ANALYSIS OF PUBLISHED LITERATURE IN MOLECULAR BIOLOGY WITH SPECIFIC REFERENCE TO HEMATOLOGICAL DISORDERS: A SCIENTOMETRIC STUDY USEFUL FOR LIBRARIANS

A

THESIS

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PUNE.

OCTOBER

2021

D E C L A R A T I O N

I hereby declare that the thesis entitled **"Analysis of published literature in molecular biology with specific reference to hematological disorders: A scientometric study useful for librarians"** completed and written by me has not previously been formed as the basis for the award of any Degree or other similar title upon me of this or any other Vidyapeeth or examining body. I understand that if my Ph.D. Thesis (or part of it) is found duplicate at any point of time my research degree will be withdrawn.

Mrs. Kala Ajit. Desai Gorakshakar
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Place: Mumbai
Date: 21 OCT 2021

C E R T I F I C A T E

This is to certify that the thesis entitled **"Analysis of published literature in molecular biology with specific reference to hematological disorders: A scientometric study useful for librarians"** which is being submitted herewith for the award of the Degree of Vidyavachaspati **"Doctor of Philosophy"** in **Library and Information Science** of Tilak Maharashtra Vidyapeeth, Pune is the result of original research work completed by **Ms. Kala Ajit Desai Gorakshakar** under my supervision and guidance. To the best of my knowledge and belief the work embodied in this thesis has not earlier formed the basis for the award of any degree or similar title of this or any university or examining body upon her.

Dr. Pratibha Gokhale
Research Guide

Place: Mumbai

Date: 21 OCT 2021

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Abstract

Introduction

Scientometrics deals with qualitative, quantitative and computational analysis of scientific literature. In modern times, advances in various branches of science are responsible for increase in the quantify of publications and research outcomes. Similarly the development of various computer programmes help to analyze this data in the context of library science, using the parameters like citation analysis. Bibliographic coupling, co-authorship network, identification of most prominent journals etc. In short, such studies are used to understand the evolution of the literature or trends of publications on a particular subject. This type of data developed in the area of library science is also useful to researhers working on that particular subject as it helps to develop the research projects to fill up the lacunae or gaps in the subject identified by the scientometric analysis.

The present scholar has been working at ICMR – National Institute of Immunohaematology (ICMR-NIIH), Mumbai for more than thirty years. This is one of the permanent and premier institutes of Indian Council of Medical Research (ICMR) where work on several hematological /genetic (hereditary) disorders is being carried out. This includes the diseases like Down syndrome, hemophilia A, hemophilia B, von Willebrand disease, sickle cell anemia and G6PD deficiency. Therefore these diseases are selected for the present scientometric analysis. Polymerase Chain Reaction (PCR) which was discovered in 1988, revolutionized the field of molecular biology. It was introduced in the routine laboratory in around 1990.

Significance of the study

Extensive search of various databases as shown that scientometric analysis of the literature published on various diseases like Typhoid, Dengue, Leprosy, Measles etc. has been done. Similarly, scientometric analylsis of the articles published from various institutes or published by renowned personalities has been done. However,

scientometric analysis of the articles published on various genetic disorders has not been done properly. Only two articles on this subject have been published so far.

Therefore, in the present study literature published on six genetic disorders viz. Down syndrome, Sickle cell anemia, G6PD deficiency, Hemophilia A, Hemophilia B, von Willebrand disease has been assessed by using various scientometric indicators. This study can help as a guideline for scholars or librarians to perform such analysis on literature published on other genetic disorders. It can also help Government agencies for funding appropriate projects. It can also help other institutes to identify lacunae in the research on various genetic disorders.

Need of the study

Information explosion in any particular area is a major challenge for scientists and researchers, policy makers, funding agencies etc., because they have to update their knowledge with recent developments in that area. Information is continuously pouring in and it is very difficult to understand the pattern of growth of the subject without analyzing this information systematically. Scientometric analysis of such data is the correct answer to this problem.

So the present scientometric study has been planned to understand the impact of molecular biology on publications of above mentioned genetic disorders in our country as well as globally.

Statement of the problem

In need of the above context the research topic has been titled as "Analysis of published literature in molecular biology with specific references to haematological disorders: A scientometric study useful for librarians."

Research questions

1) What is the trend of publications on six genetic disorders from India as well as from all over the world?

2) Is there any impact of molecular biology on the publications of articles during 1991-2014 from India as well as from all over the world?

3) What are the most prominent journals where maximum number of articles on these disorders have been published?

4) What is the pattern of growth rate on publications of articles from India on six genetic disorders?

5) Is there any difference in growth rate on publications of articles from all over the world as compared to that from India?

6) Who are the scientists or authors putting maximum efforts on research on these genetic disorders?

7) What is the extent of author collaborations in the articles published from India on the above mentioned genetic disorders?

Based on these research questions following objective have been designed.

Objectives of the study

1) To know the growth of knowledge in molecular biology in the area of certain genetic disorders (viz. Down syndrome, Sickle cell anemia, Hemophilia A, Hemophilia B, von Willebrand disease and G6PD deficiency).

2) To know the most productive year / years when the literature has been published on this subject.

3) To identify most prominent journals where maximum articles are published on the above mentioned genetic disorders.

4) To study the pattern of publications in different journals on six genetic disorders from India a well as from all over the world.

5) To study the year- wise publications of articles under different categories on six genetic disorders from India as well as from all over the world.

6) To assess the growth of literature on this subject, from India and compare it with the literature published from all over the world

7) To study the collaborative pattern among the authors who published the article on these genetic disorders from India.

8) To identify top ten authors from India on the basis of fractionalized published articles on each of the six genetic disorders.

9) To identify top ten articles published on six genetic disorders from India and receiving maximum citations.

Hypothesis

"There may be impact of molecular biology on publications of six genetic disorders (viz. Down syndrome, G6PD deficiency, Hemophilia A, Hemophilia B, Sickle cell anemia, von Willebrand Disease) published during 1991 to 2014".

Scope of the study

This study included comprehensive scientometric analysis of the articles published on six genetic disorders viz. Down syndrome, sickle cell anemia, G6PD deficiency, hemophilia A, hemophilia B and von Willebrand disease. These disorders were selected because lot of research work is being done on these disorders at ICMR-National Institute of Immunohematology (ICMR-NIIH) Mumbai where the present scholar is working. It was also thought that results of this study would help the scientists at ICMR-NIIH to design the projects to fill up the gaps in various areas. Similarly extensive scientometric analysis of the articles published on these disorders from India has not been done so far. The data was retrieved from PUBMED database for basic analysis and was retrieved from SCOPUS database for network analysis.

Limitations

In any research work limitations arise due to some constraints. This may affect the findings or conclusions. In the present study the main constraint was getting the literature on the above mentioned six genetic disorders. After extensive search only two articles were found where scientometric analysis of literature on haemophilia (combined A and B), Sickle cell anemia and von Willebrand Disease published during ten years period has been done. So sufficient data was not available for comparison with the findings of the present study.

The second limitation was development of network for author collaboration for authors from all over the world. Due to technical problem of the software it was not possible. Therefore, author collaboration was studied for authors from India only.

Review of literature

When the litetrature on scientometric analysis was reviewed extremely from various databases; it was found that only two articles were published where scientometric analysis of the literature on genetic disorders has been done. Apart

from this scientometric analysis of the literature published on various aspects has been compiled in this chapter. Those aspects are as follows;

1. Articles published in a particular journal over a particular time period.
2. Articles published by renowned personalities.
3. Articles published academic institutes.
4. Articles published on particular subject during a specific time period.
5. Articles published under medical sciences.
6. Research on other related areas like Anemia, Life science etc.
7. Biochemistry, genetics, molecular research.

Research methodology

Research means detailed study of a particular problem using scientific methods. The main objective of research is to explore the unknown and during the process unlock new possibilities.

Methodology

The present study has involved two types of research.

1) Descriptive research
2) Exploratory research

The descriptive research explains the background of the subject. This includes brief description about various metrics.

Exploratory research focuses on the analysis of the data on six genetic disorders using scientometric indicators. These two approaches gave the scholar opportunity to investigate the research problem and find out results to the research questions.

Survey Method

In the present study preliminary data was collected through survey method. The data for the present study was collected by surveying and screening the various databases viz. PUBMED, INDMED, SCOPUS, SCIENCE DIRECT. Finally PUBMED was found suitable for this study.

Data analysis

Articles published from India as well as from all over the world were analyzed using following indicators.

1) Year-wise trend of publications
2) Journal-wise trend of publications
3) Category-wise separation and analysis of the articles
4) Calculation of growth rate of the publications published during 1991-2014.

Network analysis

Network analysis was used for quantitative analysis of the data. Bibliometrix package recently developed by Aria and Cuccurullo (2017) was used for this purpose SCOPUS database was accessed to retrieve the articles published on six genetic disorders during 1991-2014.

The following indicators which were directly obtained after analysis of the data for each set of genetic disorders are included in this report.

1) Coupling or collaboration of authors in the form of network.
2) Top 5 – 8 journals revealing trends of articles published on the particular disorder from India during 1991-2014.
3) Average article citations per year.
4) Average total citations per year.
5) Most productive top ten authors calculated on the basis of fractionalized articles.
6) Highly cited top ten articles .

Hypothesis testing

"There may be impact of molecular biology on publications of six genetic disorders (viz. Down syndrome, G6PD deficiency, Hemophilia A, Hemophilia B, Sickle cell anemia, von Willebrand Disease) published during 1991 to 2014".

Articles published on six genetic disorders during 1991 – 2014 from India as well as all over the world were separated into two groups; articles published during 1991- 2002 and articles published during 2003 – 2014 to see growth rate of publications on these disorders. Articles from both the groups were further separated into six different categories (viz. Original articles, Articles based on clinical findings, Case reports, Reviews, Miscellaneous articles and Articles published using molecular biology tools). Data from category 6 (Articles published using molecular biology tools) was considered to test the hypothesis.

It was observed that 18.23- 42.4% of the articles published on six genetic disorders during 1991-2014 from India as well as from all over the world were under category six. Chi square test of contingency was applied to see the growth rate and also to test the hypothesis. It was non-significant for all the disorders in India. When articles published from all over the world under category 6 were compared for two

time intervals (1991-2002 and 2003-2014), articles published on Down syndrome and Hemophilia B showed statistically significant values. It shows that number of articles published on these two disorders under category 6 has increased significantly during second time interval (2003-2014).

Above results show that growth in number of articles published on six genetic disorders from India is steady during 1991-2014. However in the case of all over the world, articles on Down syndrome and Hemophilia B showed statistically significant increase in numbers.

It proves that there is an impact of molecular biology on publications on six genetic disorders published during 1991-2014. Thus the hypothesis stated earlier was proved.

Findings

1)The present study evaluated the research output of six genetic disorders viz. Sickle cell anemia, G6PD deficiency, Down syndrome, Hemophilia A, Hemophilia B and von Willebrand disease This study has helped us to understand the impact of molecular biology on the publications on these disorders from India as well as from all over the world.

2) This is a study where the research output on six genetic disorders published during last 60-70 years has been analyzed. This type of study has been attempted for the first time in India in the subject of Library and Information Science.

3) A Scientometric study of research output on Down syndrome and G6PD deficiency was not done earlier. In the present study it is done for the first time. Similarly analysis of the publications on Hemophilia A and Hemophilia B has been attempted separately.

4) This study presents journal-wise , year-wise and category-wise analysis of the articles. Similarly it shows authorship pattern, most productive authors, publication pattern and highly cited articles of six genetic disorders.

5) Totally 1,175 articles from India and 60,826 articles from all over the world on six genetic disorders were evaluated. This accounts for 1.90% of India's share in the global publications on these disorders.

6) 276 articles were published up to 1990 from India on the above mentioned six genetic disorders as compared to 24,710 articles published from all over the world.

7) During 1991-2014, 899 articles were published from India on six genetic disorders as compared to 36,116 articles published from all over the world.

8) Category-wise analysis of the articles published on the six genetic disorders has been done. The articles are separated into following six categories viz. original / research articles, articles based on clinical findings of the patient, case reports, reviews on various aspects of the particular disorder, miscellaneous articles describing non-scientific aspects of the particular disorder and finally articles published using various molecular biology techniques. This has helped us to get clear idea about publication pattern on these disorders over the years. This type of data is useful to the scientists working on these disorders and library professionals who can provide such data to research scholars.

Conclusion

1) From the analysis of data, it can be concluded that India's share in the global publications on individual disorders varies between 1.01% to 3.39%.
2) Also, from the analysis we can conclude that the articles published during 2015-2020 on these disorders showed that the Indian share in the global publications is increased and varies between 1.92- 8.65%. Again the maximum share of 8.65% was seen in the case of G6PD deficiency.
3) It is further concluded that the range of articles published on Down syndrome, G6PD deficiency and sickle cell anemia during 1991-2014 using molecular biology based tools varies between 18.23 - 21.81%.
4) In the case of hemophilia A, hemophilia B and von Willebrand disease which are all coagulation related disorders i.e. related to blood clotting, it varies between 28.57 - 42.40%.
5) The articles published from all over the world did not show such difference in the case of six genetic disorders. It varies between 18.23% - 26.91%
6) Data retrieved from SCOPUS shows that only 10% of the articles on these disorders are single authored, while 90% of the articles are multi authored, indicating that they are collaborative work.
7) This analysis shows that in a vast country like India only 3-4 main research institutes like ICMR National Institute of Immunohematology (NIIH), Mumbai, All India Institute of Medical Sciences (AIIMS), New Delhi, Christian Medical College (CMC), Vellore etc. are working on these disorders.

8) The information on top five journals publishing maximum number of articles on above mentioned six genetic disorders are helpful to the scientists, research scholars who are working in the research institutions.
9) Also we can conclude that the output of this work will be useful to the librarians who are working in the institutes where the research work on these disorders is being carried out. They can provide this information to their scientists who can design their future projects accordingly.

Suggessions

Following are the suggessions based on the research findings.

1. Network analysis shows that authors collaboration is not satisfactory in India. There is scope to improve this scenario.
2. More and more scientists from different institutes should collaborate with each other to conduct the research work on these disorders.
3. This information will be useful to policy makers and various research institutes to design future projects in the given area.
4. This information is also useful for funding agencies, policy makers who can precisely fund the projects which will be useful to fill up the gaps in the research on these disorders.
5) This type of study will help the librarians to subscribe the prominent journals, which will give access to the scholars to understand the current status of these disorders and to design the projects in such a way to fill up the lacunae.

Further research

Considering the findings and suggestions, researcher has noted some points which can be considered for further research for future scholars who would like to undertake the similar topic in other genetic disorders.

1. Studies can be conducted on other genetic disorders like Thalassemia, Cystic Fibrosis, Duchene Muscular Dystrophy, Huntington Disease etc.seen in India.
2. Such type of Scientometric analysis can be used to see the growth rate of publications.
3. It can also useful to know the trend of publications and to identify the gaps to understand the disease process.

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Sr.No.	Abbreviations	Full form
1	ACPP	Average Citations Per Paper
2	CSIR	Council of Scientific & Industrial Research
3	DBT	Department of Bio Technology
4	DS	Down syndrome
5	DNA	Deoxyribo Nucleic Acid
6	HepA	Hemophilia A
7	HepB	Hemophilia B
8	G6PD	Glucose 6 Phosphate Dehydrogenase
9	HIV	Human Immuno deficiency Virus
10	ICAR	Indian Council of Agricultural Research
11	ICMR	Indian Council of Medical Research
12	IQ	Intelligence Quotient
13	NGO	Non Government Organization
14	NIIH	National Institute of Immunohematology
15	NLM	National Library of Medicine
16	PCR	Polymerase Chain Reaction
17	RBC	Red Blood Cells
18	RFLP	Restriction Fragment Length Polymorphism
19	RNA	Ribo Nucleic Acid
20	SCA	Sickle Cell Anemia
21	SCI-E	Science Citation Index-Expanded
22	VNTR	Variable Number of Tandem Repeats
23	vWD	von Willebrand Disease
24	vWF	von Willebrand Factor
25	WoS	Web of Science

Chapter I

Introduction

1.1) Introduction

Research means to find out the answer in a scientific way to solve the particular problem. Therefore research is an intellectual activity which is used to solve problems in various disciplines of knowledge. It can be carried out in one of the following ways.

1) Collection of data through interview, discussion or questionnaire followed by its analysis to find some meaningful answer.

2) Generation of data by performing experiments in a laboratory followed by analysis.

3) First some hypothesis is designed and then the data is collected to say whether the hypothesis is correct or wrong?

4) Download the data from various databases or search engines on a particular aspect and then analyse it.

In the field of Library and Information Science (LIS) also research has been conducted by using one of the above mentioned ways over the years. Once the data is generated it is analyzed by using several formulas. These formulas or overall quantitative tools have been developed by many scholars to assess or to evaluate the progress in particular field of library science. Using these tools which are generally called as 'metrics', published literature in the form of books, periodicals, articles etc. published during particular time period on specific scientific area can be carefully analyzed to understand the pattern of growth. These metrics are broadly classified into three categories viz. classical metrics, neo-classical metrics and modern metrics. Bibliometrics, Scientometrics, Informetrics and Librametrics are considered as 'classical metrics'.Cybermetrics is considered as 'neo-classical metrics' while Wikimetrics, Journalmetrics , Authormetrics and Altmetrics are considered as 'modern metrics'.

1.1.1) Development of quantitative tools or metrics in Library and Information Science.

The term 'metrics' is originated from two languages. The word 'metricus' in Latin language and 'metrique' in French language means 'a measure for something'. The word 'metrics' was first introduced in 1864.Since then it has been used in various areas including Library and Information Science as a unit of measurement. When

something in large scale needs to be measured or calculated, we need statistics. It is used in several development or forecasting studies. When particular subject is studied in depth using statistics, a new area is developed. For example, application of statistics to study economics in depth gives rise to ecometrics and so on. Therefore earlier work in library science which involved bibliographical analysis using quantitative techniques of statistics was called as 'statistical bibliography'.

Campbell (1896) was the first person who performed bibliometric study using statistical techniques. However Lawani (1981) stated that the study conducted by Cole and Eales in 1917 should be considered as the first biliometric study. They studied the growth of literature published on 'comparative anatomy' during more than 300 years (1550-1860) by plotting various graphs. They called it as 'statistical analyses'. In 1923, E.W. Hulme, another gentleman, counted various documents published on the 'history of science and technology'. He called it as 'statistical bibliography'. He defined the term 'bibliography' as the science of organization of knowledge. Shapiro (1992) writes that citation indexes have been used since 1743 while Weinberg (1997) in his article states that Hebrew citation indexes were in use since 12^{th} century.

In 20^{th} century, various scholars in the area of Library Science were trying to develop the methods to assess the publications of various authors published in different journals. They were also trying to study relationships amongst academic journal citations. Meantime three research workers from the field of library science put forward different laws to study the various aspects of literature. In 1926 Alfred Lotka put forward the 'Law of scientific productivity' of an author. To study this, number of articles published by a particular author in any particular area is evaluated. Samuel Bradford, another scholar in 1934 tried to study the articles published in different journals. He arranged the journals on a particular subject based on a number of articles. This is known as 'Law of Scatter' which actually states that 'if the journal containing articles on a particular subject are arranged in descending order of the number of articles they called on that subject, then successive number of periodicals containing the same number of articles on that subject form the simple geometric series $1:n:n^2:n^3$..... However, this law is not statistically accurate. In 1947 George Zipf another gentleman working on Library Science studied certain words in an article. He put forward 'Zipf equation' which was used to study the occurrence of certain words in a text based on frequency of their occurrence.

In 1948, Dr. Ranganathan, a renowned librarian from our country, also known as a 'father of library science in India' suggested that there is a need to develop certain programs, mathematical models or statistical tools to study library systems and library services. He gave a name as 'Librametry' to this type of study. Unfortunately this idea was not properly developed till 1970.

In 1969 Alan Pritchard used the term 'bibliometrics' to replace the term 'statistical bibliography'which was confusing. He defined this new term as 'the application of mathematical and statistical tools to books and other media of communication'. However, Wilson (1995) points out that this term was originated from French language. He shows that a gentleman,Paul Otlet (1943) wrote a book on Library Science in French language. There he used the term 'bibliometrie' which has a similar meaning. However due to language problem this was overlooked.

In the same year 1969, two Russian scientists Nalimov and Mulchenko used the Russian term 'Naukometria' which is equivalent to 'scientometrics'. Now this is another metrics which involves comprehensive study of scientific literature. This includes qualitative, quantitative and computational analysis. Subsequently in 1978,Tibor Braun from Hungary started the journal 'Scientometrics'. This helped to develop this new metrics properly, as this journal started publishing various studies on scientific literature. Today lot of advances have taken place in various branches of science. Therefore number of publications on each subject has increased tremendously. However various computer based programs are now available to analyse this huge data related to library science. The various parameters developed include citation analysis, bibliographic coupling, co-authorship network etc. Thus scientometric analysis covers study of all such parameters for overall evaluation of the subject.

Practically this is done by counting how many articles are published by the scientists, how many articles are published by particular Institute or organization on particular subject or how many articles are published from particular country on a specific topic or area over the years. Such type of analysis which is called 'scientometric analysis' which helps to assess the trends in publications and subsequently overall growth of the subject. Such type of data is useful to the scholars working in that particular area to design the projects in such a way to fill up the lacunae seen in these studies. This data is also useful to government organizations, funding agencies and policy makers to design the strategy for funding of specific

projects for the overall development of the subject. From the above discussion it is clear that much of scientometrics is indistinguishable from bibliometrics.

Informetrics is the most recent classical metric. The term was used by Otto Nacke in 1979. This metrics is developed to study quantitative aspects of informal, spoken or recorded communication alongwith material from literature and other documents. It can be used for several aspects of quantitative measurement of information which lie outside the scope of bibliometrics and scientometrics.

Webometrics is another metric which was introduced by Almind and Ingwersen in 1997.This metric is used to study World Wide Web (www). Here the study of various types of hyperlinks available on net is done. This includes the study of construction of various information sources and their patterns. The main use of this metrics is ranking of universities or similar organisations after studying their webpages. This is done using parameters like Impact rank, Openness rank, Excellence rank etc.

Cybermetrics is a neo classical metric where electronic media based online communication is analyzed. After the introduction of internet in the last decade of 20^{th} century several new websites are being developed every day. Some of them are maintained properly for a long time while some of them have a very short life. Different websites on the same topic differ in quality. In Cybermetrics, techniques are developed to rank these websites depending on their content to calculate Web Impact Factor (WIF) and to study cited sites. Thus the terms Webometrics and Cybermetrics are synonymous and used in the content of measuring links, structure of usage patterns of www.

Wikimetrics is a modern metric. It is basically a webtool earlier known as UserMetrics which was developed to design the measurement of onsite user. Today we all know that Wikipedia, a modern encyclopaedia was launched in 2001. It contains detail and extensive information on several subjects and new information is being continuously included. Data analysis of these pages which are also called as wikipages is known as Wikimetrics.

Journalmetrics is another modern metric. The famous parameter this metric is called 'Impact Factor' (IF) which was introduced by Eugene Garfield in 1975. This indicates the speed with which articles get cited in various publications. The other parameters include cited half-life, the median age of articles cited, Aggregate impact factor – the impact factor for an entire subject and so on.

Authormetrics measures research impact of articles by various authors. The parameters include citations – How many times article is cited in the literature? Mentions – How many times the article is used in a blog or Wikipedia or in Facebook share, Tweets, Linked In Shares etc.

It also includes personal indicators like h-index, g-index, i-10 index etc.

Altmetrics is the most recent metric developed by Jason Priem in 2010. This is applied for quantitative measures of journals, books, data sets, presentations, videos, webpages etc. It covers information about how many times data is referred to, how many times article is viewed or downloaded or mentioned in news media or social media.

1.1.2) Introduction to the subject

The present scholar has been working at one of the prime research institutes in Mumbai for more than thirty years. In this institute work on several hematological /genetic (hereditary) disorders is being carried out. This includes the diseases like Down syndrome, hemophilia A, hemophilia B, von Willebrand disease, sickle cell anemia and G6PD deficiency. Therefore these diseases are selected for the present scientometric analysis. Polymerase Chain Reaction (PCR) which was discovered in 1988, revolutionized the field of molecular biology. It was introduced in the routine laboratory in around 1990. So scientists from all over the as well as from ICMR- NIIH and some major institutes from India started analyzing the landscape of these genetic defects in India by PCR based tools. The main idea of these studies was to unfold the uniqueness of these defects or otherwise in Indian patients and the geographical aggregation of these disorders based on population dynamics, culture, marriage, genetic admixing and way of life and from this data develop diagnostic, preventive and management strategies suitable for the country. This added a new dimension to study the above mentioned disorders at molecular level. Subsequent years have witnessed explosive growth and understanding in the area of hematological / genetic disorders at molecular level. Therefore the present scholar decided to analyze these publications from India as well as from all over the world. It was also thought that such study would tell us India's global publications share in the cumulative publication output in this subject and also would tell us the impact of molecular biology on the publications on above mentioned genetic disorders from India as well as from all over the world.

1.1.3) What is molecular biology?

Molecular biology is a separate branch of science which was established in the world in 1930s. This term was first used by Dr. Warren Weaver of Rockefeller Foundation USA who predicted that biology would undergo significant changes due to advances in technology.

Today molecular biology is not an independent branch. It has inter-relationship with biochemistry and genetics. Because biochemists study the role, function and structure of biomolecules while geneticists study DNA alternations in gene and various genetic interactions. Both of them use techniques of molecular biology for this purpose. So all the three subjects are overlapping. Molecular biology tells us biology at molecular level. In other words it is the study of life at the level of atoms and molecules.

Therefore molecular biology is a study of function, control and management of genetic information encoded in the DNA and RNA of living organisms and from this study scholars developed techniques to establish diagnostic, prognostic, therapeutic and industrial approaches to improve human lives in all its complexity including health. Structure of DNA was discovered in 1952. The discoveries of techniques like cloning, gel electrophoresis, southern blotting etc. and restriction enzymes helped the scientists to study various genetic disorders at molecular level. Similarly discovery of Polymerase Chain Reaction (PCR) in 1988 was the significant milestone and it totally revolutionized the field of molecular biology. After its discovery the scientists stated analyzing the defects causing various disorders by PCR based tools like PCR-RFLP (Restriction Fragment Length Polymorphism). Thus in subsequent years there was an extensive growth in the area of molecular biology to understand the alterations at molecular level which are responsible to cause various genetic disorders.

1.2) Significance of the study

Several genetic disorders like thalassemia, sickle cell anemia, cystic fibrosis, Duchene muscular dystrophy etc. are seen in our country. Many NGOs and other organisations have been preparing registries of these disorders. Apart from identifying and counselling such cases, lot of work is being done to understand the pathophysiology of these procedures. All this work is being published by the

researchers in several journals over the years. From the library science point of view, scientometric analysis of this literature would help us to know the evolution of literature over the years.

Extensive search of various databases has shown that scientometric analysis of the literature published on various diseases like Typhoid, Dengue, Leprosy, Measles etc. has been done. Similarly, scientometric analylsis of the articles published from various institutes or published by renowned personalities has been done. However, scientometric analysis of the articles published on various genetic disorders has not been done properly. Only two articles on this subject have been published so far.

Therefore, in the present study literature published on six genetic disorders viz. Down syndrome, Sickle cell anemia, G6PD deficiency, Hemophilia A, Hemophilia B, von Willebrand disease has been assessed by using various scientometric indicators. This study can help as a guideline for scholars or librarians to perform such analysis on literature published on other genetic disorders. It can also help Government agencies for funding appropriate projects. It can also help other institutes to identify lacunae in the research on various genetic disorders.

1.3) Need of the study

In 1983, Dr. M.S. Swaminathan, a renowned geneticist and known for his role in India's green revolution, organized an International conference on genetics in New Delhi. Since then India has made tremendous progress in the field of molecular biology. The first major step was the establishment of Department of Biotechnology (DBT) in 1986. DBT along with other government organizations like Indian Council of Scientific and Industrial Research (CSIR), Indian Council of Agricultural Research (ICAR) subsequently established several research institutes across the country. They also established co-operations with world class scientists to develop trained manpower in our country. Because of these efforts the major area which was developed was 'molecular genetics'.

Molecular biology in general is interrelated with biochemistry and genetics; and is considered as one of the sub-disciplines under biotechnology. The development of molecular biology started in our country in early 1970s with a very few laboratories and handful of scientists working on it. As mentioned earlier the discovery of PCR in 1988 followed by development of PCR based tools helped the scientists to study the various aspects in molecular biology. Therefore subsequent

years have witnessed explosive growth and understanding in the area of various disorders including genetic disorders at molecular level.

Information explosion in any particular area is a major challenge for scientists and researchers, policy makers, funding agencies etc., because they have to update their knowledge with recent developments in that area. Information is continuously pouring in and it is very difficult to understand the pattern of growth of the subject without analyzing this information systematically. Scientometric analysis of such data is the correct answer to this problem.

So the present scientometric study has been planned to understand the impact of molecular biology on publications of above mentioned genetic disorders in our country as well as globally.

1.4) Statement of problem

In need of the above context the research topic has been titled as "Analysis of published literature in molecular biology with specific references to haematological disorders: A scientometric study useful for librarians"

1.5) Research questions

1) What is the trend of publications on six genetic disorders from India as well as from all over the world?

2) Is there any impact of molecular biology on the publications of articles during 1991-2014 from India as well as from all over the world?

3) What are the most prominent journals where maximum number of articles on these disorders have been published?

4) What is the pattern of growth rate on publications of articles from India on six genetic disorders?

5) Is there any difference in growth rate on publications of articles from all over the world as compared to that from India?

6) Who are the scientists or authors putting maximum efforts on research on these genetic disorders?

7) What is the extent of author collaborations in the articles published from India on the above mentioned genetic disorders?

Based on these research questions following objective have been designed.

1.6) Objectives of the present study

i) To know the growth of knowledge in molecular biology in the area of certain genetic disorders (viz. Down syndrome, Sickle cell anemia, Hemophilia A, Hemophilia B, von Willebrand disease and G6PD deficiency).

ii) To know the most productive year / years when the literature published on this subject.

iii) To identify the most prominent journals where maximum number of articles are published on the above mentioned genetic disorders.

iv) To study the pattern of publications in different journals on six genetic disorders from India a well as from all over the world.

v) To study the year- wise publications of articles under different categories on six genetic disorders from India as well as from all over the world.

vi) To assess the growth of literature on this subject, from India and compare it with the literature published from all over the world

vii) To study the collaborative pattern among the authors who published the articles on these genetic disorders from India.

viii) To identify top ten authors from India on the basis of fractionalized published articles on each of the six genetic disorders.

ix) To identify top ten articles published on each of the six genetic disorders from India and receiving maximum citations.

1.7)Hypothesis

A research hypothesis is generally a statement of prediction that is tested by conducting the research work. The prediction is based on the relationship between the variables. In the present study variables are publications published from India as well as from all over the world on six genetic disorders. The hypothesis for this study is as follows-

"There may be impact of molecular biology on publications of six genetic disorders (viz. Down syndrome, G6PD deficiency, Hemophilia A, Hemophilia B, Sickle cell anemia, von Willebrand Disease) published during 1991 to 2014".

1.8) Research design

Research design is the set of methods and techniques used in planning of collection and analysis of the data. This helps to find answers to the research questions. Generally three main steps are involved in their process. They are data or variable collection, measurement and analysis. Data is usually collected from primary sources (i.e. database) directly and then analyzed as it has more confidence level than the data from secondary source. So after collection of the variables from proper sources different techniques including statistical tools are selected to find answers to the research questions.

In the present study the variables are publications on six genetic disorders retrieved from PUBMED up to 2014.They were published from India as well as from all over the world. After preliminary screening of the publications they were analyzed using following scientometric indicators

1) Year-wise and journal-wise analysis of the articles

2) Category-wise analysis of the articles

3) The growth rate of the articles published from India as well as from all over the world

4) Network analysis – For this purpose articles published from India were retrieved from SCOPUS database. This database was selected for this analysis because the scientometric package used for building matrices for co-citation or coupling analysis is easily compatible with the data retrieved from SCOPUS and converting into R format. This analysis was done to see the extent of collaborations among the scholars or scientists working in India.

1.9) Scope of the study / Flow charts

This clearly tells us the extent of work which will be covered during the present study in order to reach some meaningful conclusions.

This study included comprehensive scientometric analysis of the articles published on six genetic disorders viz. Down syndrome, sickle cell anemia, G6PD deficiency, hemophilia A, hemophilia B and von Willebrand disease. These disorders were selected because lot of research work is being done on these disorders at ICMR-National Institute of Immunohematology (ICMR-NIIH) Mumbai where the present scholar is working. It was also thought that results of this study would help the scientists at ICMR-NIIH to design the projects to fill up the gaps in various areas.

Similarly extensive scientometric analysis of the articles published on these disorders from India has not been done so far.

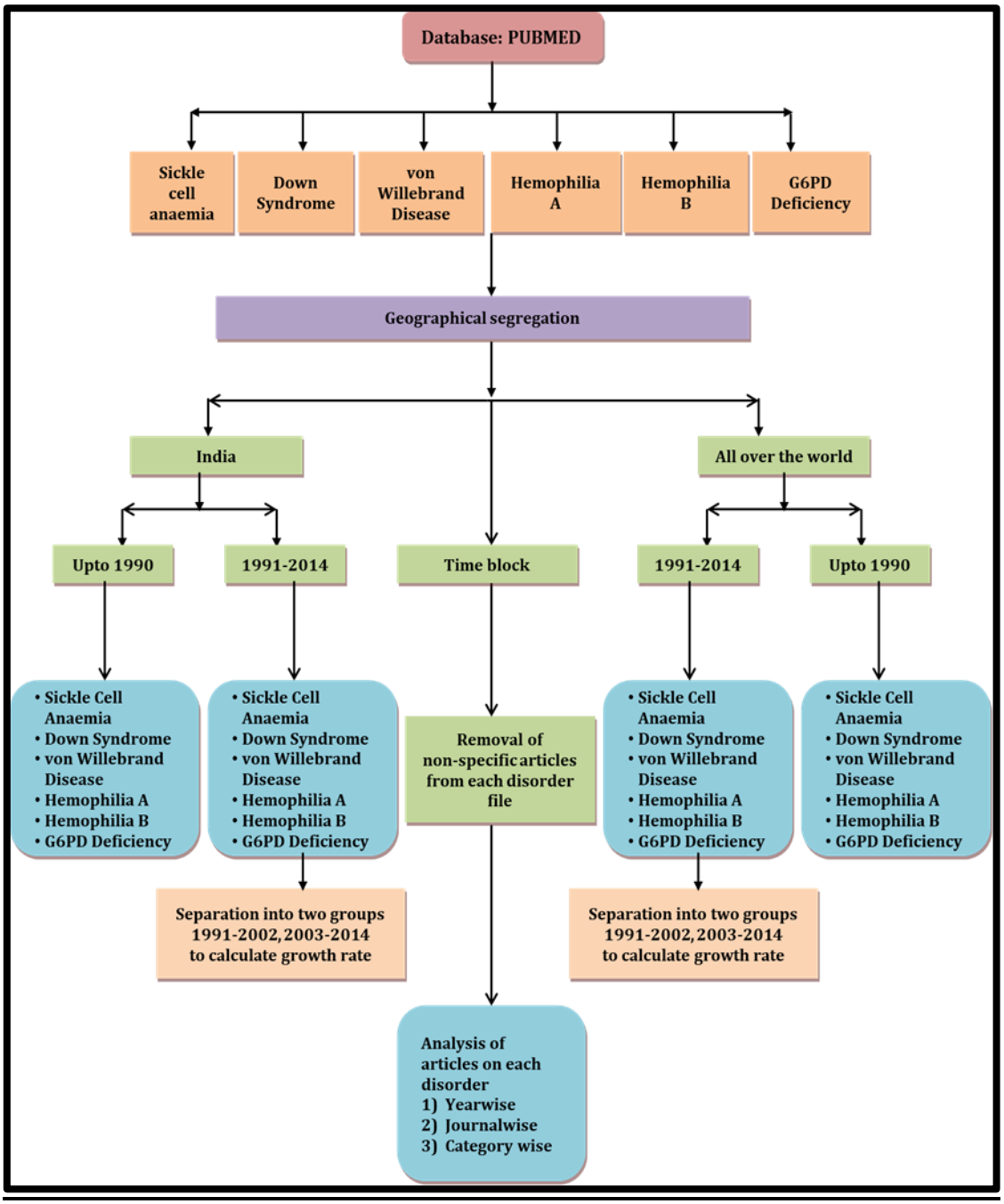
It was decided to retrieve the data from PUBMED database for the reasons mentioned under 'selection of a database'. The time period for each disorder was since the year when the first article on each disorder was listed in PUBMED and up to 2014; the span of about 60-70 years for each disorder. The span for each disorder was different depending upon the year when the first article on it was listed.

Initially the articles published from India as well as from all over the world on the six genetic disorders were downloaded from PUBMED database. Then the geographical segregation of these articles was done. i.e. the articles on each disorder were separated into two files. The first one was 'articles published from India' and the second one was 'articles published from all over the world'. Thus twelve files (two for each disorder) were created. The articles from each file were further separated into two groups viz. 'articles published up to 1990' and 'articles published during 1991-2014' to see the impact of molecular biology on publications of each disorder.

Using scientometric indicators like 'year-wise and journal-wise analysis', the basic analysis of the articles was done. Then the articles published during 1991-2014 from India as well as from all over the world were further separated into six different categories as described under 'pattern of analysis' to see the pattern of publications on different genetic disorders. The articles published during 1991-2014 were again separated into two groups viz. 'the articles published during 1991-2002' and 'the articles published during 2003-2014'. This helped to calculate the growth rate of articles published on each disorder during 24 years. Schematic presentation of all these steps is shown in flow chart 1.

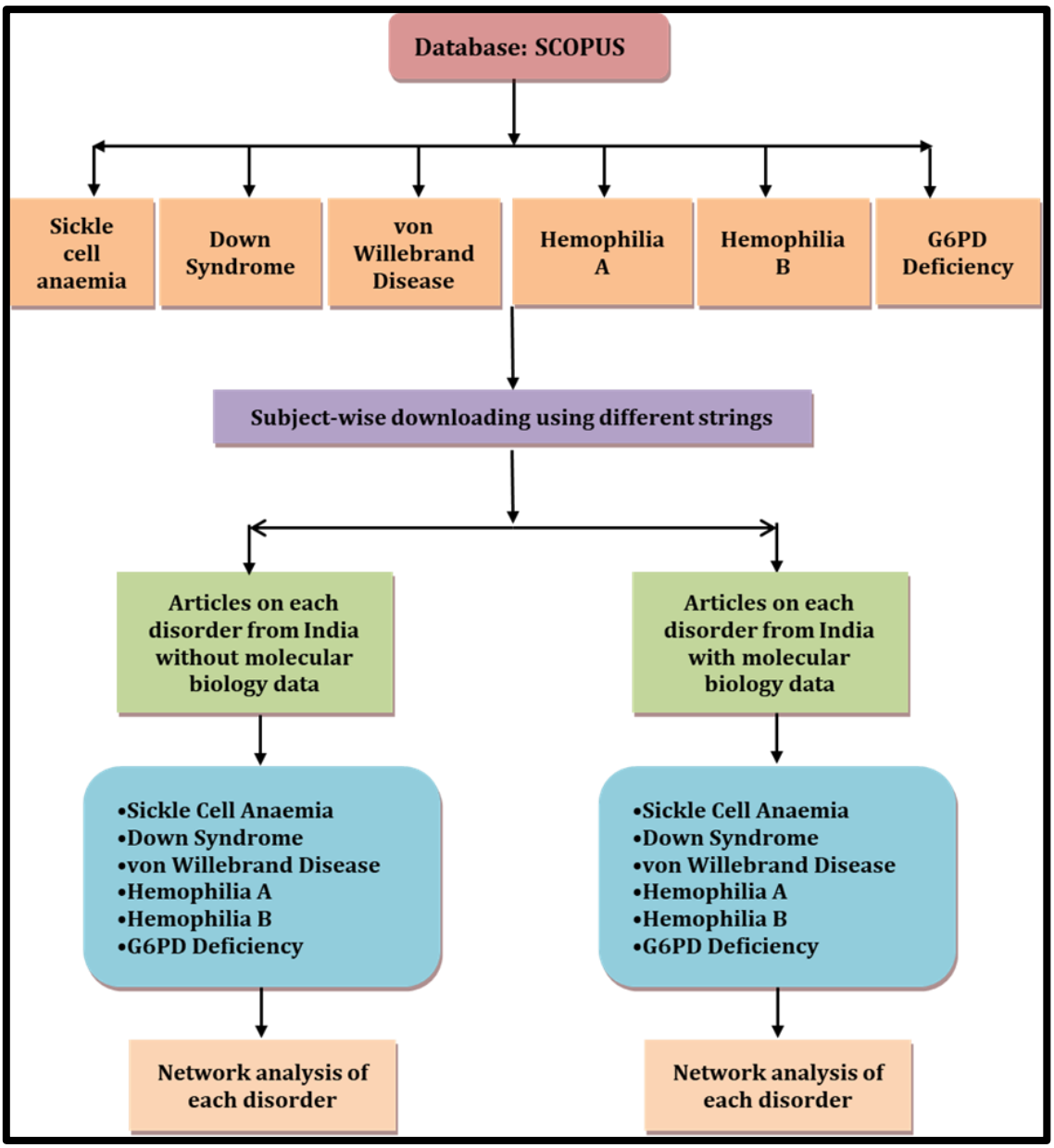
For network analysis, the articles on each disorder published from India were downloaded from SCOPUS database for the reasons mentioned above. These articles were separated into two groups. The first group consisted of 'the articles which were published without any data related to molecular biology' while the second group consisted of 'the articles involving data generated by using molecular biology tools'. Thus for each disorder two files were created. These files were subjected to network analysis using R tool format to look for author collaboration. This was recently developed by Aria and Cuccurullo (2017).

Flow Chart 1



Schematic presentation of algorithm used for analysis of the data.

Flow Chart 2



Schematic presentation of algorithm used for network analysis of authors in India.

Thus for each disorder two networks along with basic information like total number of articles ,total number of sources, number of single authored articles, number of multi-authored articles, average citations per article etc. was obtained. Schematic presentations of all these steps are shown in flow chart 2.

1.10) Limitations

In any research work limitations arise due to some constraints. This may affect the findings or conclusions. In the present study the main constraint was getting the literature on the above mentioned six genetic disorders. After extensive search only two articles were found where scientometric analysis of literature on haemophilia (combined A and B), Sickle cell anemia and von Willebrand Disease published during ten years period has been done. So sufficient data was not available for comparison with the findings of the present study.

The second limitation was development of network for author collaboration for authors from all over the world. Due to technical problem of the software it was not possible. Therefore, author collaboration was studied for authors from India only.

1.11) An overview about the genetic disorders which are selected for the present study

1.11.1 Introduction

Blood is the most essential component of our body which is made up of liquid and solid parts. The liquid portion is called plasma which is pale yellow in color. In this many cells like red blood cells, white blood cells and platelets are floating in our body. Hemoglobin is present in red blood cells. It carries oxygen from lungs to various parts of our body and brings back carbon dioxide from body parts to lungs from where it is exhausted out. White blood cells fight against various microorganisms causing infections; while certain white cells are involved in our body's defence system. A big group of various proteins is involved in blood clotting process. All these proteins are collectively called as 'Coagulation Factors' which along with platelets are involved in clot formation. If any defect occurs in production of these cells, the individual's health is greatly affected. Similarly is the levels of proteins of coagulation cascade if the level of coagulation factors is reduced then the clotting process is affected. Some disorders of blood are acquired after birth due to

medication or blood loss; while some disorders which are transmitted from parents to children are called hereditary disorders.

G6PD deficiency is a disorder where an important biochemical enzyme Glucose 6 Phosphate Dehydrogenase (G6PD) is not produced in an individual's body. Down syndrome is a genetic disorder which occurs as random events during the formation of eggs and sperms. An error in division of these cells results in the formation of cells with an abnormal number of chromosomes.

Genetic information of all the living organisms is stored in DNA and RNA present in their body. Therefore a genetic disorder is a disorder which is caused due to an alteration in DNA or RNA sequence. These alterations are called as mutations. A mutation can be present in a single gene then it is called a monogenetic disorder (e.g. Sickle cell anemia) or it can be present in multiple genes in an individual. Sometimes genetic disorders can be caused due to a combination of gene mutations, some environmental factors (e.g. smoking) and/or damage to entire chromosome. Some genetic disorders are caused by acquired mutations during person's life e.g. cancer.

1.11.2 Down syndrome: This is a condition in which a person has an extra chromosome which is formed by a random error during the formation of eggs and sperms. This chromosome comes from chromosome 21. Therefore this is also called 'Trisomy 21'. In these individuals each cell in the body has three copies of chromosome 21. Some individuals have an extra chromosome in some of the body cells. This is called "Mosaic" Down syndrome. In some individuals Down syndrome is caused because part of chromosome 21 becomes attached or translocated to another chromosome before or at the time of conception. This is called translocation. Therefore most of the times Down syndrome is not inherited, only in the case of translocation type it can be inherited. It is named after Dr. John Down, the British doctor who fully described the syndrome in 1866. This disease can be detected by chromosome analysis on a blood or skin sample. The main symptoms include distinct facial appearance, poor muscle tone, intellectual disability and developmental delay since infancy. It occurs in about 1 per 1000 babies born every year. About 23000 to 25000 babies are born in India every year with this disorder which is the highest rate in the world. The average IQ of these children is about 50. Today the average life span of these people is about 60 years in developed countries

where proper care of these children is taken. Today molecular biology based techniques are available to detect this disorder during pregnancy in our country. This is called prenatal diagnosis.

1.11.3 Sickle cell anemia: It is an inherited group of disorders that affects production of normal hemoglobin molecule. This condition was first described by American physician Dr. James Herrick in 1910. People with this disorder produce a typical hemoglobin molecule called hemoglobin S (Sickle hemoglobin). This is formed due to a mutation in the beta chain of hemoglobin, the gene for which is located on chromosome 11. Normal hemoglobin in our body is involved in transporting oxygen from lungs to other organs of the body. However, sickle hemoglobin molecules stick to one another and form long rod like structures which are responsible for making RBCs stiff, rigid and ultimately they assume sickle or crescent shape. Such RBC's pile up and causes blockages and damages various organs in the body. These RBC's also die early in their body causing anemia. Blockages are responsible for pain. Apart from it frequent episodes of infections, swelling of hands and feet, fatigue are some other symptoms. About 250 million people from all over the world carry this abnormal gene. It is mainly seen in Africa, India (mainly among tribal population) and the Arabian Peninsula.

A baby with sickle cell anemia inherits one copy of abnormal gene from each parent. When both the parents have one copy of abnormal gene, there is a 50% chance that this baby will carry one abnormal gene the condition is called as sickle cell trait, 25% chance that baby will not carry any abnormal gene and will be born as normal baby and 25% chance that their baby will carry both the abnormal genes (one from each parent) which is called Sickle cell anemia. Thus this is an autosomal recessive disorder.

1.11.4 G6PD deficiency: Glucose 6 Phosphate Dehydrogenase (G6PD) deficiency is an inborn error of metabolism. The deficiency of the enzyme G6PD causes red blood cells to breakdown prematurely which is called hemolysis. This causes anemia.

It is an X linked disorders inherited in a recessive pattern. Therefore, generally males are affected and females are carriers. Most of the time the affected males have no symptoms. However, certain specific triggers like stress due to infections, certain medications like anti material drugs like quinine, primaquine etc.,

ingestion of foods like Fava Beans may develop systems of intravascular hemolysis like dark urine, shortness of breath, jaundice, renal failure and in some cases even death.

Thus avoiding such triggers is the best option. About 400 million people all over the world are affected by this condition. It is commonly seen in Africa, Asia the Mediterranean and the Middle East.

The next three disorders viz. Hemophilia A, Hemophilia B and von Willebrand disease (vWD) are all related to blood coagulation or blood clotting process. In normal individuals platelets and fibrin are required to stop blood flow and to form a clot when blood vessel is injured several specific proteins known as 'Coagulation factors' are activated one after the other in a 'Cascade effect'. This leads to fibrin formation. Fibrin is an insoluble protein which forms a fibrous net like structure to stop the flow of blood. Similarly when the endothelium (A thin membrane that lines the inside of heart, blood vessels and lymph vessels) is damaged, underline collagen is exposed to circulating platelets which directly attach to collagen. This binding is strengthened by von Willebrand Factor (vWF). It helps platelets to stick together like glue to form a clot.

In the case of hemophilia blood does not clot properly. This can lead to spontaneous bleeding as well as bleeding following injuries or surgery. People with hemophilia have low levels of certain coagulation factors. People with von Willebrand disease either have a low level vWF in their blood or the vWF does not work the way it should. So it takes longer time to form a clot so bleeding takes longer time to stop.

People in ancient times were aware about bleeding problems. They observed that in most of the individuals clot was formed normally but in some individuals it was not formed normally. However, very little was known about clot formation. The first article on hemophilia was published in 1813 in 'New England Journal of Medicine'. Hemophilia has been called a 'Royal disease' because Queen Victoria of England had the gene for hemophilia which was occurred due to spontaneous mutation in her body. Her one son was hemophilic while two daughters were carriers for the gene. Their daughters passed on this gene to Spanish & Russian royal families after marriage. This gene was for hemophilia B. In 1947 Dr. Pavlovsky from Argentina distinguished two types of hemophilia viz. Hemophilia A and Hemophilia B.

1.11.5 Hemophilia A: Hemophilia is caused by mutation or change in one of the genes that provides instructions for making the clotting factor proteins required to form a blood clot. This disorder is inherited as an 'X' linked recessive pattern. Therefore usually males are affected while females are carriers. Father cannot pass this disease to their sons. In this disorder there is a deficiency in clotting factor VIII protein which results in increased bleeding. Individuals generally suffer due to internal and external bleeding episodes. External bleeding include prolonged bleeding during surgery or superficial bleeding due to cuts; while serious internal sites of bleeding are joints, muscles, brain etc. It's prevalence in India is about 4 per 1, 00,000 individuals or 1 out of 10,000 male births.

1.11.6 Hemophilia B: It is also an inherited coagulation disorders. It is formed due to deficiency of factor IX. Like hemophilia A it is also inherited as X linked recessive pattern. It is also known as 'Christmas Disease' named after Stephan Christmas , the first patient suffering from this disorder was detected in 1952. The common symptoms include easy bruising and bleeding. In this disorder periodontal disease and bleeding is high. This disorder is less common than Hemophilia A . It's prevalence in India is 0. 1 per 1,00,000 individuals or 1 out of 30,000 male births.

1.11.7 von Willebrand disease (vWD): This disease is names after the Finnish physician Eric Adolf von Willebrand who first detected this condition in a small girl in 1924. Subsequently he published this case in 1926 where he called it as 'pseudo hemophilia'. It is the most common hereditary blood clotting disorder seen in humans. It is inherited in an autosomal dominant fashion. It is caused due to deficiency in quality or quantity of vWF which is required for platelet attachment. There are three main categories of this disease, they are hereditary, acquired and pseudo type. It occurs among males and females equally. The symptoms include varying degrees of bleeding tendency which are in the form of nose bleeds, bleeding of gums while among females heavy menstrual periods and blood during delivery. Its prevalence is about 1% of the general population in India. It remains as an under diagnosed in India till 2005.

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Chapter II

REVIEW OF LITERATURE

2.1 Introduction

'Review of literature' is a process where the earlier findings by the scholars on the topics related to the present subject are collected and presented. Similarly the current status about the subject is also presented. This is important information as it helps to design the research questions and accordingly our research work.

Scientific development is a continuous process which is generally assessed by analyzing various research projects of scientists, academicians, researchers etc. The results of this work are published in the form of research articles in various national and international journals. Many times these journals are subject or discipline specific. This helps to increase the visibility of their work at regional, national or international level. These publications are listed in various databases.

The analysis of journals or articles on a particular subject or discipline from these databases gives us an idea about the trend of publications on these areas. This process is called 'Scientometric analysis'. If such analysis is carried out on a particular disease, then it gives us a fare idea about the growth in knowledge of that disease over the years. When this data is observed by the medical professionals or researchers working on that disease, then this would help to improve the health of the patients suffering with that disease or to improve the prognosis of critically ill patients.

Apart from this, scientometric analysis can also be done of the articles published on particular subject or articles published in different volumes of a particular journal over the years. Using this process of analysis, articles published by renowned scientists on a particular subject can also be evaluated to understand the impact of that scientist. Articles published from different Institutes can also be evaluated to assess the progress of that Institute. In the area of medical sciences, scientometric analysis has been carried out in sub speciality like genetics, biochemistry, biotechnology etc. In some cases articles published on particular disease (malaria, tuberculosis etc) over a time period of 10-15 years have been analysed. Examples of all these categories have been listed in the following paragraphs.

2.2 Articles published in a particular journal over a particular time period.

This include following examples. Scientometric analysis of 177 articles published in Annals of Library and Information Studies during 2009-2013 (Paliwal 2015), analysis of 3,174 articles published by Indian scientists (2.37% of the total number of articles) in PLOS ONE during 2012-2016 (Ahmadi 2018).

Similarly Davarpah and Aslekia (2009) conducted scientometric analysis of 894 articles published in 56 LIS journals during 2000-2004; while 316 articles published in Indian Journal of Biochemistry and Biophysics (IJBB) during 2009-2013 have been evaluated by Bala and Singh (2014).

The scientific articles which have been published during 1989-2015 and cited more than 100 times have been analysed by Kolle and Hanumaiah (2016). They retrieved the data from WoS. Marisha (2019) analysed 18,897 records published in Current Science during 1990-2017.

2.3 Articles published by renowned personalities

This category generally provide the basis for measuring the outputs of renowned individuals including scientists.

Kalaiappan et al (2010) evaluated the scientific literature published by G.N. Ramchandran a renowned physicist worked on X-ray crystallography, molecular biophysics and peptide synthesis while Kalaiappan and Yesudoss (2018) compared the scientific publication pattern of Prof. G.N. Ramchandran and Prof. C.N.R. Rao, a renowned chemist worked in solid state and structural chemistry. They found that Prof. Ramchandran showed solo article pattern as 35.5% of his articles were written by him alone while Prof. Rao showed collaborative article pattern as more than 95% his articles were multi authored thus showing good degree of collaboration.

Kulkarni (1997) analysed 147 articles published by Mr. M.V. Bhole, a renowned personality in the field of yoga. These articles were published during a span of 30 years (1965-1995). He observed that 60% of the articles were based on collaborative work.

Kannappanavar et al (2004) analysed the overall publishing trends of Indian chemical scientists who published their articles during 1996-2000. The average number of authors per paper increased from 7.52 to 8.39 during this period. This is a healthy trend indicating increase in collaborative pattern during this time period. Tilak

et al (2003) published the analysis of contributions of Indian foresters (some different field) and found the degree of collaboration was 0.64 among the authors.

2.4 Articles published by academic institutes

To evaluate the growth of a particular Institute, scientometric analysis of the research output of that Institute is done.

Jeevan and Gupta (2002) evaluated the publications of the scientists from Indian Institute of Technology, Khargpur. 1,172 articles published from this Institute during 1994-1997 were evaluated. The data was extracted from annual reports of the Institute and Science Citation Index (SCI). 47.6% of the articles from Department of Chemistry received normalised impact above 2.0. Chemistry department, Physics and Meteorology department and Electronics and Electrical Communication Engineering department were found to be the top three departments as far as publications were concerned.

901 articles were published by the scientists from Indian Institute of Technology Roorkee during 1993-2001 have been analysed by the Singh et al (2005). The articles were extracted from Science Citation Index (SCI). Maximum number of articles (280) were published by Engineering and Technology Unit. Out of 901 articles, 23(8.22%) articles were involved in international collaboration while 32(11.43%) articles were involved in national collaboration. However, Department of Mathematics, Department of Biology and Department of Clinical Medicine even though contributed less articles as compared to other departments during above mentioned time period, got good average normalized impact factor per paper.

1,518 articles published in 405 journals during 1996-2006 by the scientists from different departments of Science and Technology of University of Mysore was extracted from SCOPUS database (Kumbar et al2008). Annual growth rate was found to be 23.9%. Average Citation per paper increased from 1.53 in 1996 to 2.62 in 2003. 14% of the total publications were through international collaboration.

Bala and Gupta (2009) analysed 754 articles published during 1992-2007 by the scientists from Government Medical College and Hospital (GMCH), Chandigarh. The data was extracted from SCOPUS database. This data has been compared with the data published during the same period from top 15 medical colleges from India. Amongst these GMCH ranked 9^{th} in the research output while ranked 13^{th} in the average citation per paper. Thus authors concluded that substantial improvement is

required in research environment. Good infrastructure facilities, recruitment of qualified staff, increase in International collaboration are some other aspects which need to be looked into.

2.5 Articles published on a particular subject during a specific time period.

Some studies have been conducted on research articles published on a particular subject during a specific time period to understand the growth of that subject during that period.

Nattar (2009) analysed 829 articles published during 2004-2008 on various aspects of physics in volume numbers 78-82 of Indian Journal of Physics to understand the growth of the subject. He found that 12.42% of articles were single authored while 33.17% of the articles were multi-authored. The degree of collaboration seen in this journal during the above mentioned period was 0.781.

Pathak and Prasanna Kumari (2019) analysed 14,474 articles published during 2014-2018 in the area of Pharmaceutical Research. They were extracted from web of Science. 3,568 articles (24.65%) published by Indian scientists were with International collaboration. Overall research in this field in our country is on the rise. There were 617 publications in 2014 in this field from India which increased to 809 publications in 2018. India ranked 4th in global research output in this field.

Mohan and Kumbar (2020) analysed 1,569 research articles published during 1999-2018 on Solar physics. The data was extracted from Web of Science (WoS) core collection database using Clarivate Analytics. The data was analysed using R program. VOS viewer software was used for the visualized analysis of the articles. This included bibliometric networks for journals, authors, co-authors, citations, bibliometric coupling etc. The research on this field was increased with the annual growth rate of 8.9% with a high degree of collaborative work. India collaborated with 787 Institutes from all over the world. This articles received 21.792 citations with average number of citations per document is 13.89.

Satish Kumar (2020) assessed 20,311 articles published on Astronomy and Astrophysics research in India. The data was retrieved from Web of Science (WoS) published during 1988-2017 and analysed using various scientometric parameters like literature growth, document types etc. The research work progress in this area was very good during the above mentioned period except during 2008-2014 when it was showing steady growth. India was on 13th position in this field in the world.

Interestingly 30.51% of the articles from India have been published in open access journals. Tata Institute of Fundamental Research (TIFR), Mumbai is the highly productive organization in this area. Indian scientists working on Astrophysics are more interested in collaborative research than independent research.

Jeyasekar and Sarvanan (2014) evaluated the literature published on Forensic Science from all over the world during 1975-2011. Totally 13,626 articles were extracted from SCOPUS database. India ranked 8th with 414 articles published on this subject during the above mentioned time period. The authors observed that the literature has grown exponentially during 36 years. Journal of Forensic Sciences is the top ranking journal which published 33% of the articles on this subject during the above mentioned time period. Apart from the topic specific journals, the literature on this subject has been scattered in many journals.

2.6 Articles published under medical sciences

This branch of science covers various areas from cancer to biotechnology and neurological diseases to genetics, heredity, life sciences etc. Various scholars have evaluated the growth of these areas by analysing the research output published under different subjects. These publications have been systematically listed and their salient features are discussed in the following paragraphs.

2.6.1 Articles published on neurological diseases

Neurological diseases generally affect the brain, the nerves and spinal cord of our body. Alzheimer's disease, Parkinson's disease and multiple sclerosis are the most common neurological diseases.

2.6.1.1 Neuroscience

Bala and Gupta (2010) analyzed 4,51,331 articles published from all over the world during 1999-2008 on neuroscience which have been downloaded from SCOPUS database. India ranked 21st in the 26 top productive countries with 4,503 publications on this subject during this period. India's global publication share was 0.99%. Cumulative publication output of India witnessed growth of 59.24%. India's annual average publication growth rate in neuroscience was 11.37%. The average citation per paper published from India was 4.21. India published 781 papers with

international collaboration during above mentioned period which accounted for 17.34%.

Shahbuddin (2013) also evaluated the articles published on neuroscience from India during 1992-2005. Although the time period was overlapping with earlier publication, in this article the data was retrieved from Neuroscience Citation India (NSCI) and PUBMED databases. Therefore different set of articles were available for analysis. Totally 18,138 articles were published from India on this subject during 14 years period. The author observed that multi authored articles which indicate collaborative work on this topic are well cited. Similarly it was found that Indian authors have international collaborations in only 12% of the articles indexed in NSCI. Unfortunately more than 70% of the articles on this subject have not been cited at all. Therefore as suggested by Gargouri et al (2010), the present author also suggests that scientists working on neuroscience in India should publish in 'open access' journals. This will help to improve the visibility and hence increasing the possibility of citing this work by other scientists.

2.6.1.2 Dementia

Dementia is not a particular disorder but a common term used to describe impaired ability to remember, to think or to make certain decisions.

Gupta et al (2011) retrieved 89,129 papers from SCOPUS database published during 2002-2011 on dementia from all over the world and observed that India published 1,109 articles during this period and ranked 16th with its share of 1.24%. The authors suggested that there is a need to have good funding programs to conduct research in this area.

2.6.1.3 Alzheimer's disesase

It is a progressive neurological disorder where brain is mainly affected. It shrinks and brain cells die. These irreversible changes in brain are responsible for loss of memory and thinking skills. This is the most common type of dementia.

Gupta and Bala (2013) also evaluated research output of another important neurological disorder, Alzheimer's disease from India during 2002-2011 using SCOPUS database. Only 900 articles were published on this disease from India during the ten years' time period. India ranked 16th among the top 20 most productive countries. Its global publication share was only 1.33%. In addition,

citation impact per article and international collaborative publications share decreased considerably during this period. To improve this situation, authors concluded that there should be increased funding for research on this disorder. Serious efforts should be made to improve national and international collaborations.

2.6.1.4 Parkinson's disease

It is a progressive nervous system disorder which affects the overall movements of the patient. It first starts with tremors in hands and subsequently causes stiffness or slowing of movements. Sometimes speech is also affected.

Gupta and Bala (2013) extracted 31,132 articles published on Parkinson's disease from all over the world from SCOPUS database. They were published during 2002 to 2011. India ranked 16th among the top 20 countries from all over the world with the total publications of 458 articles. Its global publication share was 1.47% and an annual average publication growth rate was 26.05% during the above mentioned period. Overall the prevalence of this rare neurological disorder is very low in India. Exception for this is Parsis which is a very small community where the highest prevalence of this disease in the world is reported (Tags 2012). 328 out of 1,00,000 Parsis are affected with this disease while in the remaining population of India 70 out of 1,00,000 people are affected. In spite of very low prevalence of this disease in India, the research performance is very good in global context.

2.6.1.5 Autism

Autism is a broad term used to describe various neuro-developmental disorders. People suffering with it have problems with communication and social interactions. Generally males are more affected than females with the ratio of 4:1.

Jeyshankar and Vellaichamy (2016) harvested 13,079 articles from SCOPUS database published on Autism during 2007-2011. Indian scientists published 134 articles on Autism during this period. India ranked 17th among the top productive countries with a global publication share of 1.01% during this period. Out of total articles published from India, 64.76% of the articles were 'original articles' published in various journals while 16.06% of the articles were 'review articles'. 79.61% of the articles were multi authored showing good degree of collaboration in

this area. The degree of collaboration was 0.77. Journal of Autism and Developmental Disorders was the core journal which published 7.19% of the total articles.

2.6.2 Mycobacterium genus

In microbiological classification of bacteria 'mycobacterium' is a genus which is under phylum Actino bacteria and family Mycobacteriaceae. It includes pathogens which can cause serious diseases in humans like tuberculosis and leprosy.

2.6.2.1 Tuberculosis

Tuberculosis is a disease which is caused by an organism called Mycobacterium tuberculosis. They generally attack lungs but other parts of the body are also affected. This disease is spread through air when a person with tuberculosis coughs or sneezes. It is a major health problem in India as around 2,20,000 deaths occur every year due to this disease.

The three articles where research output on tuberculosis is evaluated are listed below. The three authors selected different time intervals to extract the data and also selected different databases to extract the data. Therefore their results show variation.

Gupta and Bala (2011) downloaded 77,713 articles published during 1998-2009 from 21 countries on tuberculosis research. The articles were downloaded from SCOPUS database. India ranked 3rd with 6,004 articles published during the above mentioned period. Global share of research output was 7.73%. The cumulative publication output recorded a growth of 90% when two time intervals viz. 1998-2003 and 2004-2009 were compared; while annual average publication growth rate during 1998-2009 was 10.16%. International collaboration was involved in 722 articles out of 6,004 articles, which accounted for 12.03% of the total publications from India. The average citation per paper registered by these articles was 3.73.

Maharana (2015) evaluated 48,444 documents published on tuberculosis during 2004-2013 from all over the world. They were retrieved the documents from World of Science database. Of these, 5,073 documents were published from India accounting for 10.47% of the global output. Considering the number of deaths of due to tuberculosis, India ranked 58th among tuberculosis affected countries in the world and 13th among tuberculosis affected countries in Asia. India collaborated with researchers from 20 different countries and contributed 1,463 articles through their

collaboration. This accounted for 28.84% of the total articles published from India during above mentioned period.

Ravi and Kumar (2015) analysed 1,310 articles published on tuberculosis from India during 1997-2006. The data was retrieved from three databases viz. PUBMED, SCI, and BBCI. The study included identification of Institutes where research on tuberculosis was being conducted, identification of high impact journals where research work on tuberculosis was published and international collaboration pattern.

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2.6.2.2 Leprosy

Leprosy is an infectious disease caused by bacteria called Mycobacterium leprae. It is also known as Hansen's Disease (HD). The infection damages nerves, skin, eyes or respiratory tract. This chronic disease can be cured within 6-12 months with multi drug therapy. Today the National Leprosy Eradication Programme is being run in our country. In 551 districts of the country, the prevalence of leprosy was less than 1 per 10,000.

Rajeshwari et al (2014) analysed 3,583 articles published on leprosy from India during 1960-2012. The articles were retrieved from PUBMED database. They observed that in 1960 only 4 articles were published while in 2012, 138 articles were published on leprosy. The growth rate fall from 0.81 in 1960 to 0.04 in 2012 due to elimination of this disease from the country.

2.6.2.3 Mycobacterium genus

As mentioned earlier, Mycobacterium is a genus which includes bacteria which cause tuberculosis and leprosy.

Rahul and Nishy (2016) extracted data on this genus. The data was retrieved from Web of Science database published during 1987-2012. Totally 79,628 records were obtained which were published from all over the world. India contributed 6,470 articles which accounted for 8.12% of the global research output occupying the 3rd position, however ranked 12th when quality and quantity of the research output was considered together. International collaboration accounted for 17% of the total Indian publications.

2.6.3 Cancer related topics

Cancer is an umbrella like term which includes a group of disorders which involve abnormal and uncontrolled cell growth. Therefore it can spread to other parts of the body. On the other hand there are tumors which are benign i.e. they are restricted to certain parts of the body. More than 100 types of cancers have been identified so far. Depending on which part or organ of the body is affected, they are called accordingly. For example, skin cancer, breast cancer, blood cancer (Leukemia), prostrate cancer etc.

2.6.3.1 Cancer research

Patra and Bhattacharya (2005) extracted 6,484 records published during 1984-2004 on cancer research. India's share was 648 articles with 0.42% in the global context. 6,484 articles published from all over the world were published in 868 journals. There were 19 authors who published more than 50 articles during this period.

2.6.3.2 Oncology research

Oncology is a branch of medicine which deals with the study of cancer. It mainly involves three areas viz. medical, surgical and radiation treatment. A doctor who is treating cancer patients is generally called oncologist.

Muthukrishnan and Senthilkumar (2017) analysed 10,807 records published on oncology research in India. The data was retrieved from Web of Science database published during 1989-2015. The data was analysed using Histcite and Citespace software. Various parameters like institutes, authors, years etc. were evaluated. G.K. Rath contributed maximum articles (227) during this period.

2.6.3.3 Leukemia research

Leukemia is a cancer of blood cells. In this disease, excess white cells are formed in an individual's body and they do not work in a proper way. Its symptoms include night sweat, weight loss, headache, vomiting, swollen lymph nodes etc. This is a rare disease.

Jeyshankar and Rameshbabu (2013) assessed 2,120 articles published during 1960-2011 on leukemia research by Indian scientists. The data was retrieved from

SCOPUS database. Authorship pattern, growth, co-authorship index, institutions' contribution were

some of the parameters which were analysed. The average growth rate of the research output was 1.12% . Three authored articles were maximum with 23.30% of the total output.

2.6.3.4 Bone marrow research

Bone marrow is a spongy tissue present in some of the bones in our body. These include thigh and hip bones. Cells present in this tissue are called stem cells. This soft tissue produces 200 billion red blood cells, white blood cells and platelets every day. Patients suffering from blood cancer (Leukemia) or lymphoma, thalassemia, sickle cell anemia require bone marrow transplant to survive.

Gupta and Bala (2013) retrieved 1,11,610 articles published on bone marrow research from all over the world. The articles were published during 2003-2012 and downloaded from SCOPUS database. 2,613 articles were published by Indian scientists on this subject during above mentioned period. The average citation impact per paper was 2.04 while international collaborative share of India was 11.56% . The major focus of Indian publications was in the area of medicine as 76.58% of articles by Indian scientists were published on this subject.

2.6.3.5 Stem cell research

Stem cells are special cells which are present in bone marrow of our body. They can develop into various cells of our body. They are also involved in repair system. They can change into specialized cells like muscle cells, blood cells, brain cells etc. They are used as a therapy to regenerate damaged tissue. Embryonic stem cells are there in embryos which are formed at very early stage of development while adult stem cells can differentiate into varieties of a particular type of cell which is determined by the type of tissue in which they are seen.

Karapagam et al (2012) evaluated the growth of stem cell research in India and compared it with the research output from all over the world on this topic by extracting 3,05,885 articles published during 2001-2010. These articles were extracted from Web of Knowledge database. India ranked 13th among the top 20 countries with 5,610 publications during ten year period. India's global publication share was 1.83%. On an average India published 56 articles per year. India's output increased from 1,733 papers during 2001-2005 to 3,877 papers during 2006-2010

time block with a growth rate of 12.37%. India's 5,610 papers received 37,511 total citations during the above mentioned 10 years with an average citation per paper was 6.69. Considering the world research output on stem cells, the journal 'Blood' published 9,794 articles during 10 years which received 1,50,500 total citations.

The authors observed that in a vast country like India, research on stem cells is restricted to very few institutes due to various reasons. Similarly the quality of research what is being carried out is very low which is evident from low share of highly cited articles. So the authors suggested that there is a strong need to develop proper plan to have goal oriented and need based progress which will enable the scientists to publish good articles on this subject.

2.6.3.6 Lymphoma research

It is one type of cancer where the cells of the immune system called lymphocytes are affected. These cells are present in lymph nodes, spleen thymus, bone marrow etc. Its symptoms include swollen glands, cough, fever, night sweats etc. Less than one million people in India are affected by this type of cancer every year.

Gupta and Gupta (2014) analysed 98,283 articles from 15 countries published during 2004-2013 on lymphoma research. The articles were downloaded from SCOPUS database. Amongst these, 2,402 articles were published from India with a global share of 2.44%. India ranked 11th among the top 15 most productive countries. India witnessed annual average publication growth rate of 20.17% while world witnessed annual average publication growth rate of 6.0% during the above mentioned period. However average citation per Indian publication was 5.02 and 37.34% of the articles did not receive any citation. International collaboration was involved in 15.04% of the Indian articles.

2.6.3.7 Mouth cancer research

Mouth cancer is a type of cancer which develops in any part of the mouth. It can occur on lips, tongue, gums, inner lining of cheeks, floor of mouth etc. Its risk factor involve use of tobacco or alcohol, infection with Human Papilloma Virus (HPV). More than one million cases per year are seen in India with this type of cancer.

Gupta et al (2014) evaluated 37,049 articles published from all over the world during 2003-2012. They were downloaded from SCOPUS database. 88.14% of the articles were published by top 15 countries. Indian Scientists published 1,832 articles during this time period. India ranked 7th with the publication share of 4.94%. Indians average annual growth rate was 5.15%. The largest share of publication came from the branch of medicine (73.96%). Maximum publications involved tongue (12.17%) and salivary glands (9.28%). 14.85% of Indian publications were through international collaborations. Scientists and institutes from
Maharashtra, Karnataka, Delhi and Tamilnadu working on mouth cancer contributed 61.74% of the total articles.

2.6.3.8 Lung cancer research

It is a type of cancer that begins in the lungs, many times this spreads to other parts of the body like bones and brain. The symptoms include nausea, headache, fever etc. Once it is spread to other parts of the body then it is not curable. More than one million people are affected by lung cancer in India every year.

Jeyshankar and Vellaichamy (2015) assessed the articles published during 1984-2013 on Lung cancer. They werw extracted from SCOPUS database. Good collaboration among the scientists was seen as 94% of the articles were multi authored. USA was a main collaborative partner of India as 24.66% of the total articles were shared by Indian scientists with USA scientists. Tata memorial Hospital, Mumbai was the most productive institute as 16.90% of the total articles were published from this institute. Indian Journal of Cancer published 8.65% of the total articles and was the most productive journal.

2.6.3.9 Prostrate cancer research

The prostrate is a small gland located next to urinary bladder in males. Prostrate cancer is a type of cancer that develops in this gland. More than one million males with this cancer are reported every year from India.

Gupta and Gupta (2015) analysed 89,994 articles published on prostrate cancer from all over the world during 2004-2013. The data was retrieved from SCOPUS database. Of these 1,368 articles were published by the Indian scientists on this subject. Thus Indian global publication share was 1.52% during the above mentioned period. The average citation per article registered by Indian publications was 5.23. Out of 1,368

articles, 67.03% of the articles were original articles followed by reviews (21.35%) and letters (3.07%). 28.65% of the articles did not receive any citation while 0.95% of the papers received more than 100 citations. Indian scientists through international collaboration could publish 320 (23.39%) articles during this time period. India ranked 14th among the top 15 countries. Subjective distribution of articles showed that the highest number of articles were published from medicine (59.50%) followed by biochemistry, genetics and molecular biology (40.13%). Scientists from Delhi, Maharashtra, Uttar Pradesh and Tamilnadu published 57.28% of the total articles published on Prostrate cancer during 2004-2013. Punjab University formed maximum national linkages (15) with 5 research institutes and organisations from India to conduct research work on prostrate cancer. This was followed by Post Graduate Institute of Medical Education and Research, Chandigarh who formed 14 linkages with 3 organisations.

2.6.4 Endocrinology research

Endocrinology is a branch of biology and specifically of internal medicine and it deals with the endocrine system, its diseases and diseases related to specific secretions of the body called hormones.

2.6.4.1 Endocrinology research

Hari kumar et al (2013) analysed 1,160 articles published during 2001-2012 on endocrinology from India. They were retrieved from PUBMED database. The distribution of articles showed that 46% of that articles were original articles followed by case reports (26%) and reviews (15%). Maximum number of articles were published from Delhi followed by Chandigarh and Lucknow. Subjective separation showed that 25% of the total articles were on pancreas and diabetes, followed by bone (19%) and pituitary glands (16%). Indian Journal of Endocrinology and Metabolism (24%) and Journal of Association of Physicians of India (6%) received maximum contribution from Indian authors Dr. Anil Bhansali from PGIMER, Chandigarh published maximum papers as the first author while Dr. Nalini Shah from Seth G.S Medical college, Mumbai published maximum papers as the last author.

2.6.4.2 Diabetes research

Diabetes mellitus is a metabolic disease which is responsible for high sugar level in our body. It is one of the important diseases of endocrinology affecting pancreas, an organ in our body which produces insulin which digests sugar. Gupta et al(2011) analysed 34,744 articles published on diabetes mellitus from all over the world during 1999-2008, using SCOPUS database. Of these 4,824 articles were published from India during the above mentioned period. India ranked 11th among the top 18 countries in this area with its global publication share of 2.86% during this period. However when the cumulative publications output from India was compared during two time blocks(Viz 1999-2003 and 2004-2008), an impressive growth of 114.47% was observed. The 4,824 articles published from India received 20,678 citations during the reported period. The average citation per paper was 4.29. India has contributed 12.15% (586 articles) international collaborative papers share in this field during 1999-2008. Subjective distribution of publication output has shown that 2,840 articles (58.87%) were from medicine followed by biochemistry, genetics and molecular biology (1,382 articles28.65%). High cited top 100 articles received citations in the range of 45-266 citations per article, during the above mentioned period.

2.6.5 Research output on other diseases

Apart from the above mentioned areas, some scholars have evaluated the research output on some diseases. They are listed here.

2.6.5.1 HIV / AIDS research

Acquired Immune Deficiency Syndrome(AIDS)is caused by Human Immune Deficiency Virus (HIV). This retrovirus attacks human immune system and interferes with ability of a person to fight against infections. It is transmitted through contact with infected blood, semen, or vaginal fluid. This is a chronic disease, can last for few years or it can be lifelong. In India AIDS is an epidemic and according to one estimate 2.17 million people were suffering from HIV in India in 2017.

Gupta et al(2011) analysed 1,60,878 articles published from all over the world on HIV/AIDS during 1999-2008.The data was extracted from SCOPUS database. During this period India published 3,325 articles and ranked 12th among the top 20 countries with a global share of 2.07%. The cumulative publication output witnessed

the growth of 108.44% when publications from two time blocks(viz. 1999-2003 and 2004-2008) were considered. The citation impact of 4.22 per article was registered by Indian publications. Only 13 authors from India published more than 25 articles on AIDS during this period. Totally they published 587 articles which accounted for 17.65% of the total research output. These 587 articles received 3,385 total citations with an average of 5.77 citations per articles. When 100 most cited articles were considered they received 7,020 citations with an average of 70.20 citations per articles. These 100 articles were published in 69 journals. International collaboration was involved in 758(22.81%) articles published by Indian scientists. When the articles published under different subjects were considered, the highest research output(2,032 articles,61.11%) was from medicine.

2.6.5.2 Celiac disease research

Celiac disease is an autoimmune disease and is triggered due to a substance called gluten which is a protein and is present in wheat, barley and other grains. An immune reaction occurs in our body with gluten. This is responsible for inflammation and it damages our small intestine. The disease symptoms include diarrhoea, weight loss, gas formation, constipation, abdominal pain etc. About 6.8 million people are suffering from this disorder in India.

Bharadwaj (2013) analysed 14,356 records published on celiac disease from all over the world during 2001-2012. The data was harvested from SCOPUS database. They found that the highest number of articles were published during 2011. 37.35% of the articles were published on adults. During the above mentioned period 299 articles were published by Indian scientists on this disorder. India ranked 15th among the top 20 countries. Indians share of research output was 2.08% in the global research output. 299 articles received 1,870 total citations with 6.25 average citations per article.

2.6.5.3 Asthma research

Asthma is a condition where extra mucus is produced in respiratory track due to which air pipes swell and become narrow. This creates difficulty in breathing and triggers coughing. This cannot be cured but with proper treatment it can be controlled. In 2018, about 1.31 billion people in our country were suffering from Asthma.

Gupta et al(2011) extracted 68,091 records published during 1999-2008 on Asthma from all over the world. SCOPUS database was used for this purpose. The authors observed that only 862 articles were published from India during this period. India ranked 15th in 23 top productive countries with global publication share of 1.27%. The international collaboration output accounted 10.09%(87articles) share. Subject wise 62.88%(542 articles) were published under medicine followed by 200 articles(23.20%) which were published under toxicology, pharmacology and pharmaceutics. The 13 most productive Indian institutes contributed 396 articles (45.94%) on asthma research during above mentioned period. The top 14 authors published 275 articles (31.9%) in the country's output in asthma. The top 100 highly cited papers received citations in the range of 14-373 per article during 1999-2008. Thus the authors concluded that the impact and quality of Indian research is very low as compared to other countries.

2.6.5.4 Typhoid research

Typhoid is an enteric fever caused by bacterial infection. It is caused by the bacteria Salmonella typhi. They can pass through contaminated food and drinking water. The symptoms include high fever, diarrhoea and vomiting. It's incidence in India is 2/1000 population/year under 10years of age.

Gupta et al(2011) evaluated 16,742 articles published during 2000-2009 on typhoid fever from all over the world. The data was extracted from SCOPUS database. During this period India published 940 articles with a global share of 5.61% and ranked 3rd in the top 21 most productive countries. These 940 articles received 2,221 citations with average citation per paper was 2.36 during the above mentioned period. The cumulative publication output of India witnessed growth of 78.93%. When two time blocks (viz 2000-2004 and 2005-2009) were considered. India published 115 articles through international collaborations. This accounts for 12.23% of the total publications. USA was the major international partner publishing 37.39% (43 articles)of the total international publications. Subject wise the highest publication output was from medicine (493 articles and 52.45% share) followed by immunology and microbiology (246 articles, 26.17% share). The top 16 productive Indian institutes published 339 articles (36.06%) out of 940 articles. The top 15 most productive authors contributed 192 articles accounting for 20.42% of total articles published during this period. Indian Journal of Medical Research published 42 articles while

Indian Journal of Microbiology published 34 articles on typhoid during the above mentioned time period. Top 30 most productive journals published 428 articles on this subject during 2000-2009. Top 100 highly cited articles published in 73 journals totally received 2,187 citations with an average of 22 citations per article.

2.6.6 Vector borne disease research

Vector borne diseases are those which are transmitted by the bite of infected insect like mosquitoes, ticks, sand flies etc. Most of them are preventable if we take proper protective measures. The burden of these diseases is the highest in tropical and sub-tropical areas. Malaria and dengue fever are the most common vector borne diseases seen in our country.

2.6.6.1 Malaria research

Malaria is transmitted through mosquito bite. An Anopheles mosquito carries the Plasmodium parasite. The most common parasites are Plasmodium vivax and plasmodium falciparum. When the infected mosquito bites an individual, it transmits the parasites into blood stream of a person. From there the parasite travels to liver where they mature and after several days enter the blood stream and start infecting red blood cells. This results in symptoms of malaria which generally occurs in cycles and lasts for two to three days. India is one of the countries known to have widespread malaria. It shows distinct geographic patches with high or low prevalence of the disease. Such features have burdened our country with high morbidity and socio economic losses.

Gupta and Bala (2011) evaluated articles published on this important disease from India during 1998-2009 using SCOPUS database. It was observed that 2,786 articles from India were published on malaria during this period with an average citation per article of 3.49. India had good publications share of 6.47% during the above stated period. However considering the citations per article it was argued that funding agencies from India should invest more to establish international and national collaborations to improve the quality of research.

2.6.6.2 Dengue research

Dengue fever is a tropical disease caused due to dengue virus. This virus is transmitted through mosquito bite. The mosquito Aedes aegypti and Aedes albopictus

are the primary vectors of dengue. The females of these species transmit the virus through the bites. Their peak biting time is early in the morning and in the evening before sunset. The symptoms include high fever, vomiting, headache, muscle pain etc. It is epidemic in all the states of India. In 2019 more than 67,000 cases of dengue fever have been reported from India. About 2.5% of dengue fever cases die.

Gupta et al(2014) evaluated 8,905 articles published from all over the world on dengue fever during 2003-2012. They were retrieved from SCOPUS database. These articles received 66,948 citations during this period with 7.52 average citations per papers. India published 910 articles on dengue fever during 2003-2012 with 10.22% share in international context. India ranked 2nd in the tap 15 productive countries. Indian 910 articles received 2,978 citations during above mentioned period. Of these, 96 articles (10.55%) were published through international collaboration. Subject wise maximum publication output was from medicine (2,686 articles, 75.38% share) followed by immunology and microbiology (266 articles, 29.13% shares). The top 15 institutes involved in dengue research in India published 381 articles (41.87%) during above mentioned time period; with average publication productivity per institutes was 25.4. the top 15 most productive authors published 298 articles (32.75%) on dengue fever during 10 years. The top 15 most productive journals published 306 Indian research articles (33.63%) in dengue research during 2003-2012.

The severity of dengue in India is under estimated as data on incidence or cost of illness is not properly collected. Therefore, the authors felt that India's research efforts on this disease were low and they should be improved substantially to increase research impact mainly through enhanced collaboration at national and international level.

2.6.7 Articles on global research

Some scholars have tried to understand the growth of the particular subject or area in the world literature by considering their publication output from all over the world. Details about some of the topics are listed here.

2.6.7.1 Hematology research

Kamble and Pradeepa (2015) evaluated the global research output on hematology during 2004-2013. Using ISI Web of Science database 2,50,310 articles were retrieved. Year-wise publications of articles showed that maximum number of

articles i.e. 28,282 were published during 2013 followed by 27,145 articles were published during the year 2007. USA ranked first in the list publishing 37.43% of the total articles while India ranked 21st publishing 1.17% of the total articles during the above mentioned period.

Philip and Abraham(2017) analysed the publications on research in hematology conducted in India during 1996-2012 and compared it with the publication output from all over the world during the same time period. The data was extracted from web based portal of SCImago. They found that India ranked 18th when the countries were arranged according to number of publications in the area of medicine. Amongst these publications they found that hematology is a neglected subject as only 6.5% of these publications were related to hematology. The authors observed that there is plateau in the research trends of Japan and India as against China which showed increasing trend in hematology research publications. They put forward following reasons to explain this. Many doctors after obtaining basic degree in medicine migrate to foreign countries. This increases the burden on the doctors who are working in India. This limits their research activity. With advent in technology, discovery of various aspects explaining the disease causing mechanism there is an ample scope for research but unfortunately a culture of research, mentorship, collaboration is not yet properly recognized by these medical doctors. This is required to fuel the growth of hematology or any other medical speciality in our country.

2.6.7.2 Hemophilia research

Vellaichamy and Jeyshankar (2018) recently assessed the growth in publications on hemophilia research output globally during 2003-2017 using SCOPUS database. Totally 13,503 articles were published on hemophilia (Hemophilia A + Hemophilia B) from all over the world during this period. Of these only 18.48% were single authored underlying the importance of collaborative research in this field. However, degree of collaboration increased gradually from 0.691 in 2003 to 0.890 in 2017. The authors found that there is a rapid growth in literature on hemophilia from 2011 onwards. However Indian contribution to a global research output is only 3.74% thus requiring more focused research on this subject.

2.6.7.3 Measles research

Infection with rubella virus causes measles. This virus enters person's body through mouth, nose or eyes. It travels to lungs from where it starts infecting immune cells. There infected cells move to lymph nodes from where virus is transferred to other cells and finally reaches the blood stream. This is a contagious disease and spreads through physical contact with the patient or being near the patient when he/she cough, sneezing, watery eyes and a red rash throughout the body.

Bala et al (2012) analysed 9,829 articles published on measles from all over the world during 2001-2010. The data was extracted from SCOPUS database. These 9,829 article received 53,672 total citation during the above mentioned period with average citation per paper was 5.46. India ranked 5th among the top 20 countries in measles research. India published 414 articles with the global share of 4.21% during this period. These articles received 1,144 citation with average per paper was 2.98 during this period. India published 65(15.70%) of articles through international collaboration. India established 30 collaboration linkages with USA, 15 with UK and 8 each with France and Switzerland. In the context of different subjects, the highest publication output was from medicine (7,148 articles, 72.72% share) followed by immunology and microbiology (2,460 articles, 25.03% shares). Separate articles were published on various complications seen in measles. The largest number of articles were on respiratory infection (777 articles) followed by encephalitis (735 articles) and pneumonia (615 articles). The top 15 institute from all over the world published 1,704 articles accounting for 17.34% share in the cumulative publication output. Top 15 authors published 774 articles received 9,359 citations during above mentioned period with average of 12.58 citation per article. 'Vaccine' was the most productive journal publishing 490 articles on measles. This was followed by Journal of Virology and Lancet (170 articles each) and Journal of Infectious Diseases (164 articles), during above mentioned period. Among 77 highly cited articles, one articles received more than 1000 citation while 5 articles received citation in range of 500-1000 citations per article during 2001-2010.

2.6.8 Biotechnology/ Biomedicine/ Medicine research

Some scholars evaluated the overall growth of above mentioned topics by considering the publications research output in their areas.

2.6.8.1 Biotechnology research

Patra and Chand (2005) evaluated this subject in India during 1982-2003 by retrieving articles from four different databases. Totally 2,083 articles were analysed. They found that overall growth rate was good except in 1997 where there was a dip. 89% of the articles were co- authored, underlying the importance of collaborative work in this area.

Recently Sharma et al (2019) analysed the same subject. However, the time period selected was different. It was 2008 – 2017, a ten year period. The authors mentioned that today India has a very big pool of qualified workforce and well equipped modern laboratories which can be utilized to develop various areas in biotechnology research. Using SCOPUS database authors found that 73,153 articles were published globally on biotechnology research during the above stated 10 year period. India ranked 3rd among the top ten most productive countries in biotechnology research with a publication share of 7.62%. They found that 5,574 articles were published from India on this subject during 10 year period with an average citation per article was 14.25 and annual mean relative growth rate was 0.33%. Out of these publications 48.79% publications were related to biochemistry, genetics and molecular biology. However they observed that the average citation per publication received by India during 2008-2017 was less as compared to other developed countries. So they finally commented that in spite of having good facilities for research, there is a need to increase the quality of research and also to strengthen the international collaboration.

2.6.8.2 Biomedical research

Satyanarayana (2001) analysed Indian scientist's contribution in biomedical research. Surprisingly he observed that there was a decline in publication under this subject in 1990 which declined to 3,241 articles published during 1994.

2.6.8.3 Research output in medicine

Gupta and Bala (2011) evaluated research activities of India in the area of medicine during on various aspects of medicine from all over the world during the above mentioned time period were extracted from SCOPUS database. In this, Indian cumulative output was 65,745 articles with the annual publication growth rate of

11.57%. India accounted for 11.87% (7,824 articles) share of international collaboration papers in medicine.

2.6.9 Research on other related areas

Some scholars assessed the growth of certain topics like anemia, genetics, and heredity, life science etc. by analysing the publication output on these subjects. Some of such examples are listed here.

2.6.9.1 Anemia research

Anemia is a common symptom in all the disorders selected in the present study. In simple term it can be explained as it is caused due to low hemoglobin level. It is a risk factor for increasing morbidity and mortality in several disorders.

. Vellaichamy and Jayshankar (2014) evaluated research output on anemia research in India during 1993-2013 by retrieving data from SCOPUS database. 5,085 articles were published on anemia during above mentioned ten year period. The most productive year was 2013 when 739 articles were published while the least productive year was 1996 when only 47 articles were published. Inverse relationship was seen when the citation data was considered. 739 articles received only 178 citations while 47 articles received 3,245 citations. 92.27% of the total articles were multi authored. Articles on anemia during 1993-2013 from India have been published under 27 different subjects. Amongst these 768 articles with a publication share of 11.73% have been published under biochemistry, genetics and molecular biology. This ranked second in the 27 subject list.

2.6.9.2 Life science research

Garg and Kumar (2014) conducted quantitative analysis of 9,957 articles published by Indian scientists on 12 various disciplines of life science during 2008-2009 and found that maximum articles were published by the scientists who were working in different academic institutes. Interestingly they observed that women scientists generally work in small teams and have less international collaborations as compared to male scientists.

2.6.9.3 Genetics and heredity research

Garg et al (2010) performed quantitative analysis of 2,899 articles published on genetics and heredity by Indian scientists during 1991-2008 using Science Citation Index-Expanded (SCI-E) database. They observed that during initial stages the growth was slow but was maximum during 2005-2008. Growth in the area of molecular genetics was maximum during 2005-2008. Molecular genetics was the area which showed maximum output (70.16%) during this period. The citation analysis data indicates that 2,899 articles received 31,299 citations. So the average rate of citation per article for the Indian research output was 10.80. About 30% of the articles were cited more than 10 times.

2.6.9.4 Hereditary disorders research

This is the only article where various heredity disorder have been evaluated by analysing the research output published on them.

In 2012, Gupta published an article on scientometric analysis of articles published on some hereditary disorders seen in India using various parameters like the global publications share of India, India's growth in this area, citation impact etc. Time period selected was 2002-2011. Five hereditary disorders viz. sickle cell anemia, thalassemia, von Willebrand disease, hemophilia and thrombopenia were selected for the study. Of these three disorders viz. sickle cell anemia, von Willebrand disease and hemophilia are studied in the present work. Therefore, details about these three disorders published in this article is compiled here. The publication output from India during 2002-2011 on sickle cell anemia was 193 articles (20.96% share) followed by hemophilia (105 articles, 11.40% share) and von Willebrand disease (99 articles,10.75% share). The author observed that articles published on hereditary blood disorders from India during 10 years (2002-2011) were under six subjects. Amongst these, 175 articles (19.00% of the total share) were published under biochemistry, genetics and molecular biology. However, these articles received only 2.30 citations per year. However international collaborative publications share data revealed that 19.19% of the articles were on von Willebrand disease followed by hemophilia (14.29%) and sickle cell anemia (6.74%). When the most productive institutes in India where work on these disorders is being carried out, were considered, National Institute of Immunohematology Mumbai ranked first (145 publications) followed by All India Institute of Medical Sciences, New Delhi (107

publications) and King Edward Memorial Hospital, Mumbai (86 publications). Christian Medical College, Vellore (65 publications) ranked fourth but they ranked first in Average Citations Per Paper (ACPP) – 6.97 and H index (14) amongst the top productive institutes.

2.6.10 Biochemistry, genetics, molecular biology research

The subject of molecular biology is inter-related with biochemistry and genetics. Some scholars have tried to analyze the research output on all the three subjects collectively from India as well as from other countries like China and South Africa.

Bala and Gupta (2010) analyzed the research output from India on this subject. 1998-2007 was the time period selected for the study and the data was retrieved from SCOPUS database. Totally 45,712 articles from India were published on this subject during the stated time period. This was 2.37% of the cumulative publication output from all over the world. India ranked 10th among the top 30 most productive countries in the world. India's global publications share was 1.77% in 1998 which increased to 3.07% in 2007. India's annual publication growth rate was 11.5% during 1998-2007; however, when four year block (2003-2007) was considered, the growth rate was 16.96%. The authors identified 20 institutes in India as the most productive institutes where more than 500 articles have been published from each of those institutes during 1998-2007. The Average Citation per Paper (ACPP) received for those publications was 3.21. When two time blocks viz. 1998-2002 and 2003-2007 were considered the average growth rate of these 20 institutes was 89.15% which is a very impressive achievement.

Amin and Parekh (2019) attempted to evaluate the research output on biochemistry, genetics and molecular biology from Gujarat University, Ahmedabad during 38 years (1980-2018) time period using SCOPUS database. Only 400 articles on this subject were published from this University during the above mentioned time period. They found that publication trend increased considerably during 2012-2017 with qualitative articles. Otherwise the overall performance was very poor

He et al (2005) analyzed the research articles published in biochemistry and molecular biology from China. They constructed a database for China for a period of three years (1999-2002) using Science Citation Index – Expanded (SCI-E) as a main source of data. Totally 4,502 articles were published from China during three years

in 272 journals. When the growth rate was considered, it was 5.96%, 19.53% and 8.83% for three years with an average rate of increase of 12.6%. Out of 4,502 articles, 91.56% of articles were published.

Molatudi and Pouris (2006) assessed the research output of microbiology, genetics and molecular biology in South Africa during 20 year period from 1980-2000 after extracting data from the ISI database. It was observed that maximum number of articles on genetics and molecular biology were published in 1996 however subsequently they were decreased by 17% during the last four years. On the other hand, there was a sharp increase (60%)in number of articles published in the area of microbiology during 1996-2000.

Recently Ramdoss and Yadalla (2020) analysed 2,633 articles published in Indian Journal of Ophthalmology during 2005-2017. Subspecialty wise analysis of distribution of these articles has been done across the time. Similarly the articles have also been analysed as original articles, care reports, editorials, letter to editor, review articles etc. The authors found that there is a healthy trend of progressive increase in the number of paper published year after year. Original articles and care reports were the main types of articles published during this period. There is a slight reduction in the number of care reports over the time. The authors claim that this is the first article where data from Indian Journal of Ophthalmology has been analysed scientometrically. This eventually will help the readers to get a better perspective of the direction in which the journal is headed in future.

Thus in the present chapter the scholar has compiled the result of the various scientometric analysis conducted on various disorders and other areas of science particularly medical science. If we observe the detail of these references it can be seen that most if these studies have been published during 2010-2014. Thus this time period can be considered os 'Golden Period' for scientometric studies in medical sciences. However amongst these studies there are only 2-3 reports where scientometric analysis of research output on hereditary disorders has been assessed.

In the present study, the scholar has extensively evaluated the research output of six genetic/hereditary disorders Viz. Down syndrome, Sickle cell anemia, G6PD deficiency, Hemophilia A, Hemophilia B and von Willebrand disease, by scientometric analysis which is a new area. In this study the scholar has tried to understand the impact of molecular biology on research output on the above

mentioned disorders from India as well as from all over the world. In the next chapter, the basic information about the above mentioned six genetic disorders is presented.

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Chapter- III

REASEARCH METHODOLOGY

Research Methodology

Research means detailed study of a particular problem using scientific methods. The main objective of research is to explore the unknown and during the process unlock new possibilities.

Research methods are specific procedures where data is collected and analysed systematically. Selection of method depends on what type of data is required to answer research questions.

3.1 Methodology

This is the path through which any researcher plans to conduct research. Initially the researcher formulates the problem, accordingly design the objectives and then using various steps or specific methods, the answers are derived. The present study has involved two types of research.

1) Descriptive research
2) Exploratory research

The descriptive research explains the background of the subject. This includes brief description about various metrics, detail descriptions of scientometrics review of various scientometric studies conducted by different scholars.

Exploratory research focuses on the analysis of the data on six genetic disorders using scientometric indicators. These two approaches gave the scholar opportunity to investigate the research problem and find answers to the research questions.

3.1.1) Survey method

In the present study preliminary data was collected through survey method. The survey was conducted to screen the database and to download the research output on six genetic disorders. Here the variables were considered as publications on six genetic disorders retrieved from database. These publications were analysed systematically to get the answers to the research questions.

3.2) What is a database?

Data is information collected on any aspect. It can be simple names of individuals, weight of individuals, addresses of employees etc. Using computers this

information is changed into a particular format for efficient movement and processing. Thus database is a place where different types of data can be stored in an organized way and it can be accessible by everybody. It can be updated regularly by the administrator. In the case of Library and Information Science database is mainly list of articles obtained from various sources. These sources can be scientific journals, proceedings of the conferences, newspapers etc.

Depending on the sources, number of items in the database varies considerably. Each data base has its own advantages and disadvantages. Access to database is either free or charged.

3.2.1) Selection of the database for the present study

To conduct this study following databases were taken into consideration – INDMED, SCIENCE DIRECT, SCOPUS and PUBMED. Basic information about these databases is given below.

i) INDMED - It is a database of selected peer reviewed medical journals published from India. It covers more than 100 journals indexed from 1985 onwards and supplements PUBMED.

ii) SCIENCE DIRECT – It is a website owned by Elsevier which provides subscription based access to a large database of scientific and medical research articles. It has 12 million articles from over 3500 journals.

iii) SCOPUS – It is a private, largest multidisciplinary database owned by Elsevier. It was developed in 2004. It covers more than 20,000 peer reviewed journals from over 5000 publishers. The journals are from scientific, technical, medical and social sciences. It is an extremely flexible database. It helps to analyze titles, keywords, coupling of authors and co-authors etc. However, access to this database is charged. The second limitation of this database is analysis of the large data as it can handle only 2000 titles for further analysis.

iv) PUBMED - It is a free database. The United States National Library of Medicine (NLM) maintains this database. It contains more than 28 million articles from MEDLINE, online books, life science journals etc. It provides quality control in scientific publishing. Only journals that meet PUBMED's scientific standards are indexed or listed. It provides access to older references of ' Index Medicus' back to 1951 and even earlier. Articles on selective topics are there up to the year 1865 while articles on very selective topics are there even up to 1809. It has also very

recent entries to records. Sometimes it provides link to full text of articles or publishers website from PUBMED central. About five lakh new records are added every year to this database.

From the above mentioned information it was evident that PUBMED was the most comprehensive database to retrieve the data on the above mentioned genetic disorders. Similarly in the present study older publications up to 1990 and during 1991-2014 were required to evaluate the growth in publications related to these disorders. Therefore, it was decided to use PUBMED database to retrieve data for the present study.

3.3) Retrieval of the data

The following strings were used to retrieve the data on six genetic disorders.

1) Sickle cell anemia - Sickle cell anemia, Sickle cell disease, sickle hemoglobin

2) Hemophilia A - Hemophilia A, Factor VIII

3) Hemophilia B - Hemophilia B, Christmas disease, Factor IX.

4) von Willebrand disease – von Willebrand disease, vWD, von Willebrand Factor, vWF.

5) Down syndrome - Down syndrome, Trisomy 21, Aneuploidy.

6) G6PD deficiency - G6PD deficiency, Fava beans.

Thus data on six genetic disorders published up to 2014 was retrieved. Totally six files were generated. An additional string 'India' was added to each file. This helped to get the articles published from India on each genetic disorder. The remaining data included the articles published from all over the world on each disorder. Thus finally twelve files were generated. Six files contained the data from India while six files contained the data from all over the world on the above mentioned six genetic disorders.

Details of this preliminary data retrieved from PUBMED are shown in Table 3.1. Totally 1,606 articles published on six genetic disorders from India were found while 93,919 articles were published from all over the world. The table shows that India's share on publication of the articles on these disorders is 1.71% as compared to the world publications. India's share on individual disorders ranges between 1.01%-3.39%.

As seen in the table, the world wide data on different disorders was in thousands. So it was very difficult to handle these files. Therefore for convenience

each file was divided into separate files, each file containing 2000 publications. This generated a preliminary database for the present work. While scrutinizing the files containing the articles published from all over the world, it was found that there were many annotation errors in various publications, especially those which were published during 1940s and 1950s. These included absence of PMID number, absence of proper title etc. All these incomplete entries were deleted. Similarly lines showing headings like only title or only author name or only

Table 3.2 reveals number of articles under each disorder available for analysis after deleting annotation errors. Now record of each article contained title of the article, names of all the authors, name, volume and page number of the journal and year of publication. This data was fed into MS Excel worksheet for further analysis.

As mentioned earlier, the discovery of PCR revolutionized the field of molecular biology. Scientists from all over the world started using PCR based techniques to study the above mentioned disorders. Therefore to understand the impact of molecular biology on the publications of these disorders it was decided to separate the publications into two groups – articles published up to 1990 and articles published during 1991-2014. Therefore articles published from India as well as from all over the world were separated in to two groups. Thus for each disorder four files were created as follows.

1) Articles published from India

a) Up to 1990

b) During 1991-2014

2) Articles published from all over the world

a) Up to 1990

b) During 1991-2014

Totally 24 files were created for six disorders for further analysis.

While going through these publications it was observed that many nonspecific publications not related to that particular disorder were also listed under that disorder. So it was decided to delete those nonspecific publications from the list. Following are some of the examples or reasons why nonspecific publications which were not directly related to the particular disorder but were still listed under it.

Table 3.1: Details of preliminary retrieval of data from PUBMED database on six genetic disorders published from India as well as from all over the world

Sr. No.	Disease	Country	PubMed Record
1	Down syndrome	India	430 (1.36)
	Down syndrome	Worldwide	31,548
2	G6PD deficiency	India	252 (3.39)
	G6PD deficiency	Worldwide	7,424
3	Hemophilia A	India	249 (1.29)
	Hemophilia A	Worldwide	19,312
4	Hemophilia B	India	83 (1.89)
	Hemophilia B	Worldwide	4,391
5	von Willebrand disease	India	75 (1.01)
	von Willebrand disease	Worldwide	7,435
6	Sickle cell aanemia	India	517 (2.17)
	Sickle cell anemia	Worldwide	23,809
TOTAL		**India**	**1,606 (1.17)**
TOTAL		**Worldwide**	**93,919***

Figures in brackets indicate percentage values

*After deleting the entries showing annotation errors like absence of PMID number, absence of proper titles and also deleting the lines showing headings like only author name, journal name, etc., this number is reduced to 86,555 for further analysis.
PMID number were also deleted. Then the total number of international publications reduced to 86,555.

Table 3.2: Number of articles published from India on six genetic disorders and available for analysis after deleting nonspecific publications

Genetic Disease	Up to 1990		1991-2014		Pub Med Records	After Non specific Deletions
	Pub Med Record	After non specific Deletions	Pub Med Record	After non specific Deletions		
Down syndrome	88	32	342	192	430	224
G6PD deficiency	124	117	128	118	252	235
Haemophilia A	19	13	230	210	249	223
Haemophilia B	07	06	76	73	83	79
vonWillebrand disease	0	07	75	63	75	70
Sickle cell anemia	174	101	343	243	517	344
Total	**412**	**276**	**1,194**	**899**	**1,606**	**1,175**

Explanation - 1,175 articles published from India were available for detail analysis.

1) Down syndrome - Any article with the word 'Down' or 'syndrome' in its title but not related to Down syndrome was listed under this disorder. Similarly articles on other cytogenetic disorders were also listed under this disorder. All of them were deleted.

2) Sickle cell anemia - Any article on beta globin gene or some times on thalassemia was listed under this disorder. They were all deleted.

3) Hemophilia A - Publications on other coagulation disorders like hemophilia B or Factor XIII deficiency were also listed under this disorder. Similarly articles on hepatitis B, hepatitis C, HIV were also listed under this disorder. They were all deleted.

4) Hemophilia B - Publications on hemophilia A or on other coagulation disorders were listed under this disorder. Similarly articles on viral diseases like hepatitis, HIV etc. were also listed under this disorder. They were all deleted.

5) von Willebrand disease - Along with articles on Hemophilia A and Hemophilia B some articles on viral diseases like hepatitis and also on diabetes mellitus, primary biliary cirrhosis etc. were also listed under this disorder. They were all deleted.

6) G6PD deficiency - Articles related to hemolytic anemia due to various cancers including leukemia were listed under this disorder. Similarly articles on selenium poisoning, lead poisoning, Paroxysmal Nocturnal Hematuria (PNH) or articles published on red cell enzymes other than G6PD were also listed under this disorder. They were all deleted.

While deleting non-specific articles from the list, help was taken from the expert who was well versed with these disorders and as per his decision the nonspecific publications were removed. After deleting these articles, the number of articles under each disorder was reduced.

Table 3.3 depicts number of articles published from India and was available for analysis after removing nonspecific articles. As compared to PUBMED records out of 1,606 articles only1,175 articles were available for further analysis. In the case of Down syndrome, out of 430 records only 224 articles were taken for further analysis.

Table 3.4 shows number of articles published from all over the world on each disorder up to 1990 and available for analysis after deleting nonspecific articles. In the case of Down syndrome only 58.5% of the total articles retrieved from PUBMED

were available for further analysis while in the case of remaining five disorders 61.2-83.1% articles were available for further analysis.

Table 3.5 shows number of articles published from all over the world during 1991-2014 and available for further analysis after deleting nonspecific articles. In the case of Down syndrome only 46.11% articles were available for analysis while in the case of remaining disorders 78.99-94.33% articles were available for further analysis.

3.4) Pattern of analysis

Articles published from India as well as from all over the world were analyzed using following indicators.

1) Year-wise trend of publications
2) Journal-wise trend of publications
3) Category-wise separation and analysis of the articles
4) Calculation of growth rate of the publications published during 1991-2014.
5) Trend of publication of articles on six genetic disorders during 2015-2020.

Table 3.3: Preliminary analysis of articles published from all over the world and retrieved from PUBMED database on six genetic disorders

Disease	Up to 1990	1991-2014	Total
1. Down syndrome	9,325	20,981	30,306
2. Sickle cell anemia	8,392	11,704	20,096
3. Hemophilia A	8,652	10,274	18,926
4. Hemophilia B	1,683	2,607	4,290
5. von Willebrand disease	2,580	4,746	7,326
6. G6PD deficiency	3,544	2,067	5,611
Total	**34,176**	**52,379**	**86,555**

Explanation - 86,555 articles published from all over the world were available for detail analysis after deleting entries showing annotation errors.

Table 3.4: Number of articles published from all over the world up to 1990 available for analysis after deleting nonspecific publications

Sr. No.	Disease	Up to 1990	Articles for further analysis	
			Number	Percentage
1	Down syndrome	9,325	5,455	58.5
2	G6PD deficiency	3,544	2,666	75.2
3	Hemophilia B	1,683	1,399	83.1
4	Hemophilia A	8,652	6,962	80.5
5	Sickle cell aanemia	8,392	6,649	79.2
6	von Willebrand disease	2,580	1,579	61.2
	Total	**34,176**	**24,710**	**72.3**

Explanation - 24,710 articles i.e. 72.3% of the total articles (34,176) published from all over the world up to 1990 were analysed in detail.

Table 3.5: Number of articles published from all over the world during 1991 to 2014 available for analysis after deleting nonspecific publications.

Disease	Published during 1991-2014	Publications for further analysis	
		Number	Percentage
Down syndrome	20,981	9,675	46.11
Sickle cell anemia	11,704	9,245	78.99
Hemophilia A	10,274	8,611	83.81
Hemophilia B	2,607	2,324	89.14
von Willebrand disease	4,746	4,477	94.33
G6PD deficiency	2,067	1,784	86.31
Total	**52,379**	**36,116**	**68.95**

Explanation - 36,116 articles i.e. 68.95% of the total articles (52,379) published from all over the world during 1991-2014 were analysed in detail.

1) Articles published under each disorder were separated on the basis of number of publications per year, which gave us the year-wise trend of publications on each disorder.

2) Number of journals in which the articles on particular disorder were published every year were counted to get journal-wise trend of publications on each disorder.

3) While going through the titles of the articles especially published during 1991-2014 on each disorder, it was realized that articles have been published on various aspects including also those using molecular biology tools. So these articles were separated into six broad categories as follows;

i) Research /Original articles – The articles explaining certain aspects or giving any new information, presenting any new technology developed to detect the disorder or finding the prevalence of the disorder in particular area or state in India or in other parts of world were included under this category.

ii) Articles based on clinical findings - All the six genetic /hematological disorders selected for the present study affect various organs or systems in our body. The extent of damage and severity is variable in different patients. Many articles were published explaining the clinical variability seen in different disorders. They were grouped under this category.

iii) Case reports - Lot of clinical variability is generally seen among individuals suffering from these disorders. Some rare cases reveal atypical clinical presentations. Such interesting cases have been published as 'Case reports'. All such case reports were clubbed under this category.

iv) Reviews - Lot of advances are taking place over the years to understand the pathophysiology of these disorders. Several review articles evaluating this information have been published on these disorders. They were put together under this category.

v) Miscellaneous - Apart from scientific aspects many times articles have been published on various nonspecific aspects related to that particular disorder. For example, articles on phonological processes, behavioral abnormalities, physical anomalies, management of mentally handicapped babies have been published under Down syndrome. Articles on counselling, nursing, cost burden etc have been published under Sickle cell anemia. All these articles were brought under this category.

vi) Articles explaining the molecular biology aspects or the articles published by using molecular biology tools - This is the most important category. Articles published under this category will tell us the impact of molecular biology in developing the research related to particular disorder. Articles involving data related to various molecular biology aspects were considered under this category.

While separating out the articles under each category, again guidance was taken from the expert in the related areas.

4) Calculation of growth rate of the publications of articles published during 1991-2014.

To calculate the growth rate of the articles published during 1991-2014 on each disorder, the articles were separated into two groups of 12 years' time interval.

i) Articles published during 1991 – 2002
ii) Articles published during 2003 - 2014

The following formula was used to calculate the growth rate.

Increase in publications = Number of publications during 2003-2014 - Number of publications during 1991-2002.

$$\text{Growth rate} = \frac{\text{Increase in number of publications} \times 100}{\text{Number of publications during 1991-2002}}$$

Using this formula growth rate for articles published on each disorder from India as well as from all over the world was calculated.

3.5) Network analysis

Network analysis was used for quantitative analysis of the data. Bibliometrix package recently developed by Aria and Cuccurullo (2017) was used for this purpose SCOPUS database was accessed to retrieve the articles published on six genetic disorders during 1991-2014. As this is a very advanced computer package and the candidate is not well versed with it so the help was taken from an expert who could handle and analyze the data comfortably. It was realized that even though SCOPUS is a very flexible database, it can handle only 2000 titles for further analysis. Therefore, it was decided to analyze only the articles published from India during

1991-2014 on six genetic disorders. Due to large sample size it was not possible to analyze the articles published from all over the world on these disorders. The articles published from India during 1991-2014 were further divided into two groups. The first group included articles published on particular disorder without involving any data on molecular aspects while the second group included articles on the same disorder involving data on molecular aspects.

The following strings were used to separate the data on molecular aspects.

1) Down syndrome: chromosome engineering, micro RNA, mitochondria, Ts65Dn.
2) G6PD deficiency: X inactivation, mutations, RFLP, Polymorphism, genotypes.
3) Sickle cell anemia: haplotype, polymorphism, DNA genomics, beta globin gene.
4) Hemophilia A: mutation linkage analysis, DNA polymorphism.
5) Hemophilia B: mutations, DNA, linkage analysis polymorphism.
6) Von Willebrand disease: mmultimers, subtypes, VNTR, molecular pathology, mutations.

Thus twelve sets of articles (two for each disorder) were prepared for further analysis of six genetic disorders.

All the twelve groups were converted into 'R-format'. Then bibliometric analysis using the above mentioned package was done to build matrices for coupling / collaboration of authors, average article citations per year, average total citations per year, growth of the particular subject judged on the basis of trend of publications etc. The following indicators were directly obtained after analysis of the data for each set of genetic disorders .

1) Coupling or collaboration of authors in the form of network. One or more clusters are seen. Separate colour to each cluster indicates that they are independent. Each circle in the cluster represents individual group or author and is called "node". The size of the node is directly related to the number of his/her documents. Different nodes are connected by lines which are called links. They represent the co-operation among the authors. The wider the line, the stronger is the co-operation. The authors in a big cluster are generally situated very close to each other showing strong

connections while those in the smaller clusters are far away from the main cluster and require to strengthen their co-operation.

2) Top 5 – 8 journals revealing trends of articles published on the particular disorder from India during 1991-2014.

3) Average article citations per year.

4) Average total citations per year.

5) Most productive top ten authors calculated on the basis of fractionalized articles.

6) Top ten articles published on different disorders and received maximum citations.

There are two main methods in bibliometrics for quantitative measurement of scientist's publications. They are i) Full counting method and ii) Fractional counting method. In a full counting method. A multi authored publication is counted with a full weight of one for each author. It means that over all weight of the publication is equal to the number of authors. While in the fractional counting method, a multi authored publication is assigned fractionally to each of the co-authors with the overall weight of the publication is equal to one. Now this technique is slightly modified to consider authors contribution to the specific publication and his/her ranking in the list of authors of that publication In the present study this method is used to give weightage to different authors.

6) Highly cited top ten articles .

Chapter IV

Data analysis

4.1 Results

4.1.1 Year-wise and journal-wise trend of publications of articles

4.1.1.1 India

4.1.1.1.1 Up to 1990.

Table 4.1 describes year-wise publication trend of articles published from India up to 1990 on six genetic disorders. Maximum number of articles have been published on G6PD deficiency (117) followed by those on sickle cell anemia (101). First article on sickle cell anemia appeared in 1952 from India; while on the remaining disorders the articles started appearing since 1962. G6PD deficiency was first reported in India in 1963. First publication on Down syndrome from India appeared in 1964 and since then only 32 publications appeared up to 1990. Very few articles have been published on hemophilia A, hemophilia B and von Willebrand disease from India up to 1990 (range 7-13 articles).

Figure 4.1 depicts journal-wise trend of articles published from India up to 1990 on six genetic disorders. Articles on G6PD deficiency have been published in 37 journals while articles on sickle cell anemia have been published in 32 journals.

Table 4.2 reveals top five journals where maximum number of articles from India has been published up to 1990. In the case of 5 out of 6 disorders maximum number of articles have been published in JAPI. Journal of Physicians of India (JAPI) is one of the oldest medical journals published from India. (started from 1944 at Madras). Therefore many articles on these disorders were published in JAPI at that time. For example, out of 117 articles published o G6PD deficiency in 37 journals, as many as 26 articles were published in JAPI. While in the case of sickle cell anemia, 29 out of 101 articles were published in JAPI. 57.43%, 53.13% and 48.72% articles on sickle cell anemia, Down syndrome and G6PD deficiency respectively were published in top 5 journals.

4.1.1.1.2 During 1991-2014.

Table 4.3 reveals year- wise trend of articles published on six genetic disorders from India during 1991-2014. Considerably more number of articles have been published during this time interval as compared to the earlier time interval (up to 1990). However the rate of publication was slow during initial years. . Therefore half-life for the articles on these disorders ranged between 13-19 years.

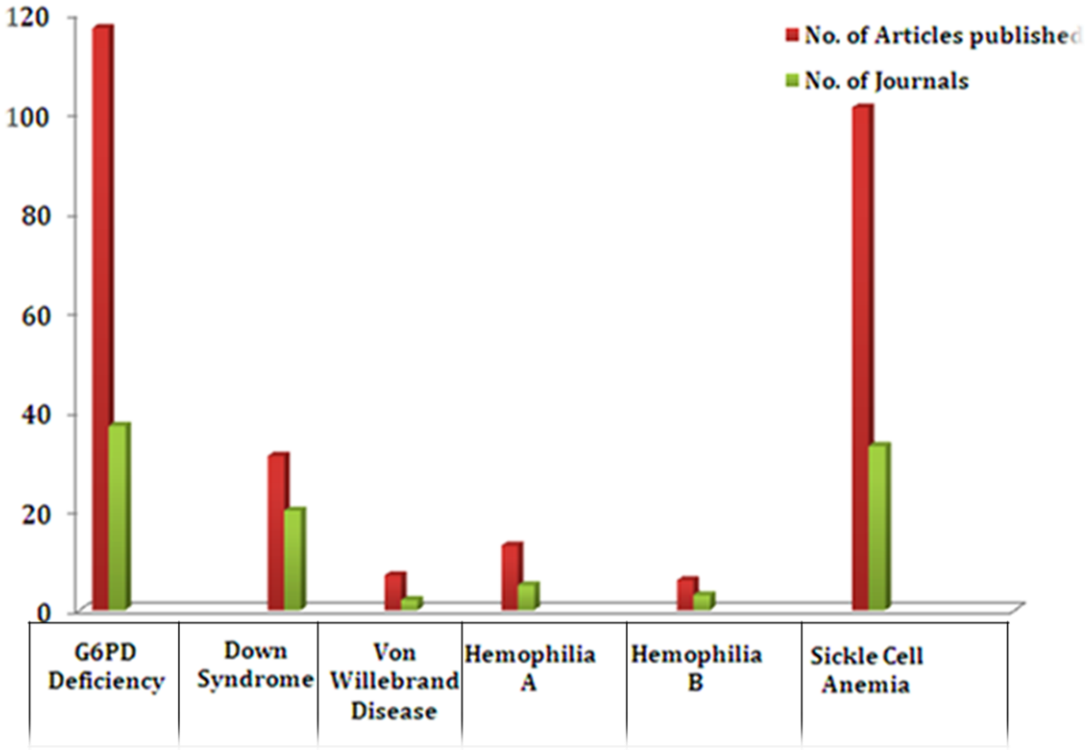
Table 4.1: Year- wise publications of articles up to 1990 published from India on six genetic disorders.

Year	Sickle cell anemia	Hemophilia 'A'	Hemophilia 'B'	von Willibrand disease	G6PD deficiency	Down syndrome
1952	2	0	0	0	0	0
1953	0	0	0	0	0	0
1954	0	0	0	0	0	0
1955	0	0	0	0	0	0
1956	0	0	0	0	0	0
1957	0	0	0	0	0	0
1958	3	0	0	0	0	0
1959	0	0	0	0	0	0
1960	0	0	0	0	0	0
1961	0	0	0	0	0	0
1962	0	1	0	0	0	0
1963	1	0	0	1	2	0
1964	1	0	0	0	1	1
1965	0	0	0	1	0	1
1966	1	0	0	1	2	0
1967	3	0	0	0	4	0
1968	4	0	0	0	7	1
1969	6	0	0	0	3	0
1970	2	0	0	0	0	1
1971	1	0	0	0	6	1
1972	0	1	2	1	6	1
1973	4	0	1	1	5	2
1974	2	0	0	0	5	0
1975	3	0	0	0	3	2
1976	2	0	0	1	4	1
1977	3	0	0	0	6	1
1978	3	0	0	0	6	0
1979	3	0	0	0	1	1
1980	3	0	0	0	3	0
1981	5	2	1	1	7	4
1982	3	1	1	0	7	0
1983	4	0	0	0	4	1
1984	3	0	0	0	6	0
1985	3	0	0	0	7	1
1986	7	1	0	0	0	1
1987	10	2	0	0	5	3
1988	9	0	0	0	3	1
1989	4	0	0	0	5	2
1990	6	5	1	0	9	6
Total	**101**	**13**	**6**	**7**	**117**	**32**

Most productive years of publications for different disorders were as follows-

SCA- 1987, HepA- 1990, HepB- --, vWD- ---, G6PD def- 1990, DS- 1990.

Figure 4.1: **Articles published up to 1990 from India on the six genetic disorders in various journals .**



Observation- Relatively small number of articles on von Willebrand disease, Hemophilia A and Hemophilia B have been published from India up to 1990 as compared to those published on remaining three disorders.

Table 4.2: List of top five journals where maximum numbers of articles from India published on each genetic disorder up to 1990.

Genetic Disorder	Down syndrome	Sickle cell anemia	Hemophilia A	Hemophilia B	*von Willebrand disease	G6PD deficiency
Top Five Journals	Ind J Paed 05	JAPI 29	JAPI 09	JAPI 04	JAPI 06	JAPI 22
	Human Genetics 04	IJMR 11	IJMR 01	IJMR 01	Ind J Paed 01	IJMR 12
	IJMR 03	JIMA 07	Blood 01	Ind J Paed 01		Ind Paed 11
	Lancet 03	Ind Paed 06	BJCP 01			Ind J Patho & M 07
	Ind Paed 02	Am J Haemat 05	Ind J Paed 01			Human Genetics 05
Total	**17**	**58**	**13**	**6**	**7**	**57**
Total No. Articles	**32**	**101**	**13**	**6**	**7**	**117**
No. of Independent Journals	**18**	**32**	**5**	**3**	**2**	**37**
% Representation of top 5 Journals	**27.78**	**15.63**	**100**	**100**	**100**	**13.51**
% Representation of Articles in top 5 Journals	**53.13**	**57.43**	**100**	**100**	**100**	**48.72**
% representation of Articles in first Journal	**15.53**	**28.71**	**69.23**	**66.67**	**85.71**	**18.8**
Articles/Journals	**1.78**	**3.16**	**2.6**	**2**	**3.5**	**3.16**

Ind J Patho & M - Indian Journal of Pathology and Microbiology, Ind Paed – Indian Paedeatrics Ind J Paed- Indian Journal of Paedeatrics, BJCP- British Journal of Clinical Practice, IMA-Journal of Indian Medical Association, IJMR – Indian Journal of Medical Research ,JAPI- Journal of Association of Physicians of India, Am J Haemat – American Journal of Haematology.

*** There are no articles seen in PUBMED database during 1982 – 1990 on von Willebrand disease**

Table 4.3: Year- wise publications of articles on six genetic disorders published during 1991 – 2014 from India.

Year	Sickle cell anemia	Hemophilia 'A'	Hemophilia 'B'	von Willibrand disease	G6PD deficiency	Down syndrome
1991	6	1	1	0	3	4
1992	5	1	0	0	11	2
1993	9	4	2	2	5	2
1994	8	0	0	1	6	2
1995	5	1	0	1	5	1
1996	7	2	1	0	1	4
1997	10	2	2	0	6	7
1998	3	10	2	0	0	6
1999	5	8	0	0	2	1
2000	11	9	2	3	1	2
2001	5	15	8	0	6	4
2002	6	7	2	0	5	7
2003	3	14	4	5	7	9
2004	7	16	6	1	3	3
2005	12	12	1	10	6	8
2006	8	7	2	5	4	5
2007	9	17	6	2	8	11
2008	7	10	6	2	7	11
2009	8	12	5	3	7	11
2010	13	8	3	4	9	16
2011	14	12	6	5	5	13
2012	32	13	6	4	5	18
2013	25	12	5	7	2	23
2014	25	17	3	8	4	22
Total	**243**	**210**	**73**	**63**	**118**	**192**
Half Life Years	**18**	**16**	**17**	**18**	**13**	**19**

Most productive years of publications for different disorders were as follows-

SCA- 2013, 2014, HepA- 2007, 2014, HepB- 2001,

vWD- 2005, G6PD def- 1992, DS- 2013.

Figure 4.2 depicts journal-wise trend of articles published from India on six genetic disorders during 1991-2014. Maximum number of journals published articles on sickle cell anemia during this period, followed by those on Down syndrome and hemophilia A.

Table 4.4 reveals top 5 journals where maximum number of articles published from India during 1991-2014 on six genetic disorders has been listed. Percent representation of articles in top five journals ranges between 24.58-43.33%. It can be seen that number of journals in which the articles on each disorder have been published, have increased considerably (range 34-115). One of the reasons for this may be introduction of subject specific journals. For example, 'Hemophilia', 'Thrombosis and Hemostasis' are the journals which were started during last decade of 20th century and are specific for coagulation disorders. Therefore 63 out of 210 publications on hemophilia A (30%) and 20 out of 73 publications on hemophilia B (27.4%) have been published in 'Hemophilia' journal during 1991-2014.

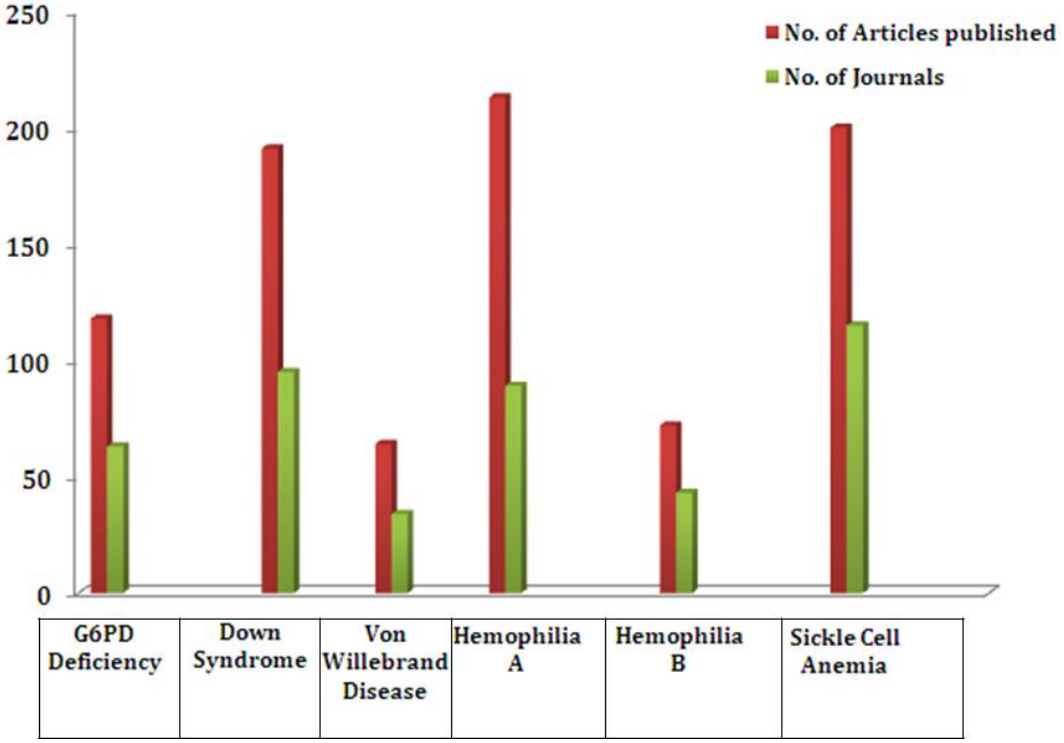
4.1.1.2 All over the world.

4.1.1.2.1 Up to 1990

Figure 4.3 represents year-wise trend of articles published on Down syndrome from all over the world up to 1990 in different journals. The first article appeared in 1945, while the sizeable Mumbai of publications started since 1950. Then the publications were steadily increasing up to 1960, and since then there was a considerable increase in number of publications. Similarly number of journals in which these articles were published, also increased significantly.

Figure 4.4 depicts year-wise trend of publications of articles from all over the world on sickle cell anemia up to 1990. The first article appeared in 1944 but the sizable number of publications started since 1946. The number of publications increased steadily up to 1970 and then there was a sharp increase. In 1990 as many as 273 articles were published on this disorder from all over the world. This was due to the fact that sickle gene was identified in many countries in Africa, India and the Middle East. The presence of this gene in many communities or in tribal groups used to be reported regularly. Similarly in 1975 Southern blotting, an important technique in molecular biology was discovered. This was used to study various aspects related to this disorder. Similarly at the same time it was realized that clinical manifestations of the patients with this disorder was extremely variable in different communities

Figure 4.2: Articles published during 1991-2014 from India on six genetic disorders in various journals.



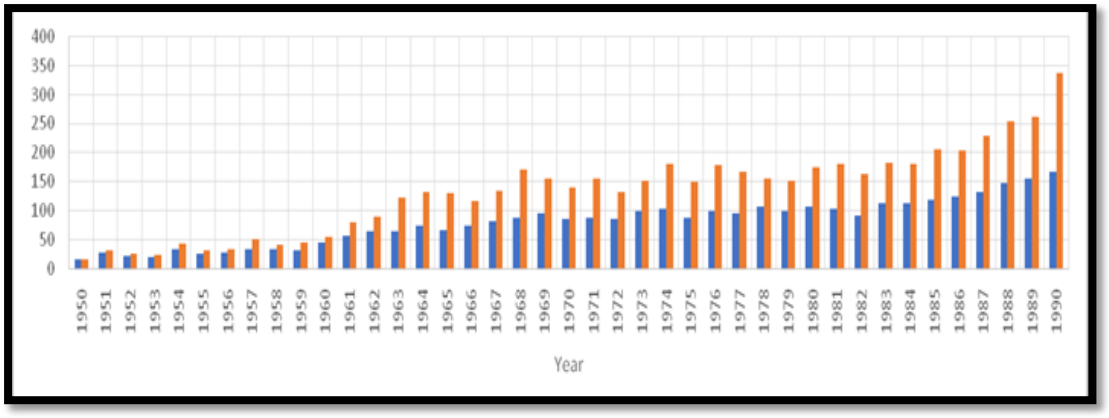
Observation- There is a considerable increase in number of articles published on von Willebrand disease, Hemophilia A and Hemophilia B from India during 1991-2014 as compared to those published up to 1990 (fig 4.1).

Table 4.4: List of top five journals where maximum numbers of articles from India are published on each genetic disorder during 1991 – 2014.

Genetic Disorder	Down syndrome	Sickle cell aanemia	Hemophilia A	Hemophilia B	von Willebrand disease	G6PD deficiency
Top Five Journals	Ind J Paed 26	JAPI 21	Haemophilia 63	Haemophillia 20	Ann Haemat 08	Ind J Paed 10
	Ind Paed 14	Ind J Paaed 13	Sem In Thrombo Hem 08	Am J Haemat 03	Haemophilia 06	BCMD 06
	IJHG 09	Haemoglobin 13	CATH 08	EuroJ Haemat 03	Natl Med J Ind 04	JAPI 05
	JAPI 06	IJMR 07	J Thrombo Hem 06	Blood Co F I 02	Sem In Thrombo Hem 04	Ind J Mal 04
	Paed Haemo Onco 05	Ind J Patho & M 06	Ind J Paed 06	J Thrombo Hem 02	J Thrombo Hem 03	Anthro ANZ 04
Total	**60**	**60**	**91**	**30**	**25**	**29**
Total No. Articles	**192**	**243**	**210**	**73**	**63**	**118**
No. of Independent Journals	**95**	**115**	**85**	**44**	**34**	**62**
% Representation of top 5 Journals	**5.26**	**4.35**	**5.88**	**11.36**	**14.71**	**8.06**
% Representation of Articles in top 5 Journals	**31.25**	**24.69**	**43.33**	**41.10**	**39.68**	**24.58**
% representation of Articles in first Journal	**13.54**	**8.64**	**30.00**	**27.4**	**12.7**	**8.48**
Articles/Journals	**2.02**	**2.11**	**2.47**	**1.66**	**1.85**	**1.90**

Sem In Thrombo Hem – Seminars in Thrombosis and Haemotostasis , Ind J Patho & M - Indian Journal of Pathology and Microbiology, Ind Paed – Indian Paedeatrics, BCMD – Blood Cells Molecules and Diseases J Thrombo Hem – Journal of Thrombosis and Haemostasis, Anthro ANZ- Anthropology ANZ, Ind J Mal- Indian Journal of Maleriology, Blood Co F I- Blood Coagulation and Fibrinolysis, Ind J Paed- Indian Journal of Paedeatrics, Am J Haemat – American Journal of Haematology, IJHG – Indian Journal of Human Genetics ,IJMR – Indian Journal of Medical Research,Paed Haemo Onco – Paediatrics Haematology and Oncology ,JAPI- Journal of Association of Physicians of India, CATH- Clinical Applications in Thrombosis and Haemostasis, Euro J Haemat – European Journal of Haematology,Ann Haemat – Annals of Haematology, Natl Med J Ind – National Medical Journal of India.

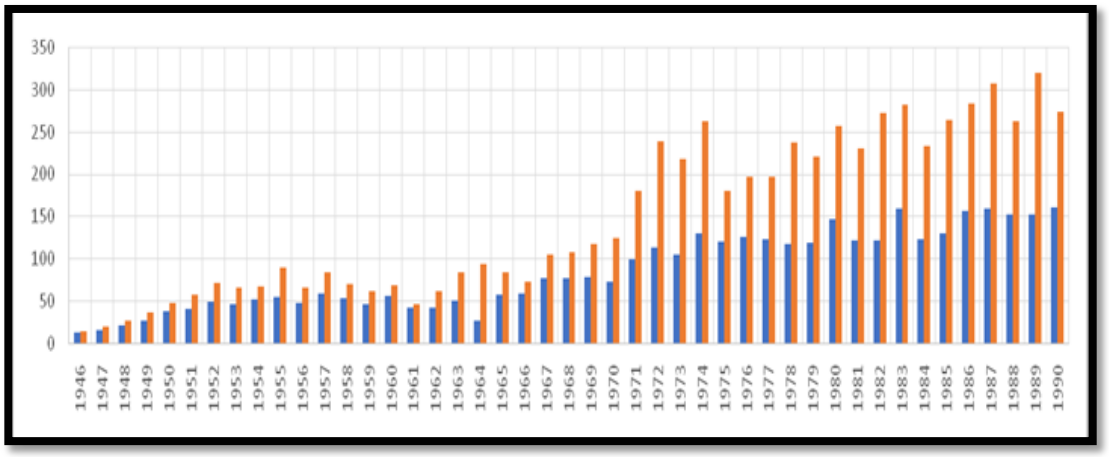
Figure 4.3: Year - wise trend of international publications up to 1990 on Down syndrome.



Observation- Publications were steadily increasing up to 1960 and since then considerable increase in number of publications is seen over the years up to 1990.

Most productive year of publications was 1990.

Figure 4.4: Year - wise trend of international publications up to 1990 on Sickle cell anemia.



Observation- Publications were steadily increasing up to 1970 and since there was a sharp increase in number of publications up to 1990.

Most productive year of publications was 1989.

living in different countries. These were some of the reasons for sharp increase in publications.

Year-wise trend of international publications on hemophilia A up to 1990 is shown in Figure 4.5. The first article appeared in 1944. It can be seen that the sizable number of publications started in 1946 and remained steady up to 1963 and then there was an increase in number of articles published up to 1990.

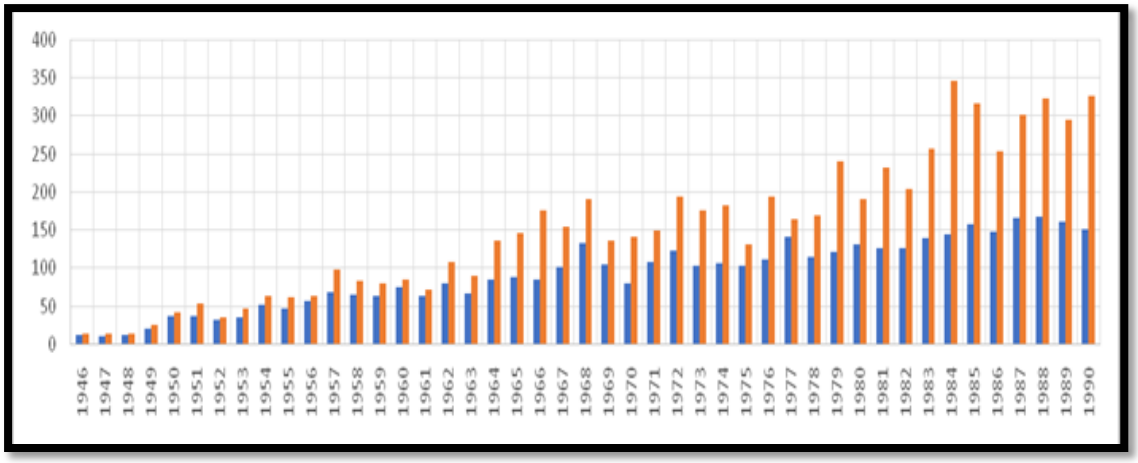
Year-wise trend of international publications on G6PD deficiency up to 1990 is depicted in Figure 4.6. The first article was published in 1947. The sizable number of articles started appearing since 1952 and the number of publications per year remained more or less same up to 1960. Then there was a sharp increase in number of publications for next four to seven years. However, then there was a gradual decrease in number of publications up to 1990. This is the only disorder where the number of publications has been decreased over the years. Overall the number of publications was relatively less as compared to those on sickle cell anemia or Down syndrome.

Figure 4.7 reveals year-wise trend of publications from all over the world up to 1990 on hemophilia B. The first article on this disorder appeared in 1946. However sizable number of publications were started since 1953 and then there was a steady increase in the number of publications up to 1982. During last eight years i.e. 1983-1990 there was a steep increase in number of publications. However the total number of publications (1,399) was considerably less as compared to other disorders.

Year-wise trend of international publications on von Willebrand disease up to 1990 is shown in Figure 4.8. The first publication on this disorder appeared in 1946 however sizable number of publications started after 12 years i.e. from 1958 onwards. It may be due to lack of awareness among scientists and clinicians about this disorder. The publications increased steadily up to 1972 and then there was a steep increase in publications over the years up to 1990. Again the total number of publications (1,579) was less as compared to other disorders.

Figure 4.9 revealed comparison of trends of total number of international publications in different journals on six genetic disorders up to 1990. Maximum numbers of articles were published on hemophilia A followed by those on sickle cell anemia and Down syndrome.

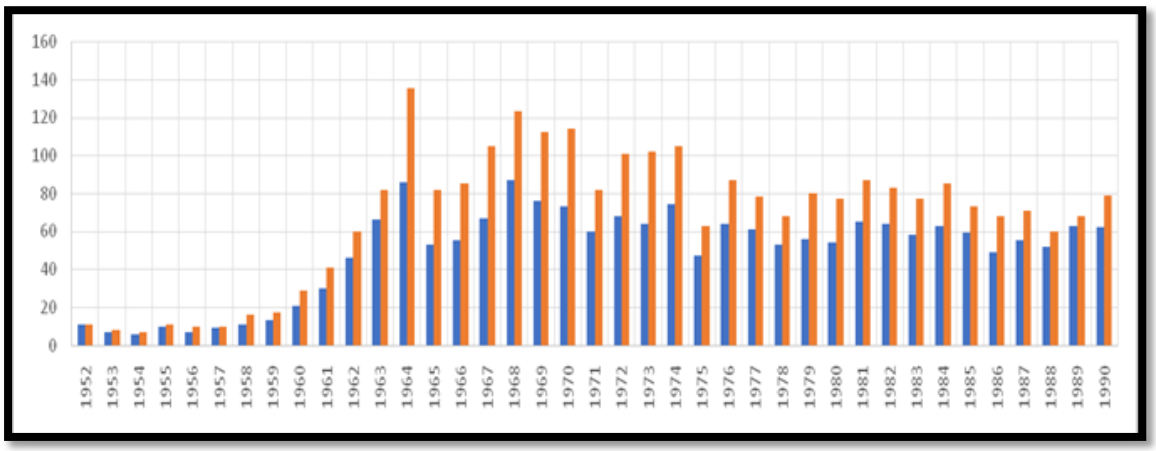
Figure 4.5: Year - wise trend of international publications up to 1990 on Hemophilia A.



Observation - Publications were steadily increasing up to 1963 and there was a considerable increase in number of articles up to 1990.

Most productive year of publications was 1984.

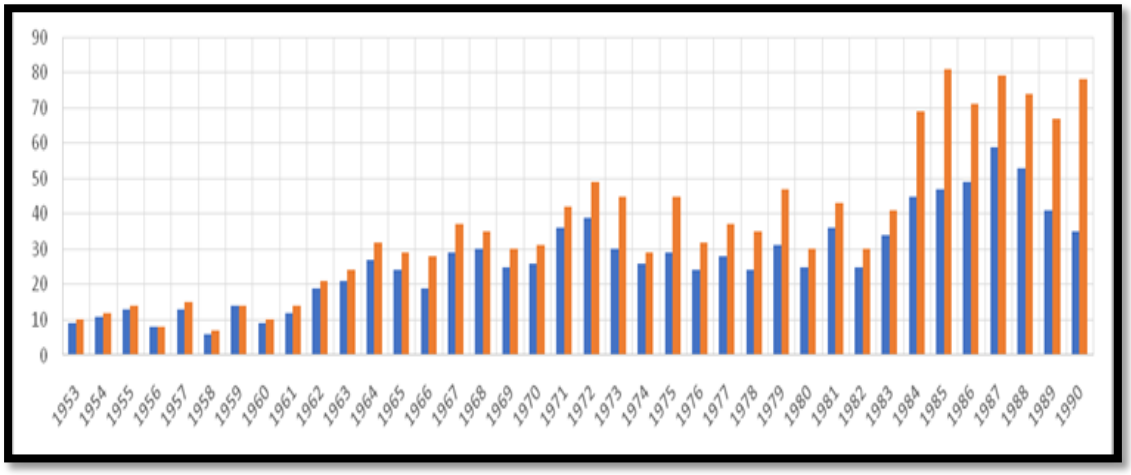
Figure 4.6: Year - wise trend of international publications up to 1990 on G6PD deficiency.



Observation- Publications remained more or less same up to 1960, a sharp increase in number of articles is seen in next four to seven years and then a gradual decrease is seen over the years.

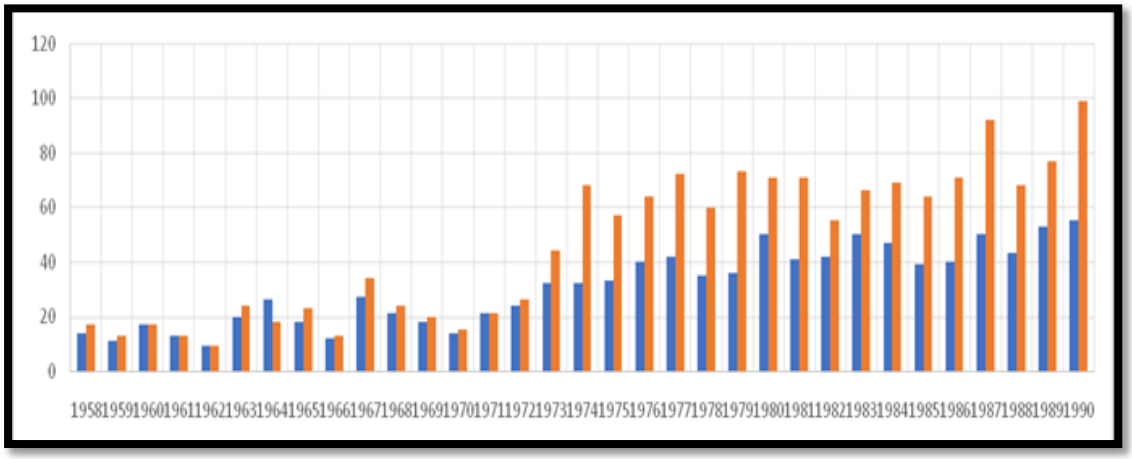
Most productive year of publications was 1964.

Figure 4.7: Year - wise trend of international publications up to 1990 on Hemophilia B.



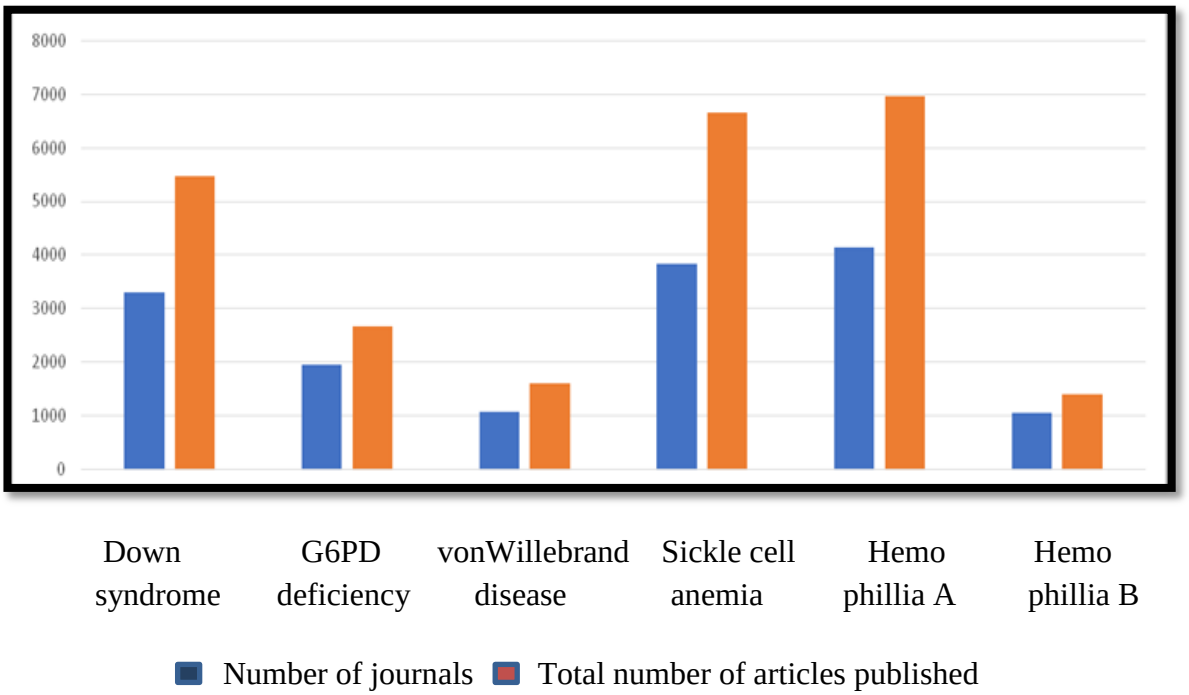
Observation- Steady increase in number of articles published is seen up to 1982 and then a sharp increase in number of publications up to 1990. Most productive year of publications was 1985.

Figure 4.8: Year - wise trend of international publications up to 1990 on von Willebrand disease.



Observation- Publications increased steadily up to 1972 and then there was a steep increase in number of publications over the years up to 1990. Most productive year of publications was 1990.

Figure 4.9: Total number of articles published from all over the world up to 1990 on six genetic disorders.



Observation- Articles published on G6PD deficiency, von Willebrand disease and hemophilia B were relatively less as compared to those published on remaining three disorders.

Table 4.5 reveals details about the publications on each disorder up to 1990. It lists the number of independent journals where articles on the six disorders were published. It also shows the list of 5 journals where maximum number of articles on each disorder was published. Number of independent journals publishing different articles on six disorders ranged between 420-2,361. These journals can be divided into two groups. In the first group, some journals publish good research work on various disorders or on various aspects related to biology. This group includes journals like 'Blood', 'Nature', 'Science' etc. In the second group, there are journals dedicated to particular disorder or topic. So they preferably accept articles related to that topic. With this background, if we observe Table 4.5, then it can be seen that 'Lancet' and 'Blood' are there in top five journals in five out of six disorders. On the other hand, 'Journal of Mental Deficiency Research' or 'American Journal of Mental Deficiency ' published articles on Down syndrome. Journals like 'Thrombosis and Hemostasis' or 'Thrombosis Research' are dedicated for publishing research on coagulation disorders. Therefore articles on hemophilia A, von Willebrand disease and hemophilia B were published in these journals. In the case of von Willebrand disease about $1/4^{th}$ of the total articles were published in top five journals while in the case of rest of the disorders this percentage ranged between 10.34-17.54%.

4.1.1.2.2 During 1991-2014

Figure 4.10 depicts year-wise trend of international publications during 1991-2014 on Down syndrome. There was a gradual increase in number of journals where the articles were published over the years. More or less similar trend was seen in the case of number of articles published on this disorder over the years.

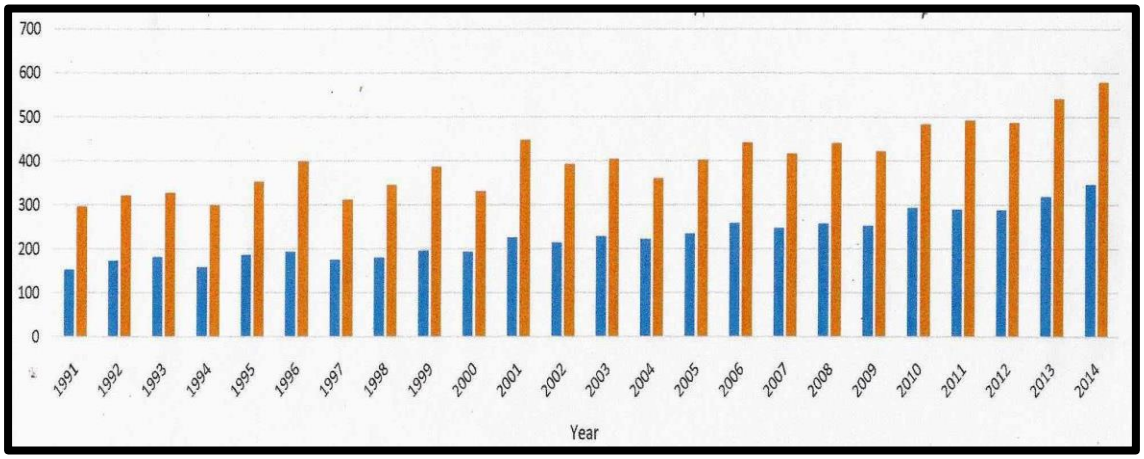
Year-wise trend of international publications on sickle cell anemia during 1991-2014 is shown in Figure 4.11. Here also increasing trend is seen. About 200-400 articles were published every year during 1991-2007; while from 2008 onwards there was a spurt seen in number of articles published every year. As many as 631 articles were published during 2014.

Table 4.5: List of top five journals where maximum numbers of articles from all over the world are published on each disorder up to 1990.

Genetic Disorder	Down syndrome	Sickle cell anemia	Hemophilia A	Hemophilia B	von Willebrand disease	G6PD deficiency
Top Five Journals	Lancet 269	Blood 241	Lancet 293	Blood 60	Blood 106	Lancet 78
	J Men Def Res 249	N Eng J Med 159	Blood 193	Br J Hemat 54	Thrombo Res 87	Blood 53
	Hum Gen 106	Lancet 154	Haemat 169	Throm Haemost 41	Br J Hemat 77	Acta Haemat 51
	Am J Men Def 236	J Paed 143	Throm Haemost 127	Lancet 39	Throm Haemost 63	HumGen 49
	J Paed 97	JAMA 127	N Eng J Med 114	Thrombo Res 29	J Clin Invest 49	AJHG 45
Total	**957**	**824**	**896**	**223**	**382**	**276**
Total No. Articles	**5,455**	**6,649**	**6,962**	**1,399**	**1,579**	**2,666**
No. of Independent Journals	**1,196**	**1,123**	**1,361**	**467**	**420**	**671**
% Representation of top 5 Journals	**0.45**	**0.45**	**0.37**	**1.07**	**1.19**	**0.75**
% Representation of Articles in top 5 Journals	**17.54**	**12.39**	**12.87**	**15.94**	**24.19**	**10.35**
% representation of Articles in first Journal	**4.93**	**3.62**	**4.21**	**4.29**	**6.71**	**2.93**
Articles/Journals	**4.56**	**5.92**	**5.12**	**3**	**3.76**	**3.97**

J Men Def Res – Journal of Mental Defect Research, J.Paed - Journal of Pediatrics , Throm Haemost- Throbosis and Haemostasis, Hum Gen - Human Genetics, JAMA- Journal of American Medical Association, Br J Hemat- British Journal of Haematology, Am J Men Def – American Journal of Mental Defects, Haemat- Haematology , Acta Haemat- Acta Haematologica, Thrombo Res – Thrombosis Research, J Clin Invest – Journal of Clinical Investigation, AJHG- American Journal of Human Genetics, N Eng J Med – New England Journal of Medicine.

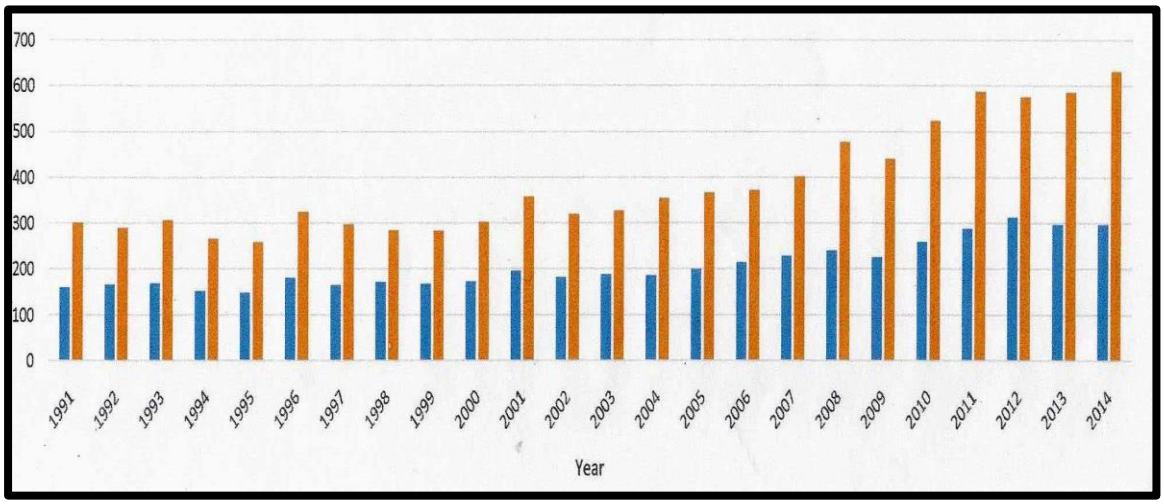
Figure 4.10: Year - wise trend of international publications during 1991-2014 on Down syndrome.



Observation- Steady increase in number of publications is seen over the years.

Most productive year of publications was 2014.

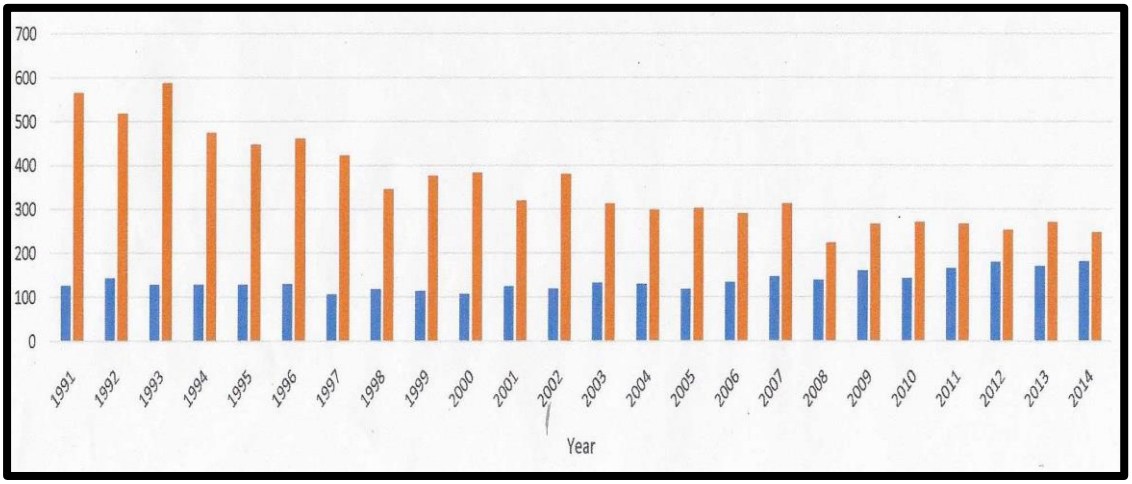
Figure 4.11: Year - wise trend of international publications during 1991-2014 on Sickle cell anemia.



Observation- Steady increase in number of publications is seen over the years.

Most productive year of publications was 2014.

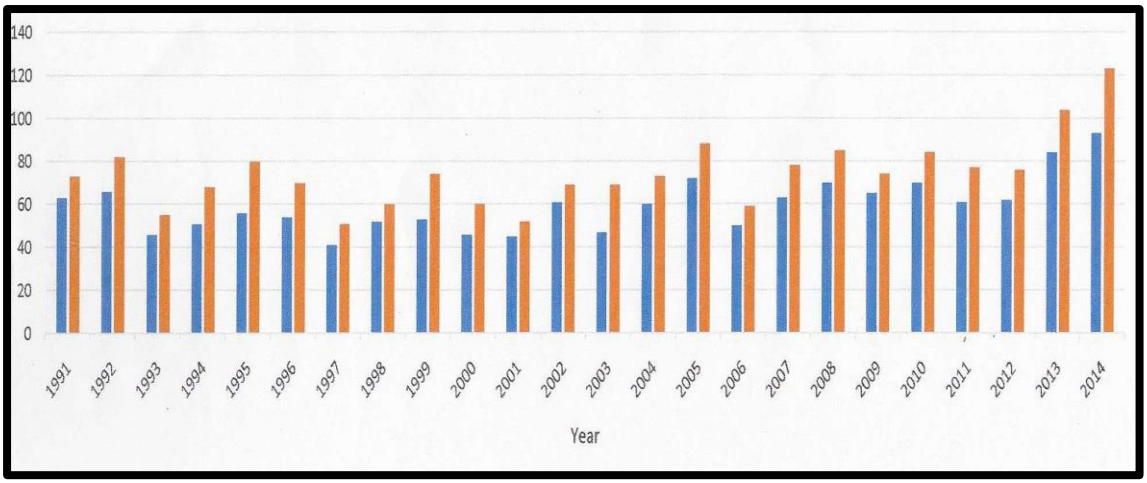
Figure 4.12: Year - wise trend of international publications during 1991-2014 on Hemophilia A .



Observation- Steady decrease in number of publications is seen over the years.

Most productive year of publications was 1993.

Figure 4.13: Year - wise trend of international publications during 1991-2014 on G6PD deficiency.



Observation- No particular pattern is observed in the number of publications.

Most productive year of publications was 1993.

Figure 4.12 reveals year-wise trend of international publications on hemophilia A during 1991-2014. As far as journals are concerned a slow but steady increase in the numbers is seen over the years. However in the case of number of articles published every year, the trend is reversed. Publications of number of articles were steadily decreasing.

Year-wise trend of articles published from all over the world on G6PD deficiency during 1991-2014 is represented in Figure 4.13. The number of journals did not show any particular pattern. In the case of number of articles, the years 2013 and 2014 showed a sudden spike in the number of articles published as compared to those during earlier years. 104 and 123 articles were published during these two years respectively.

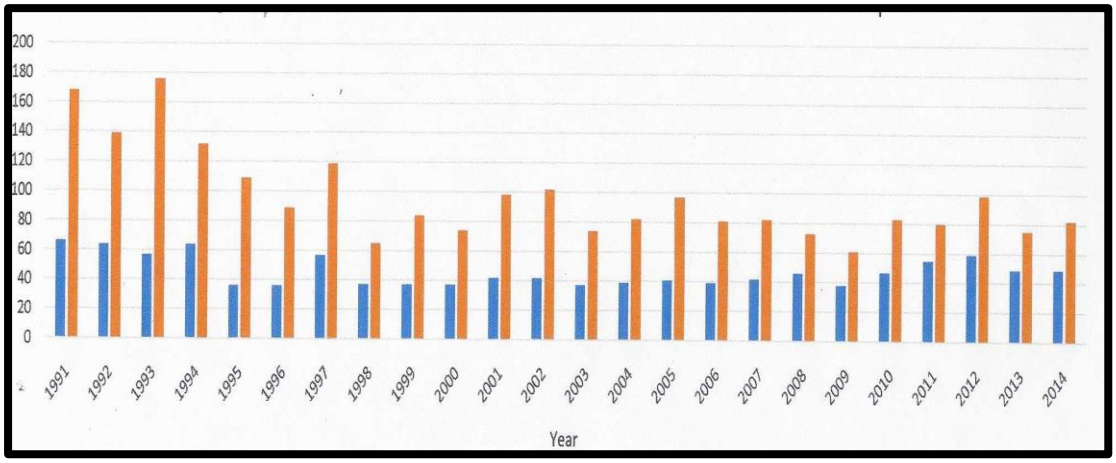
Figure 4.14 presents year-wise trend of publications of articles from all over the world on hemophilia B during 1991-2014. The number of journals where the articles were published does not show any particular trend. However in the case of number of articles published, there was a decreasing trend over the years. The number of articles published during 1991-1997 was in the range of 89-168 while afterwards the number decreased considerably (range 61-102).

The results of year-wise trend of articles published from all over the world on von Willebrand disease during 1991-2014 are shown in Figure 4.15. An increasing trend in the case of number of journals as well as number of articles published over the years has been observed.

Figure 4.16 depicts total number of articles published from all over the world on six different disorders during 1991-2014 in various journals. Maximum articles were published on Down syndrome followed by those on sickle cell anemia and hemophilia A.

Table 4.6 reveals the list of top 5 journals where maximum number of articles on each disorder were published. In the case of hemophilia B and hemophilia A percent representation of articles in top 5 journals was 42.69% and 40.47% respectively, while in the remaining disorders it ranged between 4.54%-24.68%. It can be seen that majority of the articles (4,682 out of 5,582 i.e. 83.88%) On hemophilia A, hemophilia B and von Willebrand disease were published in subject specific journals like ‘Hemophilia’ ‘Thrombosis and Hemostasis’, ‘Journal of Thrombosis and Hemostasis’ and ‘Seminars in Thrombosis and Hemostasis’. In the case of Down syndrome maximum numbers of articles were published in the journal

Figure 4.14: Year - wise trend of international publications during 1991-2014 on Hemophilia B.

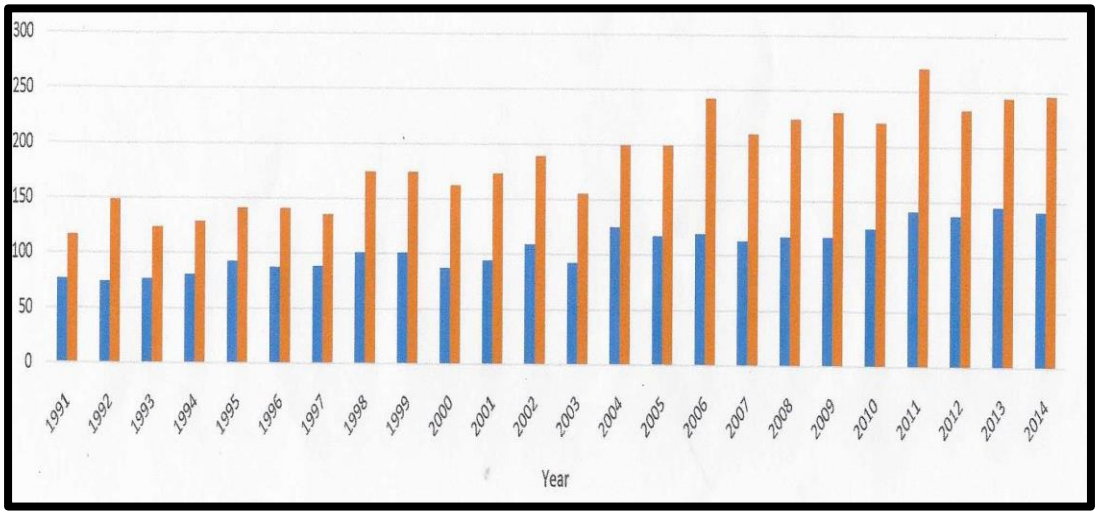


Number of journals Total number of articles published

Observation- Steady decrease in number of publications is seen over the years.

Most productive year of publications was 1993.

Figure 4.15: Year - wise trend of international publications during 1991-2014 on von Willebrand disease.

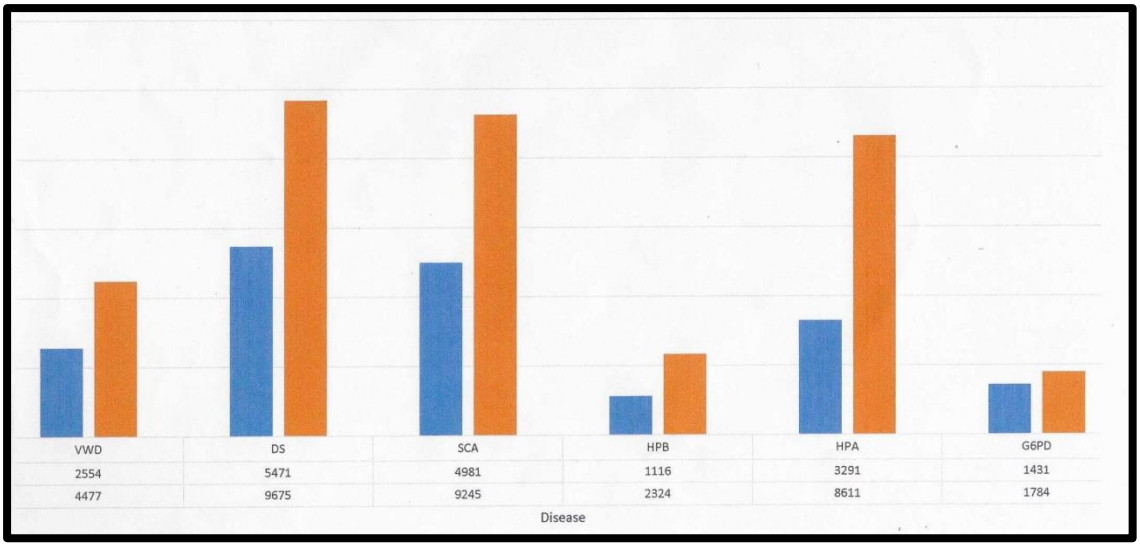


Number of journals Total number of articles published

Observation- Increasing trend in number of publications is seen over the years.

Most productive year of publications was 2011.

Figure 4.16: Total number of international publications during 1991-2014 on six genetic disorders.



Number of journals (First row) Total number of articles published (Second row)

Observation- Articles published on hemophilia B and G6PD deficiency were relatively less as compared to those published on four genetic disorders.

Table 4.6: List of top five journals where maximum numbers of articles from all over the world are published on each genetic disorder during 1991 – 2014.

Genetic Disorder	Down syndrome	Sickle cell anemia	Hemophilia A	Hemophilia B	von Willebrand disease	G6PD deficiency
Top Five Journals	PD 645	Am J Haemat 437	Hemophillia 2225	Hemophillia 634	Hemophillia 338	Blood 19
	JIDR 210	Blood 407	Throm Haemo 365	Throm Haemo 110	Throm Haem 287	Am J Haemat 17
	Ultra Obs Gyc 183	BJH 299	J Throm hem 351	Blood 97	J Throm Hem 190	BCMD 17
	AJOG 152	Paed Blood Can 225	Blood 297	B J H 84	Blood 175	B J H 15
	RDD 150	J Paed H Onco 188	B J H 247	J Throm Haemo 67	Sem In Thrombo Hem 115	Paediatrics 13
Total	**1,340**	**1,556**	**3,485**	**992**	**1,105**	**81**
Total No. Articles	**9,675**	**9,245**	**6,962**	**8,611**	**1,579**	**2,666**
No. of Independent Journals	**1,196**	**1,123**	**1,361**	**537**	**1,117**	**689**
% Representation of top 5 Journals	**0.27**	**0.2**	**0.38**	**0.93**	**0.45**	**0.73**
% Representation of Articles in top 5 Journals	**13.85**	**16.83**	**40.47**	**42.69**	**24.68**	**4.54**
% representation of Articles in first Journal	**6.67**	**4.73**	**25.84**	**27.28**	**7.55**	**1.07**
Articles/Journals	**5.29**	**5.55**	**6.50**	**4.33**	**4.01**	**2.59**

Sem In Thrombo Hem – Seminars in Thrombosis and Haemostasis, BCMD – Blood Cells Molecules and Diseases, J Thrombo Hem – Journal of Thrombosis and Haemostasis, Am J Haemat – American Journal of Haematology, PD- Prenatal Diagnosis, Ultra Obs Gyc – Ultra sound Obstetric Gynecology, JIDR- Journal of Intellectual Disability Research, BJH- British Journal of Haemotolgy ,RDD – Research in Developmental Disability, AJOG – American Journal of Obstetrics and Gynecology, Paed Blood Can- Paediatric Blood Cancer, J Paed H Onco- Journal of Paediatric Haemotolgy Oncology, Throm Haemo- Thrombosis and Haemostasis

Table 4.7: Year - wise and category - wise analysis of articles published on Hemophilia B from India during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	1	0	0	0	0	0	1
1992	0	0	0	0	0	0	0
1993	1	0	0	0	0	0	2
1994	0	0	0	0	0	0	0
1995	0	0	0	0	0	0	0
1996	0	0	0	0	0	0	1
1997	0	0	1	0	0	1	2
1998	0	0	1	0	0	1	2
1999	0	0	0	0	0	0	0
2000	0	0	0	2	0	0	2
2001	2	2	2	0	0	2	8
2002	1	0	0	0	1	0	2
2003	1	0	0	0	0	3	4
2004	1	1	0	1	0	3	6
2005	0	0	0	0	0	1	1
2006	0	0	0	0	0	2	2
2007	1	2	1	0	0	2	6
2008	1	0	0	0	0	5	6
2009	0	1	0	0	0	4	5
2010	1	0	1	0	1	0	3
2011	0	0	3	1	0	2	6
2012	2	0	2	1	0	1	6
2013	1	1	0	0	0	3	5
2014	0	0	2	0	0	1	3
Total	**13 (17.80)**	**8 (10.96)**	**14 (19.18)**	**5 (6.85)**	**2 (2.74)**	**31 (42.4)**	**73**

Figures in brackets indicate percentage values.

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Half-life of the articles published in this series is 17 years since 1991.

'Prenatal Diagnosis' while 'American Journal of Hematology' published maximum articles on sickle cell anemia.

4.1.2 Category-wise trend of publications of articles

4.1.2.1 India (During 1991-2014)

Table 4.7 reveals category-wise and year-wise trend of publications of articles on hemophilia B from India during 1991-2014. Only 73 articles have been published on this disorder during 24 years with an average of 2-3 articles per year. As mentioned earlier incidence of hemophilia A in India is about 1 in 5000 individuals while that of hemophilia B is about 1 in 20000 individuals. Because of this rarity, less number of articles has been published on this disorder. Due to small sample size, percent values were calculated only for total number of articles. 42.4% of the articles were published using molecular biology tools. Half-life of the articles in this series was 17 years since 1991.

Category-wise and year-wise analysis of articles published on G6PD deficiency from India during 1991-2014 is shown in Table 4.8. Totally 118 articles have been published during 24 years with an average of 4-5 articles per year. 54.48% of the total articles were published as 'original articles' while 20.34% of the total articles were published on molecular biology related data. Half-life of the articles in this series was 14 years since 1991.

Table 4.9 depicts year-wise and category-wise analysis of the articles published from India on sickle cell anemia during 1991-2014. This is a very common disorder among the various tribal groups from our country. So various scientists and institutes working on different tribal groups have been working on this disorder. Therefore maximum number of articles (243) has been published on this disorder during 24 years with an average of 9-10 articles per year. 36.63% of the total articles were published as 'original articles' while 21.81% of the total articles were published using data related to various molecular biology aspects. Half-life of the articles in this series was 18 years since 1991.

Category-wise and year-wise analysis of the articles published on hemophilia A from India during 1991-2014 is presented in Table 4.10. Totally 210 articles have been published on this disorder during 24 years with an average of 8-9 articles per year. 28.57% of the total articles were published using molecular biology tools while

21.43% of the total articles were published as 'original articles'. Half-life of the articles published in this series was 15 years since 1991.

Table 4.11 reveals category-wise and year-wise analysis of the articles published on Down syndrome from India during 1991-2014. Totally 192 articles have been published during 24 years with an average of 7-8 articles per year. Year-wise publication figures reveal that during 1991-2006 the range of publication of articles was 1 to 8 articles per year while from 2007 onwards the speed of publication was picked up considerably as more than 10 articles were published every year. 32.29% of the total articles were published as 'original articles'. Significant number of articles (25%) were published describing various clinical findings seen in this disorder while 18.23% of the articles were published using data related to various aspects of molecular biology. Half-life the articles published on this disorder was 19 years since 1991.

Table 4.12 presents year-wise and category-wise analysis of the articles published from India on von willebrand disease during 1991-2014. Totally 62 articles have been published during 24 years with an average of 2-3 publications per year. In fact this is one of the commonest bleeding disorders in India. In spite of this very small number of articles are published on this disorder from India. Because based on the clinical presentation this disorder is divided into three categories: mild, moderate and severe. Of these, mild and moderate cannot be diagnosed. So whatever articles are published, they are on severe cases of vWD. In addition, very few institutes in the country are working on this disorder. 37.1% of the total articles were published using data involving molecular biology aspects. Almost equal number of articles (38.71%) were published as 'original articles'. Half-life of the articles published in this series was 18 years since 1991.

Table 4.13 depicts category-wise analysis of articles published on six genetic disorders from India during 1991-2014. Over all about $1/4^{th}$ of the total articles (25.25%) have been published using data involving molecular biology aspects (Category 6). The range of articles published under this category varies between 18.23% (Down syndrome) to 42.4% (hemophilia B). Over all 33.63% of the total articles have been published as 'original articles' (Category 1). The range of articles published under this category varies between 17.8% (Hemophilia B) to 54.48% (G6PD deficiency). These articles were divided into two groups with two different time intervals i.e. publications during 1991-2002 and publications during 2003-2014

Table 4.8: Year - wise and category - wise analysis of articles published on G6PD deficiency from India during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	3	0	0	0	0	0	3
1992	9	0	0	0	2	2	11
1993	4	0	1	0	0	0	5
1994	4	0	0	0	0	2	6
1995	4	0	0	0	0	1	5
1996	0	0	0	1	0	0	1
1997	5	0	0	1	0	0	6
1998	0	0	0	0	0	0	0
1999	1	0	0	0	0	1	2
2000	0	0	1	0	0	0	1
2001	4	0	0	0	0	2	6
2002	1	0	2	1	0	1	5
2003	4	0	1	0	0	2	7
2004	2	0	0	0	0	1	3
2005	5	0	0	0	0	1	6
2006	3	0	1	0	0	0	4
2007	4	0	3	0	0	1	8
2008	1	1	0	1	0	4	7
2009	3	0	1	1	0	2	7
2010	5	0	1	0	1	2	9
2011	2	0	1	0	1	1	5
2012	3	0	2	0	0	0	5
2013	1	0	1	0	0	0	2
2014	1	0	1	1	0	1	4
Total:	**69 (54.48)**	**1 (0.85)**	**16 (13.56)**	**6 (5.08)**	**2 (1.69)**	**24 (20.34)**	**118**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools.

Half-life of the articles published in this series is 14 years since 1991.

Table 4.9: Year - wise and category - wise analysis of articles published on Sickle cell anemia from India during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	1	1	0	2	0	2	6
1992	4	0	0	0	0	1	5
1993	4	2	0	2	0	1	9
1994	2	1	0	2	0	3	8
1995	1	3	0	0	0	1	5
1996	0	0	2	2	0	2	6
1997	3	4	0	0	0	3	10
1998	3	0	0	0	0	0	3
1999	1	1	0	2	0	1	5
2000	3	3	1	0	0	4	11
2001	2	1	2	0	0	0	5
2002	3	2	0	1	0	0	6
2003	3	2	0	0	0	0	5
2004	4	1	1	0	0	1	7
2005	5	3	1	1	1	1	12
2006	4	0	2	0	1	1	8
2007	5	3	1	0	0	0	9
2008	3	2	2	0	0	0	7
2009	4	0	2	0	1	1	8
2010	1	2	4	1	1	4	13
2011	4	0	4	1	2	3	14
2012	11	5	2	2	0	10	30
2013	8	4	2	3	1	7	25
2014	10	2	2	4	1	6	26
Total:	**89 (36.63)**	**42 (17.28)**	**28 (11.52)**	**23 (9.47)**	**8 (3.29)**	**53 (21.81)**	**243**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Half-life of the articles published in this series is 18 years since 1991.

Table 4.10: Year - wise and category - wise analysis of articles published on Hemophilia A from India during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	2	0	0	0	0	0	2
1992	0	1	0	0	0	0	1
1993	1	1	1	0	1	0	4
1994	0	0	0	0	0	0	0
1995	0	0	1	0	0	0	1
1996	0	1	0	1	0	0	2
1997	0	0	0	1	0	1	2
1998	1	4	1	1	0	3	9
1999	3	0	3	0	1	1	8
2000	3	1	2	1	0	2	9
2001	3	6	3	0	1	2	13
2002	2	2	2	1	0	3	10
2003	2	3	3	3	1	5	17
2004	2	5	3	1	0	5	14
2005	4	3	0	1	0	4	12
2006	3	0	1	0	1	3	8
2007	4	6	2	0	1	4	15
2008	2	2	0	0	3	3	10
2009	1	1	3	2	0	5	12
2010	1	2	2	0	1	2	8
2011	3	2	2	0	1	4	12
2012	4	2	1	1	1	5	12
2013	3	1	2	1	0	5	12
2014	3	3	4	1	3	3	17
Total:	**45 (21.43)**	**39 (18.57)**	**36 (17.14)**	**15 (7.14)**	**15 (7.14)**	**60 (28.57)**	**210**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools.

Half-life of the articles published in this series is 15 years since 1991.

Table 4.11: Year - wise and category - wise analysis of articles published on Down syndrome from India during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	3	0	0	0	1	0	3
1992	1	0	1	0	0	0	2
1993	1	0	1	0	0	0	2
1994	0	0	2	0	0	0	2
1995	0	0	1	0	0	0	1
1996	3	0	1	0	0	0	4
1997	4	0	1	1	0	1	7
1998	1	0	3	0	1	1	6
1999	1	0	0	0	0	0	1
2000	1	0	0	1	0	0	2
2001	3	0	0	1	0	0	4
2002	3	0	0	3	0	1	7
2003	3	1	0	2	0	2	8
2004	1	1	1	0	0	0	3
2005	2	1	2	1	1	1	8
2006	2	0	2	0	0	1	5
2007	3	1	2	0	3	3	12
2008	1	2	4	1	1	2	11
2009	1	4	4	0	1	3	12
2010	6	1	5	0	1	3	16
2011	5	2	4	1	0	1	13
2012	5	0	6	0	1	6	18
2013	7	1	8	1	2	4	23
2014	5	6	0	3	1	7	22
Total:	**62 (32.29)**	**20 (10.42)**	**48 (25.00)**	**15 (7.81)**	**12 (6.25)**	**35 (18.23)**	**192**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools.

Half-life of the articles published in this series is 19 years since 1991.

Table 4.12: Year - wise and category - wise analysis of articles published on von Willibrand disease from India during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	0	0	0	0	0	0	0
1992	0	0	0	0	0	0	0
1993	1	0	1	0	0	0	2
1994	1	0	0	0	0	0	1
1995	1	0	0	0	0	0	1
1996	0	0	0	0	0	0	0
1997	0	0	0	0	0	0	0
1998	0	0	0	0	0	0	0
1999	0	0	0	0	0	0	0
2000	0	1	1	0	0	1	3
2001	0	0	0	0	0	0	0
2002	0	0	0	0	0	0	0
2003	3	0	1	0	0	1	5
2004	2	0	0	0	0	1	3
2005	2	1	0	2	0	3	8
2006	2	1	1	0	0	1	5
2007	1	0	0	0	0	1	2
2008	0	1	0	0	0	1	2
2009	1	0	0	0	0	2	3
2010	0	0	0	0	1	2	3
2011	4	0	0	1	0	0	5
2012	3	0	0	1	0	0	4
2013	1	0	0	0	0	6	7
2014	2	1	1	0	0	4	8
Total:	**24 (38.71)**	**5 (8.06)**	**5 (8.06)**	**4 (6.45)**	**1 (1.61)**	**23 (37.10)**	**62**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools.

Half-life of the articles published in this series is 18 years since 1991.

Table 4.13: Category - wise analysis of articles published on six genetic disorders from India during 1991-2014.

Genetic Disease	1	2	3	4	5	6	Total	Half life Of Publ. (years)
Sickle cell anemia	89 (36.63)	42 (17.28)	28 (11.52)	23 (9.47)	8 (3.29)	53 (21.81)	243	18
Hemophilia A	45 (21.43)	39 (18.57)	36 (17.14)	15 (7.14)	15 (7.14)	60 (28.57)	210	15
Hemophilia B	13 (17.80)	8 (10.96)	14 (19.18)	5 (6.85)	2 (2.74)	31 (42.40)	73	17
vonWillebrand disease	24 (38.71)	5 (8.06)	5 (8.06)	4 (6.45)	1 (1.61)	24 (38.10)	63	18
G6PD deficiency	69 (54.48)	1 (0.85)	16 (13.56)	6 (5.08)	2 (1.69)	24 (20.34)	118	14
Down syndrome	62 (32.29)	20 (10.42)	48 (25.00)	15 (7.81)	12 (6.25)	35 (18.23)	192	19
Total	**302 (33.63)**	**115 (12.81)**	**147 (16.37)**	**68 (7.57)**	**40 (4.45)**	**227 (25.25)**	**899**	

Figures in brackets indicate percentage values

1. Research / Original articles
2 Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

to see whether any statistical difference is there in publications of articles under different categories during two time intervals or not?

Table 4.14 shows category-wise analysis of the articles published on six genetic disorders from India during 1991-2002. During this time period totally 259 articles have been published from India. Range of articles published under category 6 varies between 17.32% (Down syndrome) to 22.78% (sickle cell anemia). Over all 40.93% of the total articles were published as 'original articles'. More than half the number of articles (51.22%) on Down syndrome were published as 'original articles'.

Table 4.15 reveals category-wise analysis of the articles published on six genetic disorders from India during 2003-2014. During this time period totally 680 articles have been published from India. Under category 6 the increase in the number of articles was also four times more (180 articles published during 2003-2014 as compared to 47 articles published during 1991-2002). Significant increase in the number of articles published during second time interval was observed in the case of 'original articles' and 'case reports. In the case of von Willebrand disease only 7 articles were published during 1991-2002. Out of 56 total number of articles published on this disorder during 2003-2014, 41.07% of the total articles were published under category 6 .

X^2 test was used to compare the data on six genetic disorders during two time intervals. Where ever sample size was small (less the 4), Yate's correction was applied while calculating X^2 values. The values were considered for one degree of freedom and $p<0.05$ was considered as statistically significant value.

The results are tabulated in Table 4.16. Only in the case of two categories viz. original articles (category 1) of Down syndrome and review articles (Category 4) of sickle cell anemia, a statistically significant difference was observed while in rest of the categories non-significant results were seen.

Figure 4.17 reveals trend of articles published under category 6 from India during 1991-2014 on six genetic disorders. Due to small sample size, instead of percent values actual numbers are used to draw the graph. In the case of G6PD deficiency the peak was seen during the year 2008 and subsequently the number started decreasing while in the case of remaining disorders the number was increasing from 2010 onwards.

Table 4.14: Category - wise analysis of articles published on six genetic disorders from India during 1991-2002.

Genetic Disease	1	2	3	4	5	6	Total
Sickle cell anemia	27 (34.18)	18 (22.78)	5 (6.33)	11 (13.92)	0 (0.00)	18 (22.78)	79
Hemophilia A	15 (24.59)	13 (21.31)	13 (21.31)	5 (8.20)	3 (4.92)	12 (19.67)	61
Hemophilia B	5 (25.00)	3 (15.00)	5 (25.00)	2 (10.00)	1 (5.00)	4 (20.00)	20
von Willebrand disease	3	1	2	0	0	1	7
G6PD deficiency	35 (68.63)	0 (0.00)	4 (7.84)	3 (5.88)	0 (0.00)	9 (17.65)	51
Down syndrome	21 (51.22)	0 (0.00)	10 (24.39)	6 (14.63)	1 (2.44)	3 (17.32)	41
Total	**106 (40.93)**	**35 (13.5)**	**39 (15.06)**	**27 (10.42)**	**5 (1.93)**	**47 (18.14)**	**259**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Table 4.15 : Category - wise analysis of articles published on six genetic disorders from India during 2003-2014.

Genetic Disease	1	2	3	4	5	6	Total
Sickle cell anemia	62 (37.80)	24 (14.63)	23 (14.02)	12 (7.32)	8 (4.88)	35 (21.34)	164
Hemophilia A	30 (15.87)	26 (13.76)	23 (12.17)	10 (5.29)	12 (6.35)	48 (25.40)	189
Hemophilia B	8 (15.09)	5 (9.43)	9 (16.98)	3 (5.66)	1 (1.89)	27 (5.94)	53
vonWillebrand disease	21 (38.18)	4 (7.27)	3 (5.45)	4 (7.27)	1 (1.82)	23 (41.07)	56
G6PD deficiency	34 (50.74)	1 (1.49)	12 (17.91)	3 (4.48)	2 (2.99)	15 (22.39)	67
Down syndrome	41 (27.15)	20 (13.25)	38 (25.17)	9 (5.96)	11 (7.28)	32 (21.19)	151
Total	**196 (28.87)**	**80 (11.78)**	**108 (15.91)**	**41 (6.04)**	**35 (5.16)**	**180 (26.47)**	**680**

Figures in brackets indicate percentage values

1. Research / Original articles
2 Articles based on clinical findings
2. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Table 4.16: X^2 contingency table to compare articles published under different categories from India on Sickle cell anemia, Hemophilia B, von Willebrand disease, G6PD deficiency, Down syndrome and Hemophilia A during two time intervals i.e. 1991 to 2002 and 2003 to 2014.

Sickle cell anemia

X^2	p value	Significance
1 x 1 - 0.142	p = 0.7067	NS
2 x 2 - 1.708	p = 0.1913	NS
3 x 3 - 1.882*	p = 0.1701	NS
4 x 4 - 4.152*	p = 0.0416	Significant
5 x 5 - NA	-	-
6 x 6 - 0.042	p = 0.8383	NS

Hemophilia B

X^2	p value	Significance
1 x1 - 0.228*	p = 0.6327	NS
2 x 2 - 0.036*	p = 0.8504	NS
3 x 3 - 0.095*	p = 0.7584	NS
4 x 4 - 0.008*	p = 0.9265	NS
5 x 5 - 0.493*	p = 0.4825	NS
6 x 6 - 1.823*	p = 0.1769	NS

von Willebrand disease

X^2	p value	Significance
1 x1 - 0.025*	p = 0.8753	NS
2 x 2 - 0.334*	p = 0.5634	NS
3 x 3 - 1.275*	p = 0.2588	NS
4 x 4 - NA	-	-
5 x 5 - NA	-	-
6 x 6 - 0.309	p =0.5784	NS

G6PD deficiency

X^2	p value	Significance
1 x 1 - 0.987	p = 0.3204	NS
2 x 2 - NA	-	-
3 x 3 - 1.253*	p = 0.2629	NS
4 x 4 - 0.107*	p = 0.7349	NS
5 x 5 - NA	-	-
6 x 6 - 0.267	p = 0.6050	NS

Down syndrome

X^2	p value	Significance
1 x 1 - 3.979	p = 0.0461	Significant
2 x 2 - NA	-	-
3 x 3 - 0.006	p = 0.9371	NS
4 x 4 - 1.796*	p = 0.1802	NS
5 x 5 - 0.510*	p = 0.4751	NS
6 x 6 - 2.332*	p = 0.1268	NS

Hemophilia A

X^2	p value	Significance
1 x1 - 1.591	p = 0.2071	NS
2 x 2 - 1.413	p = 0.2345	NS
3 x 3 - 2.250	p = 0.1336	NS
4 x 4 - 0.221*	p = 0.6386	NS
5 x 5 - 0.006*	p = 0.9393	NS
6 x 6 - 0.520	p = 0.4707	NS

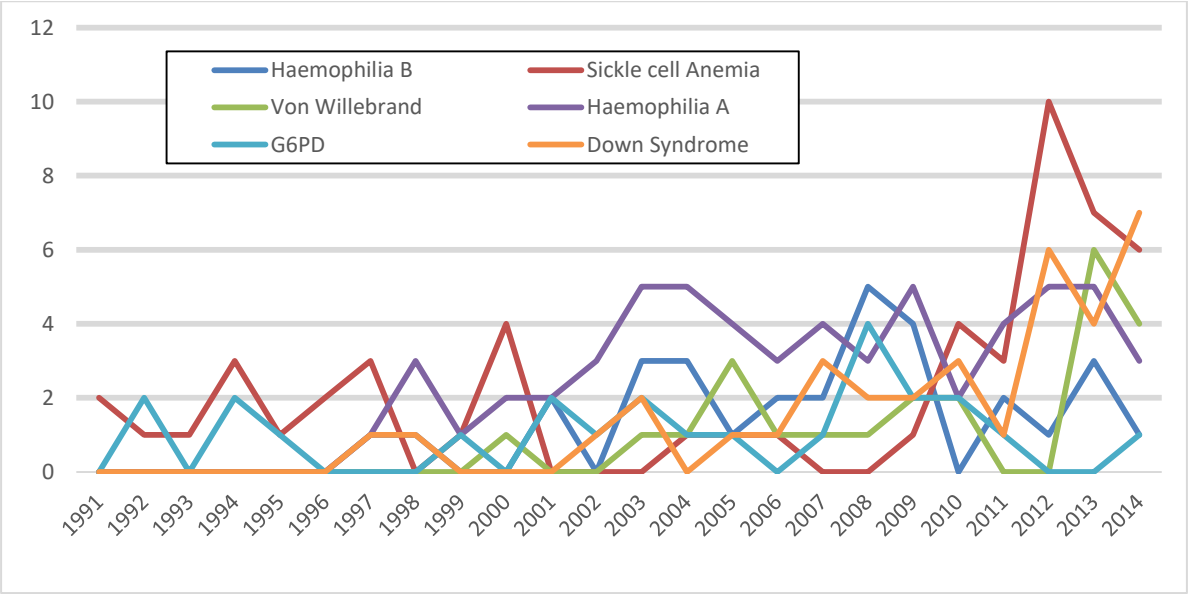
NS = Not Significant * = With Yate's correction NA = Not Applicable

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

X^2 test done for 1 degree of freedom.

X^2 test results are considered statistically significant when p value is < 0.05.

Figure 4.17: Trend of the articles published from India under category 6 (using molecular biology tools) on six genetic disorders (Viz. Down syndrome, Hemophilia A, G6PD deficiency, Hemophilia B, von Willebrand disease and Sickle cell anemia during 1991-2014 .



X axis – Years

Y axis – Number of articles published under category 6

Due to small sample size values are not expressed in percentages

Observation- Number of publications increased steadily showing maximum publications during 2011-2013.

4.1.2.2 All over the world (During 1991-2014).

Table 4.17 depicts category-wise and year-wise analysis of the articles published on von-Willebrand disease from all over the world during 1991-2014. Totally 4,477 articles have been published on this disorder on the above stated period. Out of the total number of articles published more than 50% (52.07%) of the articles have been published as 'original articles'. The last column which represents articles published using molecular biology tools reveals that about 20% of the total number of articles published every year was from this category (range 13.33-29.32%). Half-life of the articles published in this series is 15 years since 1991.

Table 4.18 reveals category-wise and year-wise analysis of the articles published on sickle cell anemia from all over the world during 1991-2014. Totally 9,245 articles have been published on this disorder during above stated time period. During the first 13 years i.e. up to 2003, total number of articles published every year was in the range of 259-327.

This number was increased considerably later on. In the next 11 years it was in the range of 353-629 articles per year. It has been observed that the original articles published on sickle cell anemia were in the range of 33.55-47.68% except during following four years (viz. 1992, 1993, 1994, and 2007) where more than 50% of the total number of articles were published under this category (range 52.32-68.63%)About $1/5^{th}$ of the articles published every year were those using molecular biology tools. During first four years (i.e. 1991-1995) the range was 11.31-14.29% while in the subsequent years the range increased gradually to 20-22% with an average of 18.23%. Half-life of the articles published in this series is 15 years since 1991 which shows that 50% of the total number of articles published in this series were during last nine years (i.e.2006-2014) .

Year-wise and category-wise analysis of the articles published on Down syndrome from all over the world during 1991-2014 is presented in Table 4.19. Totally 9,675 articles on this disorder have been published during above stated time period. During first ten years number of articles published every year were in the range of 285-367 while during remaining years there was a considerable increase in the total number of articles published every year (range359-579). It has been observed that during first ten years, about 50% of the total number of articles published every year was in the category of 'original articles' (range 46.67-56.88%) however in the next 14 years this percentage decreased considerably (range 26.29-39.55%).

Table 4.17: Category - wise analysis of articles published on von Willebrand disease from all over the world during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	53 (39.85)	18 (13.53)	7 (5.26)	16 (12.03)	0 (0.00)	39 (29.32)	133
1992	73 (46.20)	12 (7.60)	12 (7.60)	18 (11.39)	3 (1.90)	40 (25.31)	158
1993	68 (49.64)	3 (2.19)	19 (13.87)	17 (12.41)	0 (0.00)	30 (21.90)	137
1994	82 (60.74)	3 (2.22)	8 (5.93)	17 (12.59)	1 (0.74)	24 (17.78)	135
1995	81 (55.87)	6 (4.14)	18 (12.41)	7 (4.83)	1 (0.69)	32 (22.07)	145
1996	89 (58.55)	4 (2.63)	13 (8.55)	11 (7.24)	2 (1.32)	33 (21.71)	152
1997	80 (55.56)	14 (9.72)	12 (8.33)	14 (9.72)	2 (1.39)	22 (15.28)	144
1998	74 (45.40)	10 (6.13)	19 (11.66)	24 (14.72)	2 (1.27)	34 (20.86)	163
1999	106 (61.98)	7 (4.09)	11 (6.43)	15 (8.77)	3 (1.75)	29 (16.96)	171
2000	89 (51.74	12 (6.98)	12 (6.98)	18 (10.47)	1 (0.58)	40 (23.26)	172
2001	90 (52.63)	8 (4.68)	13 (7.60)	29 (16.96)	0 (0.00)	31 (18.12)	171
2002	109 (57.07)	16 (8.38)	13 (6.81)	24 (12.57)	1 (0.52)	28 (14.60)	191
2003	95 (60.90)	5 (3.21)	17 (10.90)	15 (9.62)	0 (0.00)	24 (15.38)	156
2004	94 (48.46)	21 (10.82)	18 (9.28)	15 (7.73)	2 (1.03)	44 (22.68)	194
2005	115 (58.38)	21 (10.66)	11 (5.58)	14 (7.11)	4 (2.03)	32 (16.24)	197

2006	124 (52.77)	17 (7.23)	20 (8.51)	33 (14.04)	4 (1.70)	37 (15.74)	235
2007	103 (51.24)	14 (6.97)	20 (9.95)	27 (13.43)	5 (2.49)	32 (15.92)	201
2008	127 (56.44)	9 (1.78)	15 (6.67)	38 (16.89)	6 (2.67)	30 (13.33)	225
2009	119 (54.59)	14 (6.42)	14 (6.42)	13 (5.96)	7 (3.21)	51 (23.39)	218
2010	105 (48.16)	24 (11.00)	18 (8.26)	26 (11.93)	5 (2.26)	40 (18.35)	218
2011	111 (46.44)	20 (8.37)	26 (10.88)	31 (12.97)	3 (1.26)	48 (20.08)	239
2012	102 (46.79)	21 (9.63)	14 (6.42)	30 (13.76)	13 (5.96)	38 (17.43)	218
2013	120 (49.59)	18 (7.44)	18 (7.44)	30 (12.40)	7 (2.89)	49 (20.25)	242
2014	122 (46.56)	23 (8.78)	26 (9.92)	29 (11.07)	4 (1.53)	58 (22.14)	262
Total:	**2,331 (52.07)**	**320 (7.15)**	**374 (8.35)**	**511 (11.41)**	**76 (1.70)**	**865 (19.32)**	**4,477**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools.

Half-life of the articles published in this series is 15 years since 1991.

Table 4.18: Category - wise analysis of articles published on Sickle cell anemia from all over the world during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	53 (39.85)	18 (13.53)	7 (5.26)	16 (12.03)	0 (0.00)	39 (29.32)	133
1992	73 (46.20)	12 (7.60)	12 (7.60)	18 (11.39)	3 (1.90)	40 (25.31)	158
1993	68 (49.64)	3 (2.19)	19 (13.87)	17 (12.41)	0 (0.00)	30 (21.90)	137
1994	82 (60.74)	3 (2.22)	8 (5.93)	17 (12.59)	1 (0.74)	24 (17.78)	135
1995	81 (55.87)	6 (4.14)	18 (12.41)	7 (4.83)	1 (0.69)	32 (22.07)	145
1996	89 (58.55)	4 (2.63)	13 (8.55)	11 (7.24)	2 (1.32)	33 (21.71)	152
1997	80 (55.56)	14 (9.72)	12 (8.33)	14 (9.72)	2 (1.39)	22 (15.28)	144
1998	74 (45.40)	10 (6.13)	19 (11.66)	24 (14.72)	2 (1.27)	34 (20.86)	163
1999	106 (61.98)	7 (4.09)	11 (6.43)	15 (8.77)	3 (1.75)	29 (16.96)	171
2000	89 (51.74	12 (6.98)	12 (6.98)	18 (10.47)	1 (0.58)	40 (23.26)	172
2001	90 (52.63)	8 (4.68)	13 (7.60)	29 (16.96)	0 (0.00)	31 (18.12)	171
2002	109 (57.07)	16 (8.38)	13 (6.81)	24 (12.57)	1 (0.52)	28 (14.60)	191
2003	95 (60.90)	5 (3.21)	17 (10.90)	15 (9.62)	0 (0.00)	24 (15.38)	156
2004	94 (48.46)	21 (10.82)	18 (9.28)	15 (7.73)	2 (1.03)	44 (22.68)	194

2005	115 (58.38)	21 (10.66)	11 (5.58)	14 (7.11)	4 (2.03)	32 (16.24)	197
2006	124 (52.77)	17 (7.23)	20 (8.51)	33 (14.04)	4 (1.70)	37 (15.74)	235
2007	103 (51.24)	14 (6.97)	20 (9.95)	27 (13.43)	5 (2.49)	32 (15.92)	201
2008	127 (56.44)	9 (1.78)	15 (6.67)	38 (16.89)	6 (2.67)	30 (13.33)	225
2009	119 (54.59)	14 (6.42)	14 (6.42)	13 (5.96)	7 (3.21)	51 (23.39)	218
2010	105 (48.16)	24 (11.00)	18 (8.26)	26 (11.93)	5 (2.26)	40 (18.35)	218
2011	111 (46.44)	20 (8.37)	26 (10.88)	31 (12.97)	3 (1.26)	48 (20.08)	239
2012	102 (46.79)	21 (9.63)	14 (6.42)	30 (13.76)	13 (5.96)	38 (17.43)	218
2013	120 (49.59)	18 (7.44)	18 (7.44)	30 (12.40)	7 (2.89)	49 (20.25)	242
2014	122 (46.56)	23 (8.78)	26 (9.92)	29 (11.07)	4 (1.53)	58 (22.14)	262
Total:	**2,331 (52.07)**	**320 (7.15)**	**374 (8.35)**	**511 (11.41)**	**76 (1.70)**	**865 (19.32)**	**4,477**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Half-life of the articles published in this series is 15 years since 1991.

In the case of articles involving molecular biology related aspects there was a steady increase in the number of articles published since 1991. In 1991 only 4.2% of the total number of articles has been published under this category, while from 1998 onwards about 20% of the articles have been published every year under this category. Half-life of the articles published in this series is 14 years since 1991.

Year-wise and category-wise analysis of the articles published on hemophilia B from all over the world during 1991-2014 is depicted in Table 4.20. Totally 2,324 articles were published on this disorder during the above mentioned period. During first 20 years, 60-119 articles were published every year. However, during the last four years (i.e.during 2011 - 2014) this number increased considerably as 132 -172 articles were published every year on this disorder. 23.23-41.61% of the total articles were published every year under 'original article' category. The last category which comprises of articles published using molecular biology techniques reveals some interesting pattern. About 35-45% (range 31.67-45.45%) of articles were published under this category during 1991-1996. However, later on with one or two exceptions it was observed that there was a decrease in the percentage of articles published under this category (range 13.76-28.77%). On an average 26.68% of the total number of articles were published under this category every year. Half-life of the articles published on this disorder in this series is 14 years since 1991; which suggests that 50% of the articles were published during last 10 years.

Table 4.21 demonstrates year-wise and category-wise analysis of the articles published on G6PD deficiency from all over the world during 24 years (i.e.1991-2014). Totally 1,784 articles were published during above mentioned time period. This number was considerably less as compared to those published on hemophilia A or Down syndrome. 'Original articles' published on this disorder were in the range of 37.98-67.12%. In the case of category 6 (articles published using molecular biology related techniques) no particular trend was seen. During the year 2002 and 2005, 42.03% and 36.31% respectively of the total articles were published under this category. Half-life of the articles published in this series was 14 years since 1991.

Table 4.19: Year - wise and category - wise analysis of articles published on Down syndrome from all over the world during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	145 (50.88)	15 (5.26)	23 (8.07)	32 (11.23)	58 (20.35)	12 (4.20)	285
1992	140 (46.67)	16 (5.33)	26 (8.67)	40 (13.33)	60 (20.00)	18 (6.00)	300
1993	153 (48.41)	28 (8.86)	27 (8.54)	30 (9.49)	52 (16.46)	26 (8.23)	3 16
1994	143 (47.83)	11 (3.68)	22 (7.36)	28 (9.36)	65 (21.74)	30 (10.08)	299
1995	152 (45.78)	31 (9.34)	29 (8.73)	27 (8.13)	53 (15.96)	40 (12.05)	33
1996	152 (52.23)	14 (4.81)	22 (7.56)	38 (13.06)	15 (5.15)	50 (17.18)	291
1997	186 (56.88)	14 (4.28)	16 (4.89)	31 (9.48)	26 (7.95)	54 (16.51)	327
1998	180 (49.05)	25 (6.81)	26 (7.08)	20 (5.45)	36 (9.81)	80 (21.80)	367
1999	174 (52.41)	26 (7.83)	16 (4.82)	16 (4.82)	27 (8.13)	73 (21.99)	332
2000	184 (42.89)	33 (7.69)	29 (6.46)	53 (11.80)	41 (9.13)	89 (19.82)	429
2001	137 (34.86)	46 (11.70)	24 (6.11)	49 12.47	45 (11.45)	92 23.41)	393
2002	153 (37.87)	51 (12.62)	28 (6.93)	48 (11.88)	39 (9.65)	85 (21.03)	404
2003	142 (39.55)	61 (16.99)	30 (8.36)	34 (9.47)	23 (6.41)	69 (19.22)	359
2004	126 (31.42)	89 (22.19)	33 (8.23)	56 (13.97)	24 (5.96)	73 (18.20)	401

2005	122 (27.60)	69 (15.61)	36 (8.14)	62 (94.03)	70 (15.84)	83 (18.78)	442
2006	139 (33.49)	62 (14.94)	32 (7.71)	38 (9.16)	55 (13.25)	89 (21.45)	415
2007	140 (31.89)	74 (16.86)	31 (7.06)	42 (9.57)	69 (15.72)	83 (18.91)	439
2008	146 (34.68)	65 (15.44)	39 (9.26)	37 (8.79)	59 (14.01)	75 (17.81)	421
2009	171 (35.40)	54 (11.18)	51 (10.56)	25 (5.18)	94 (9.46)	88 (18.22)	483
2010	127 (26.29)	73 (15.11)	35 (7.25)	44 (9.11)	118 (24.43)	86 (17.81)	483
2011	147 (30.18)	43 (8.83)	32 (6.57)	56 (11.50)	85 (17.45)	124 (25.46)	487
2012	171 (32.20)	52 (9.79)	35 (6.59)	57 (10.73)	97 (18.27)	119 (22.41)	531
2013	197 (35.18)	41 (7.32)	38 (6.79)	37 (6.61)	114 (20.36)	133 (23.75)	560
2014	198 (33.97)	41 (7.07)	48 (8.28)	47 (8.10)	112 (19.66)	133 (22.93)	579
Total	**3,725 (38.50)**	**1,034 (10.69)**	**728 (7.52)**	**947 (9.79)**	**1,437 (14.85)**	**1,804 (18.65)**	**9,675**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Half-life of the articles published in this series is 14 years since 1991.

Table 4.20: Category - wise analysis of articles published on Hemophilia B from all over the world during 1991-2014.

Year	1	2	3	4	5	6	Total
1991	32 ((39.51)	4 (4.94)	8 (9.88)	2 (2.47)	1 (1.23)	34 (41.98)	81
1992	29 (38.67)	7 (9.33)	7 (9.33)	2 (2.67)	2 (2.67)	28 (37.33)	75
1993	23 (23.23)	9 (9.09)	9 (9.09)	11 (11.11)	2 (2.02)	45 (45.45)	99
1994	21 (26.25)	8 (10.00)	6 (7.50)	10 (12.5)	1 (1.25)	34 (42.5)	80
1995	23 (27.71)	7 (8.43)	9 (10.84)	12 (14.46)	1 (1.20)	31 (37.34)	83
1996	20 (33.33)	8 (13.33)	4 (6.67)	9 (16.67)	0 (0.00)	19 (31.67)	60
1997	28 (38.36)	10 (13.70)	4 (5.48)	10 (13.70)	0 (0.00)	21 (28.77)	73
1998	26 (31.71)	20 (24.39)	5 (6.10)	8 (9.76)	3 (3.66)	20 (24.39)	82
1999	27 (33.33)	13 (16.05)	6 (7.41)	7 (8.64)	2 (2.47)	26 (32.10)	81
2000	36 (37.11)	10 (10.30)	11 (11.34)	14 (14.43)	2 (2.06)	24 (24.74)	97
2001	28 (34.15)	10 (12.20)	8 (9.76)	11 (13.41)	3 (3.66)	22 (26.82)	82
2002	30 (40.54)	12 (16.21)	4 (5.40)	8 (10.81)	3 (4.05)	17 (22.97)	74
2003	26 (25.49)	25 (24.51)	5 (4.90)	14 (13.73)	2 (1.96)	30 (29.41)	102
2004	34 (34.69)	18 (18.37)	6 (6.12)	10 (10.20)	3 (3.06)	27 (27.55)	98

2005	29 (39.19)	11 (14.86)	4 (5.41)	6 (8.11)	2 (2.70)	22 (29.73)	74
2006	35 (41.61)	05 (5.95)	06 (7.14)	12 (14.29)	8 (9.50)	18 (21.42)	84
2007	23 (36.50)	5 (7.94)	9 (14.29)	4 (6.35)	1 (1.59)	21 (33.33)	63
2008	38 (31.93)	10 (8.40)	18 (15.13)	12 (10.08)	11 (9.24)	30 (25.21)	119
2009	35 (39.32)	8 (8.99)	9 (10.11)	8 (8.99)	5 (5.62)	24 (26.97)	89
2010	44 (40.37)	19 (17.43)	7 (6.42)	9 (8.25)	15 (13.76)	15 (13.76)	109
2011	50 (37.87)	15 (11.36)	17 (12.88)	9 (6.82)	18 (13.63)	23 (17.42)	132
2012	60 (34.09)	22 (12.5)	11 (6.25)	28 (15.91)	19 (10.80)	36 (20.45)	176
2013	53 (38.12)	24 (17.26)	12 (8.63)	18 (12.94)	8 (5.76)	24 (17.27)	139
2014	67 (38.95)	23 (13.37)	11 (6.40)	29 (16.86)	13 (7.55)	29 (16.86)	172
Total	**817 (35.15)**	**303 (13.03)**	**196 (8.43)**	**263 (11.32)**	**125 (5.38)**	**620 (26.68)**	**2,324**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Half-life of the articles published in this series is 14 years since 1991.

Table 4.21: Category - wise analysis of articles published on G6PD deficiency from all over the world during 1991 – 2014.

Year	1	2	3	4	5	6	Total
1991	49 (67.12)	0 (0.00)	7 (9.59)	5 (6.85)	0 (0.00)	12 (16.44)	73
1992	53 (64.63)	2 (2.44)	3 (3.66)	5 (6.10)	0 (0.00)	19 (23.17)	82
1993	28 (50.91)	2 (3.64)	5 (9.09)	3 (5.45)	1 (1.82)	16 (29.09)	55
1994	27 (39.71	2 (2.94)	6 (8.82)	7 (10.24)	1 (1.47)	25 (36.76)	68
1995	30 (40.00)	2 (2.67)	8 (10.67)	13 (17.33)	1 (1.33)	21 (28.00)	75
1996	33 (47.14)	6 (8.57)	4 (5.71)	8 (11.43)	0 (0.00)	19 (27.14)	70
1997	27 (52.94)	1 (1.96)	2 (3.92)	3 (5.88)	1 (1.96)	17 (33.33)	51
1998	35 (58.33)	4 (6.67)	3 (5.00)	9 (15.00)	0 (0.00)	9 (15.00)	60
1999	35 (47.30)	11 (14.86)	3 (4.05)	6 (8.11)	2 (2.70)	17 (22.97)	74
2000	26 (43.33)	4 (6.67)	6 (10.00)	3 (05.00)	0 (0.00)	21 (35.00)	60
2001	22 (42.31)	3 (5.77)	7 (13.46)	6 (11.54)	1 (1.92)	13 (25.00)	52
2002	23 (33.33)	4 (5.79)	8 (11.59)	5 (7.25)	0 (0.00)	29 (42.03)	69
2003	38 (55.88)	1 (1.47)	8 (11.76)	7 (10.29)	1 (1.47)	13 (19.12)	68
2004	35 (47.95)	3 (4.11)	5 (6.85)	7 (9.59)	1 (1.37)	22 (30.14)	73

2005	41 (46.59)	1 (1.14)	8 (10.09)	6 (6.28)	0 (0.00)	32 (36.36)	88
2006	35 (59.32)	1 (1.69)	5 (8.47)	3 (5.08)	3 (5.08)	12 (20.34)	59
2007	40 (51.28)	3 (3.85)	5 (6.41)	7 (8.97)	1 (1.28)	22 28.21)	78
2008	42 (49.41)	0 (0.00)	10 (11.76)	7 (8.24)	0 (0.00)	26 (30.59)	85
2009	35 (47.30)	4 (5.41)	10 (13.51)	6 (8.11)	0 (0.00)	19 (25.68)	74
2010	49 (58.33)	2 (2.38)	7 (8.33)	6 (7.14)	2 (2.38)	18 (21.43)	84
2011	39 (50.65)	2 (2.60)	9 (11.69)	4 (5.19)	2 (2.60)	21 (27.27)	77
2012	48 (63.16)	3 (3.95)	6 (7.89)	4 (5.26)	1 (1.32)	14 (18.42)	76
2013	47 (45.19)	9 (8.65)	12 (11.54)	10 (9.62)	2 (1.92)	24 (23.08)	104
2014	49 (37.98)	4 (3.10)	18 (13.95)	15 (11.63)	4 (3.10)	39 (30.2)	129
Total	**886 (49.66)**	**74 (4.15)**	**165 (9.25)**	**155 (8.69)**	**24 (1.35)**	**480 (26.91)**	**1,784**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Half-life of the articles published in this series is 14 years since 1991.

Table 4.22: Category - wise analysis of articles published on Hemophilia A from all over the world during 1991 – 2014.

Year	1	2	3	4	5	6	Total
1991	119 (50.00)	18 (7.56)	19 (7.98)	24 (10.08)	1 (0.42)	57 (23.95)	238
1992	127 (46.86)	41 (15.13)	33 (12.18)	17 (6.27)	10 (3.69)	43 (15.87)	271
1993	127 (52.26)	23 (9.47)	28 (11.52)	18 (7.41)	4 (1.65)	43 (17.70)	243
1994	143 (39.94)	24 (6.70)	28 (7.82)	13 (3.63)	7 (1.96)	43 (39.94)	258
1995	136 (51.91)	12 (4.58)	23 (8.78)	20 (7.36)	13 (4.96)	58 (22.14)	262
1996	136 (52.92)	27 (10.51)	22 (8.56)	12 (4.67)	6 (2.33)	54 (21.01)	257
1997	98 (45.16)	34 (15.67)	9 (4.15)	9 (4.15)	6 (2.76)	61 (28.11)	217
1998	138 (45.39)	30 (9.87)	34 (11.18)	21 (6.91)	21 (6.91)	60 (19.74)	304
1999	100 (34.36)	51 (71.53)	36 (12.37)	15 (5.15)	17 (5.84)	72 (24.74)	291
2000	129 (42.57)	58 (19.14)	26 (8.58)	22 (7.26)	11 (3.63)	57 (18.81)	303
2001	133 (44.48)	36 (12.04)	29 (9.70)	16 (5.35)	12 (4.01)	73 (24.41)	299
2002	125 (39.81)	43 (13.69)	35 (11.15)	19 (6.05)	13 (4.14)	79 (25.16)	314
2003	117 (33.72)	48 (13.83)	39 (11.24)	28 (8.07)	30 (8.05)	85 (24.50)	347
2004	170 (44.62)	33 (8.66)	45 (11.81)	35 (9.19)	25 (6.56)	73 (19.16)	381

2005	147 (44.93)	24 (7.50)	31 (9.68)	10 (3.13)	24 (7.50)	84 (26.25)	320
2006	181 (47.26)	34 (8.28)	44 (11.49)	28 (7.31)	25 (6.53)	71 (18.54)	383
2007	175 (46.54)	39 (10.37)	50 (13.30)	16 (4.26)	19 (5.05)	77 (20.48)	376
2008	179 (42.32)	54 (12.77)	48 (11.35)	33 (7.80)	34 (8.04)	75 (17.73)	423
2009	200 (45.45)	35 (7.95)	61 (13.68)	42 (9.55)	32 (7.27)	70 (15.91)	440
2010	193 (43.08)	40 (8.93)	49 (10.94)	48 (10.71)	28 (6.25)	90 (20.09)	448
2011	210 (45.06)	57 (12.23)	40 (8.58)	29 (6.22)	28 (6.01)	102 (21.89)	466
2012	212 (36.05)	90 (15.31)	82 (13.95)	45 (7.65)	37 (6.29)	122 (20.75)	588
2013	203 (39.19)	73 (14.09)	54 (10.42)	46 (8.88)	43 (8.30)	149 (19.11)	568
2014	239 (38.93)	73 (11.89)	61 (9.93)	41 (6.68)	55 (10.59)	145 (23.62)	614
Total	**3,737 (43.40)**	**997 (11.58)**	**926 (10.75)**	**607 (7.05)**	**501 (5.82)**	**1,843 (21.40)**	**8,611**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Half-life of the articles published in this series is 14 years since 1991.

Table 4.22 demonstrates year-wise and category-wise analysis of the articles published on hemophilia A from all over the world during 1991-2014. Totally 8,611 articles were published on this disorder during the above stated time period. During the first twelve year period (i.e 1991-2002) about 260-280 articles (range 217-314) were published every year while during the next twelve years the number of articles published every year increased considerably (range 320-614). About 35-50% of the total articles (range 33.72-52.92%) were published every year as 'original articles'. Around 15-25% of the total articles (range 15.87-26.25%) were published every year under category 6. Half-life of the articles published under this series is 15 years since 1991.

Table 4.23 reveals category-wise analysis of the articles published on six genetic disorders from all over the world during 1991-2014. Articles published on G6PD deficiency (1,784) and on hemophilia B (2,324) were less as compared to those published on remaining four disorders (range 4,477-9,675). About 35-50% of the total articles (range 35.15% for hemophilia B – 52.07% for vWD) were published under category 6. (articles published using molecular biology tools).

As done for the articles published from India, the articles published from all over the world were divided into two groups with two different time intervals i.e. publications during 1991-2002 and publications during 2003-2014 to see whether any statistical difference is there in publications of the articles under different categories during two time intervals or not?

Table 4.24 reveals category-wise analysis of the articles published on six genetic disorders from all over the world during 1991-2002. Totally 14,546 articles were published on all the six disorders from all over the world during above stated period. Range of articles published as 'original articles' varies between 33.4% (Hemophilia B) to 53.1%

Table 4.25 depicts category-wise analysis of the articles published on six genetic disorders from all over the world during 2003-2014. Totally 21,560 articles have been published from all over the world on the six disorders during above stated time period. 40.44% of the total articles were published as 'original articles' while 20.45% of the total articles were published under category 6. (von Willebrand disease). About $1/5^{th}$ of the total articles published on all the disorders during this time period were from category 6.

Table 4.23: Category - wise analysis of articles published on six genetic disorders from all over the world during 1991-2014.

Genetic Disease	1	2	3	4	5	6	Total	Half life Of Publ. (years)
Sickle cell anemia	3,920 (42.40)	1,714 (18.54)	809 (8.75)	613 (6.63)	504 (5.45)	1,685 (18.23)	9,245	15
Hemophilia A	3,737 (43.40)	997 (11.48)	926 (10.75)	607 (7.05)	501 (5.82)	1,843 (21.40)	8,611	15
Hemophilia B	817 (35.15)	303 (13.03)	196 (8.43)	263 (11.32)	125 (5.38)	620 (26.68)	2,324	14
VonWillebrand disease	2,331 (52.07)	320 (7.15)	374 (8.35)	511 (11.41)	76 (1.70)	865 (19.32)	4,477	15
G6PD deficiency	3,725 (38.50)	1,034 (10.69)	728 (7.52)	947 (9.79)	1,437 (14.85)	1,804 (18.65)	9,675	14
Down syndrome	886 (49.66)	74 (4.15)	165 (9.25)	155 (8.69)	24 (1.35)	480 26.91)	1,784	14
Total	**15,416 (42.69)**	**4,442 (12.30)**	**3,198 (8.86)**	**3,096 (8.57)**	**2,667 (7.39)**	**7,297 (20.20)**	**36,116**	

Figures in brackets percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular Biology tools

Table 4.24: Category - wise analysis of articles published on six genetic disorders from all over the world during 1991-2002.

Genetic Disease	1	2	3	4	5	6	Total
Sickle cell anemia	1,582 (44.12)	714 (19.91)	314 (8.76)	197 (5.49)	170 (4.74)	609 (16.98)	3,586
Hemophilia A	1,511 (46.39)	397 (12.19)	322 (9.89)	206 (6.32)	121 (3.72)	700 (21.49)	3,257
Hemophilia B	323 (33.40)	118 (12.20)	81 (8.38)	104 (10.75)	20 (2.07)	321 (33.20)	967
vonWillebrand disease	994 (53.10)	113 (6.04)	157 (8.39)	210 (11.22)	16 (0.86)	382 (20.41)	1,872
G6PD deficiency	388 (49.18)	41 (5.20)	62 (7.86)	73 (9.25)	7 (0.09)	218 (27.63)	789
Down syndrome	1,899 (46.60)	910 (7.61)	288 (7.07)	412 (10.11)	517 (12.69)	649 (15.93)	4,075
Total	**6,697 (46.04)**	**1,693 (11.64)**	**1,224 (8.42)**	**1,202 (8.26)**	**851 (5.85)**	**2,879 (19.79)**	**14,546**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Table 4.25: Category - wise analysis of articles published on six genetic disorders from all over the world during 2003-2014.

Genetic Disease	1	2	3	4	5	6	Total
Sickle cell anemia	2,338 (41.39)	1,000 (17.70)	495 (8.76)	416 (7.36)	334 (5.91)	1,066 (18.87)	5,649
Hemophilia A	2,226 (41.58)	600 (11.21)	604 (11.28)	401 (7.41)	380 (7.10)	1,143 (21.35)	6,354
Hemophilia B	494 (36.40)	185 (13.63)	115 (8.48)	159 (11.72)	105 (7.74)	299 (22.03)	1,357
von Willebrand disease	1,337 (51.32)	207 (7.95)	217 8.33	301 (11.56)	60 (2.30)	483 (18.54)	2,605
G6PD deficiency	498 (50.05)	33 (3.32)	103 (10.35)	82 (8.24)	17 (1.71)	262 (26.33)	995
Down syndrome	1,826 (32.61)	724 (12.93)	440 (7.86)	535 (9.55)	920 (16.43)	1,155 (20.63)	5,600
Total	**8,719 (40.44)**	**2,749 (12.75)**	**1,974 (9.16)**	**1894 (8.79)**	**1,816 (8.42)**	**4,408 (20.45)**	**21,560**

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

X^2 test was used to compare the data on six genetic disorders from all over the world during two time intervals. The values were considered for one degree of freedom and $p<0.05$ was considered as statistically significant value. The results are tabulated in Table 4.26. Totally in the case of 13 combinations, statistically significant difference was observed. In the case of Down syndrome there was a statistically significant increase in the number of articles published under category 6 during 'second time interval' as compared to those published during 'first time interval'. On the other hand in the case of hemophilia B, there was a statistically significant decrease in the number of articles published under category 6 during second time interval as compared to those published during first time interval.

Figure 4.18 depicts graphically the trend of articles published from all over the world under category 6 on six genetic disorders.

4.1.3 Comparison of the data

Table 4.27 describes comparison of the data published from India as well as from all over the world on six genetic disorders during 1991-2002. Articles published from India on all the disorders were very less as compared to those from all over the world during this time period. About $1/5^{th}$ of the articles from both the groups were published on data involving various molecular biology aspects; while 40-45% of the articles were published as 'original articles' from both the groups during the above stated time period.

Table 4.28 depicts comparison of the data published from India as well as from all over the world on six genetic disorders during 2003-2014. Totally 679 articles from India and 21,560 articles from all over the world were published during above stated time periods on six disorders. 26.36% of the total number of articles published from India on all the six disorders during 2003-2014 were those involving various molecular biology aspects while 20.45% of the total number of articles published from all over the world were from the same category (i.e. category number 6). In the case of hemophilia B 50.94% of the total number of articles were published under category 6 while during the same time period only 22.03% of the total number of articles were published from all over the world on hemophilia B. Similar picture was seen in the case of von Willebrand Disease. 41.07% of the total number of articles published from India were from category number 6 as compared to 18.54% of the total number articles published from all over the world were from the same category.

In the case of original articles, there was a considerable decrease in the total number of articles published from India (28.87%) on all the disorders as compared to those published from all over the world (40.44%) during the same time period.

Comparison of the articles published from the above mentioned two groups during 1991-2014 under different categories on six disorders is shown in Table 4.29. Totally 899 articles published from India; while 36,116 articles were published from all over the world during above stated time period. 25.25% of the total number of articles were published from India while 20.20% of the total number of articles were published from all over the world on all the disorders were from category number 6. In the case of original articles there was considerable decrease in the number of articles published from India (33.63%) as compared to those published from all over the world (42.69%) on all the six disorders.

4.1.4 Trend of publications during 2015-2020.

Trend of publications of articles published on six genetic disorders from India as well as from all over the world during last five years i.e. 2015-2020 is depicted in Table 4.30.The share of Indian publications in the global context has increased considerably (range1.92 - 8.65%) However the pattern of Indian share in different disorders remained same as compared to Table3.1 which is showing pattern of Indian share up to 2014.

4.1.5 Growth rate

Based on the data published from India as well as from all over the world and separated in to two groups (i.e. publications during 1991-2002 and publications during 2003-2014) growth rate of the articles published on each disorder was calculated which is presented in Table 4.31. In the case of India, the lowest growth was seen in articles on G6PD deficiency (31.37%) while in the case of remaining disorders considerable growth was seen in the publications of articles (range 107.6-685.71%). Maximum growth was seen in the case of von Willebrand disease (685.71%) followed by Down syndrome (268.29%) . On the other hand a steady growth was observed in the publications of articles from all over the world (range 26.11.64.38%). On the other hand a steady growth was observed in the publications of articles from all over the world (range 26.11-64.38%). Maximum growth was seen in the case of hemophilia A (64.38%) followed by sickle cell anemia (57.53%).

Table 4.26: X^2 contingency table to compare articles published under different categories from all over the world on sickle cell anemia, Hemophiilia B,von Willebrand disease, G6PD deficiency, Down syndrome and Hemophilia A during two time intervals i.e. 1991 to 2002 and 2003 to 2014.

Sickle cell anemia

X^2	p value	Significance
1 x 1 - 2.6888	p = 0.1011	NS
2 x 2 - 4.8425	p = 0.0278	Significant
3 x 3 - 0.0001	p = 0.9923	NS
4 x 4 - 10.8864	p = 0.0010	Significant
5 x 5 - 5.2474	p = 0.0220	Significant
6 x 6 - 3.6662	p = 0.0557	NS

Hemophilia B

X^2	p value	Significance
1 x1 - 1.0748	p = 0.2999	NS
2 x 2 - 0.7855	p = 0.3755	NS
3 x 3 - 0.0059	p = 0.9385	NS
4 x 4 - 0.335	p = 0.5629	NS
5 x 5 - 31.279	p = <0.0001	Significant
6 x 6 - 20.138	p = < 0.0001	Significant

von Willebrand disease

X^2	p value	Significance
1 x1 - 0.399	p = 0.5277	NS
2 x 2 - 4.939	p = 0.0263	Significant
3 x 3 - 0.000	p = 0.9940	NS
4 x 4 - 0.070	p = 0.7915	NS
5 x 5 - 12.429	p = 0.0004	Significant
6 x 6 - 1.544	p = 0.2140	NS

G6PD deficiency

X^2	p value	Significance
1 x 1 - 0.0453	p = 0.8315	NS
2 x 2 - 3.5926	p = 0.0580	NS
3 x 3 - 2.7158	p = 0.093	NS
4 x 4 - 0.475	p = 0.4903	NS
5 x 5 - 2.1795	p = 0.1398	NS
6 x 6 - 0.2170	p = 0.6414	NS

Down syndrome

X^2	p value	Significance
1 x1 - 85.47	p <0.0001	Significant
2 x 2 - 56.92	p <0.0001	Significant
3 x 3 - 01.82	p = 0.1773	NS
4 x 4 - 0.6801	p = 0.4095	NS
5 x 5 - 19.4602	p <0.0001	Significant
6 x 6 - 23.6948	p <0.0001	Significant

Hemophilia A

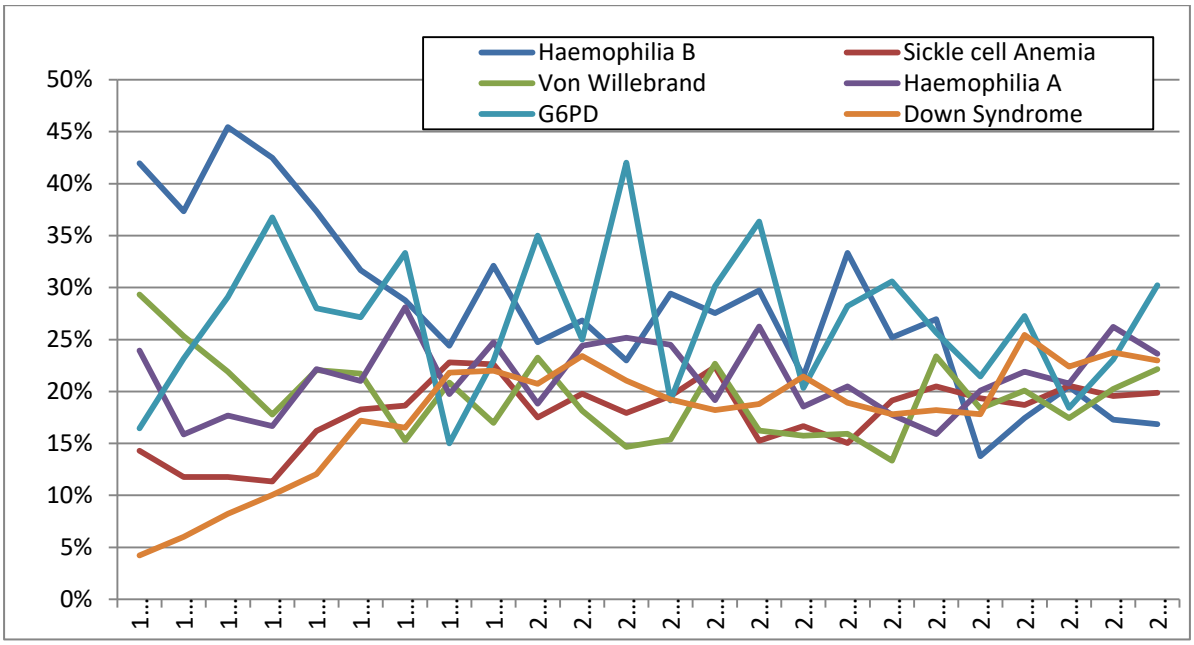
X^2	p value	Significance
1 x1 - 7.4882	p < 0.0062	Significant
2 x 2 - 1.5101	p = 0.2191	NS
3 x 3 - 3.3185	p = 0.0685	NS
4 x 4 - 3.6514	p = 0.0560	NS
5 x 5 - 37.9361	p < 0.0001	Significant
6 x 6 - 0.0161	p = 0.8991	NS

NS = Not Significant

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

X^2 test done for 1 degree of freedom.
X^2 test results are considered statistically significant when p value is < 0.05

Figure 4.18: Trend of the articles published from all over the world under category 6 (using molecular biology tools) on six genetic disorders (Viz. Down syndrome, Hemophilia A, G6PD deficiency, Hemophilia B, von Willebrand disease and Sickle cell anemia during 1991-2014.



X axis – Years

Y axis – Percent of articles published under category 6

Observation - Any particular pattern of publications is not seen.

Table 4.27: Comparison of articles published under various categories on six genetic disorders from India and from all over the world during 1991-2002.

GD	Category	1	2	3	4	5	6	Total
SCA	India	27 (34.18)	18 (22.78)	5 (6.33)	11 (13.92)	0 (0.00)	18 (22.78)	79
	International	1,582 (44.12)	714 (19.91)	314 (8.76)	197 (5.49)	170 (4.74)	609 (16.98)	3,586
Hem A	India	15 (24.59)	13 (21.31)	13 (21.31)	5 (8.20)	3 (4.92)	12 (19.67)	61
	International	1,511 (46.39)	397 (12.19)	322 (9.89)	206 (6.32)	121 (3.72)	700 (21.49)	3,257
Hem B	India	5 (25.00)	3 (15.00)	5 (25.00)	2 (10.00)	1 (5.00)	4 (20.00)	20
	International	323 (33.40)	118 (12.20)	81 (8.38)	104 (10.75)	20 (2.07)	321 (33.20)	967
vWD*	India	3	1	2	0	0	1	7
	International	994 (53.10)	113 (6.04)	157 (8.39)	210 (11.22)	16 (0.86)	382 (20.41)	1,872
DS	India	21 (51.22)	0 (0.00)	10 (24.39)	6 (14.63)	1 (2.44)	3 (7.32)	41
	International	1,899 (46.60)	310 (7.61)	288 (7.07)	412 (10.11)	517 (12.69)	646 (15.93)	4,075
G6PD	India	35 (68.63)	0 (0.00)	4 (7.84)	3 (5.88)	0 (0.00)	9 (17.65)	51
	International	388 (49.18)	41 (5.20)	62 (7.86)	73 (9.25)	7 (0.09)	218 (27.63)	789
Total	**India**	**106 (40.93)**	**35 (13.50)**	**39 (15.06)**	**27 (10.42)**	**5 (1.93)**	**47 (18.14)**	**259**
	International	**6,697 (46.04)**	**1,693 (11.64)**	**1,224 (8.42)**	**1,202 (8.26)**	**851 (5.85)**	**2,879 (19.79)**	**14,546**

Table 4.27 (Contd.)

SCA - Sickle cell anemia
HemA - Hemophilia A
Hem B - Hemophilia B
vWD - von Willebrand disease
DS - Down syndrome
G6PD - G6PD deficiency

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

*****Due to small sample size percentage values are not calculated.

Table 4.28: Comparison of articles published under various categories on six genetic disorders from India and from all over the world during 2003-2014.

GD	Category	1	2	3	4	5	6	Total
SCA	India	62 (37.80)	24 (14.63)	23 (14.02)	12 (7.32)	8 (4.88)	35 (21.34)	164
	International	2,338 (41.39)	1,000 (17.70)	495 (8.75)	416 (7.36)	334 (5.91)	1,066 (18.87)	5,649
Hem A	India	30 (15.87)	26 (13.76)	23 (12.17)	10 (5.29)	12 (6.35)	48 (25.40)	189
	International	2,226 (41.58)	600 (11.21)	604 (11.28)	401 (7.41)	380 (7.10)	1,143 (21.35)	5,354
Hem B	India	8 (15.09)	5 (9.43)	9 (16.98)	3 (5.66)	1 (1.89)	27 (50.94)	53
	International	494 (36.40)	185 (13.63)	115 (8.48)	159 (11.72)	105 (7.74)	299 (22.03)	1,357
vWD*	India	21 (38.18)	4 (7.27)	3 (5.45)	4 (7.27)	1 (1.82)	23 (40.00)	56
	International	1,337 (51.32)	207 (7.95)	217 (8.33)	301 (11.56)	60 (2.30)	483 (18.54)	2,605
DS	India	41 (27.15)	20 (13.25)	38 (25.17)	9 (5.96)	11 (7.28)	32 (21.19)	151
	International	1,826 (32.61)	724 (12.93)	440 (7.86)	535 (9.55)	920 (16.43)	1,155 (20.63)	5,600
G6PD	India	34 (50.74)	1 (1.49)	12 (17.91)	3 (4.48)	2 (2.99)	15 (22.39)	67
	International	498 (50.05)	33 (3.32)	103 (10.35)	82 (8.24)	17 (1.71)	262 26.33)	995
Total	**India**	**196 (28.87)**	**80 (11.78)**	**108 (15.91)**	**41 (6.04)**	**35 (5.16)**	**180 (26.47)**	**680**
	International	**8,719 (40.44)**	**2,749 (12.75)**	**1,974 (9.16)**	**1,894 (8.79)**	**1,816 (8.42)**	**4,408 (20.45)**	**21,560**

Table 4.28 (Contd.)

SCA - Sickle cell anemia
HemA - Hemophilia A
Hem B - Hemophilia B
vWD - von Willebrand disease
DS - Down syndrome
G6PD - G6PD deficiency

Figures in brackets indicate percentage values

1. Research / Original articles
2. Articles based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Table 4.29: Comparison of articles published under various categories on six genetic disorders from India and from all over the world during 1991-2014.

GD	Category	1	2	3	4	5	6	Total	Half life Yr
SCA	India	89 (36.63)	42 (17.28)	28 (11.52)	23 (9.47)	8 (3.29)	53 (21.81)	243	18
	International	3,920 (42.40)	1,714 (18.54)	809 (8.75)	613 (6.63)	504 (5.45)	1,685 (18.23)	9,245	15
HemA	India	45 (21.43)	39 (18.57	36 (17.14)	15 (7.14)	15 (7.14)	60 (28.57)	210	15
	International	3,737 (43.40)	997 (11.58)	926 (10.75)	607 (7.05)	501 (5.82)	1,843 (21.40)	8,611	15
HemB	India	13 (17.80)	8 (10.96)	14 (19.18)	5 (6.85)	2 (2.74)	31 (42.40)	73	17
	International	817 (35.15)	303 (13.03)	196 (8.43)	2663 (11.32)	125 (5.38)	620 (26.68)	2,324	14
vWD	India	24 (38.71)	5 (8.06)	5 (8.06)	4 (6.45)	1 (1.61)	24 (38.10)	63	18
	International	2,331 (52.07)	320 7.15)	374 (8.35)	511 (11.41)	76 (1.70)	865 (19.32)	4,477	15
DS	India	62 (32.29)	20 (10.42)	48 (25.00)	15 (7.81)	12 (6.25)	35 (18.23)	192	19
	International	3,725 (38.50)	1,034 (10.69)	728 (7.52)	947 (9.79)	1,437 (14.85)	1,804 (18.65)	9,675	14
G6PD	India	69 (54.48)	1 (0.85)	16 (13.56)	6 (5.08)	2 (1.69)	24 (20.34)	118	14
	International	886 (49.66)	74 (4.15)	165 (9.25)	155 (8.69)	24 (1.35)	480 (26.91)	1,784	14
Total	**India**	**302 (33.63)**	**115 (12.81)**	**147 (16.37)**	**68 (7.57)**	**40 (4.45)**	**227 (25.25)**	**899**	
	International	**15,416 (42.69)**	**4,442 (12.30)**	**3,198 (8.86)**	**3,096 (8.57)**	**2,667 (7.39)**	**7,297 (20.20)**	**36,116**	

Table 4.29 (Contd.)

SCA - Sickle cell anemia
HemA - Hemophilia A
Hem B - Hemophilia B
vWD - von Willebrand disease
DS - Down syndrome
G6PD - G6PD deficiency

Figures in brackets indicate percentage values

1. Research / Original articles
2. Article Based on clinical findings
3. Case reports
4. Reviews
5. Miscellaneous articles
6. Articles published using molecular biology tools

Table 4.30: Trend of publications of articles published on six genetic disorders from India as well as from all over the world during 2015-2020.

Sr.no.	Genetic disorders	Region	Number of publications
1	Sickle cell anemia	India	201(3.18)
2	Sickle cell anemia	Worldwide	6,317
3	Down syndrome	India	290 (3.05)
4	Down syndrome	Worldwide	9,510
5	G6PD	India	80(8.65)
6	G6PD	Worldwide	924
7	Hemophilia A	India	173 (3.90)
8	Hemophilia A	Worldwide	4,439
9	Hemophilia B	India	39 (3.23)
10	Hemophilia B	Worldwide	1,206
11	von Willebrand disease	India	32(1.92)
12	von Willebrand disease	Worldwide	1,670

Figures in brackets indicate percentage values.

Table 4.31: Comparison between growth rates seen in publications on six genetic disorders from India during 1991-2002 and 2003-2014 and in publications on the same genetic disorders from all over the world during the same two time intervals.

Genetic Disease	Publications during (India)			Growth during (%)	Publications during (all over the world)			Growth during (%)
	91-14	91-02	03-14	91-02to 03-14	91-14	91-02	03-14	91-02 to 03-14
Sickle cell anemia	243	79	164	107.60	9,245	3,586	5,609	57.53
Hemophilia A	210	61	189	209.84	8,611	3,257	5,354	64.38
Hemophilia B	73	20	53	165.00	2,324	967	1,357	40.33
von Willebrand disease	63	7	56	685.71	4,477	1,872	2,605	39.16
G6PD deficiency	118	51	67	31.37	1,784	789	995	26.11
Down syndrome	192	41	151	268.29	9,675	4,075	5,600	37.42

Formula to calculate % growth rate

- Increase in publications = Number of publications during 2003-2014 – Number of publications during 1991-2002

- % growth rate = $\frac{\text{Increase in publications X 100}}{\text{No. of publications during 1991-2002}}$

4.2 Network analysis

As mentioned earlier data for network analysis was downloaded from SCOPUS database. This included the articles published from India during 1991-2014 on six genetic disorders. Data on each disorder was divided into two groups, using specific keywords. The first group included the articles published during 1991-2014 on a particular disorder from India without any data related to molecular biology while the second group included the articles published from India during the same time period on the same disorder with the data related to molecular biology. Thus twelve datasets (two for each disorder) were generated and subjected to network analysis.

Table 4.32 demonstrates the preliminary data obtained from SCOPUS database involving productivity pattern of authors who published artiicles on six genetic disorders from India during 1991-2014 and without any data related to molecular biology. Maximum number of articles were published on Down syndrome followed by those on hemophilia A (459) and sickle cell anemia (436). Single authored articles on all the disorders ranged between 4.59-9.6% indicating the importance of collaboration in research on these disorders. Annual percent growth rate was the lowest in the case of G6PD deficiency (2.58%) while it was the highest (13.79%) in the case of hemophilia A.

Table 4.33 illustrates the preliminary data obtained from SCOPUS database involving productivity pattern of authors who published on six genetic disorders from India during 1991-2014 and involving some data related to various aspects of molecular biology. Relatively less number of articles were published in this group on each genetic disorders compared to the earlier group. The articles on each disorder ranged between 13-78. In the case of hemophilia B the articles were listed since 1998 onwards while in the case of von Willebrand disease the articles were listed since 2003 onwards. The range of articles published on these disorders varied between 13 (von Willebrand disease) to 78 (hemophilia A). Single authored articles in this group varied between 0-12.5%. Authors per article ranged between 2.31-3.94. Annual percent growth rate was 0% for G6PD deficiency while it was 11.64% for Down syndrome.

4.2.1 Sickle cell anemia

Figure 4.19 reveals network analysis figure of coupling of authors of the articles published on sickle cell anemia from India during 1991-2014. Totally 3 clusters were seen. They were interconnected with each other showing some interaction or collaboration among the groups. The biggest cluster showed 30 nodes. K. Ghosh showed the biggest and prominent node. R. Colah and R.B. Colah which represents the same author, revealed two independent nodes little smaller than K. Ghosh. D. Mohanty also occupied the prominent node with the same size as that of R.B. Colah. M.B. Mukherjee and A.H. Nadkarni occupied the smaller nodes with the same intensity. The second cluster depicted 11 nodes with R.S. Balgir showing the prominent node. R. Saxena revealed smaller sized node than that of R.S. Balgir. The third cluster showed 9 nodes with C.B.S. Dangi and M. Kaur occupied the prominent nodes.

Figure 4.20 revealed network analysis diagram of coupling of authors of the articles published on sickle cell anemia involving molecular biology aspects from India during 1991-2014. Totally 4 clusters were seen. Amongst these two clusters were interconnected while two small clusters were independent ones showing two nodes in each cluster. The biggest cluster showed 23 nodes with R.B. Colah and R.B. Gupta showing prominent nodes of equal size while A.H. Nadkarni showed a little smaller node than R.B. Colah. K. Ghosh and D. Mohanty showed the nodes of equal size but smaller than the above mentioned two authors. The second compact cluster revealed 22 nodes. R.B. Gupta who occupied a prominent node in the first cluster, also occupied a prominent node in this cluster. The other three prominent nodes were occupied in the second compact cluster by R.Nemer, F. Kutlar and T.H.J. Huisman, R.S. Tiwary occupied a small node in this cluster.

Figure 4.21 reveals trend of articles published on sickle cell anemia in top five journals from India during 1991-2014. 'Indian Journal of Pediatrics' published maximum number of articles during this period.

Figure 4.22 depicts trend of articles published on sickle cell anemia involving molecular biology aspects from India during 1991-2014.

Table 4.32: Productivity pattern of authors who published articles on six genetic disorders from India during 1991-2014 without any data related to molecular biology.

Disease	Arti-cles	Sour-ces	Period	Average citations per article	Authors total	Single author articles	Multi author articles	Authors per article	Annual % growth rate
DS	696	367	1991-2014	23.15	2255	47 (6.75)	649 (93.25)	3.24	12.55
vWD	180	105	1993-2014	24.69	595	12 (6.67)	168 (93.33)	3.31	11.32
SCA	436	208	1991-2014	12.83	1205	43 (9.86)	393 (90.14)	2.76	12.07
HpA	459	180	1991-2014	15.94	1182	28 (6.10)	431 (93.90)	2.58	13.79
G6PD	109	73	1992-2014	12.77	339	5 (4.59)	104 (95.41)	3.11	2.58
HpB	187	98	1991-2014	16.06	555	14 (7.49)	173 (92.51)	2.97	8.83

Source: SCOPUS database

Figures in brackets indicate percentage values

DS - Down syndrome
vWD - von Willebrand disease
SCA - Sickle cell anemia
HpA - Hemophilia A
G6PD - Glucose 6 Phosphate Dehydrogenase (G6PD) deficiency
HpB - Hemophilia B

Table 4.33: Productivity pattern of authors who published articles on six genetic disorders from India during 1991-2014 with data related to molecular biology.

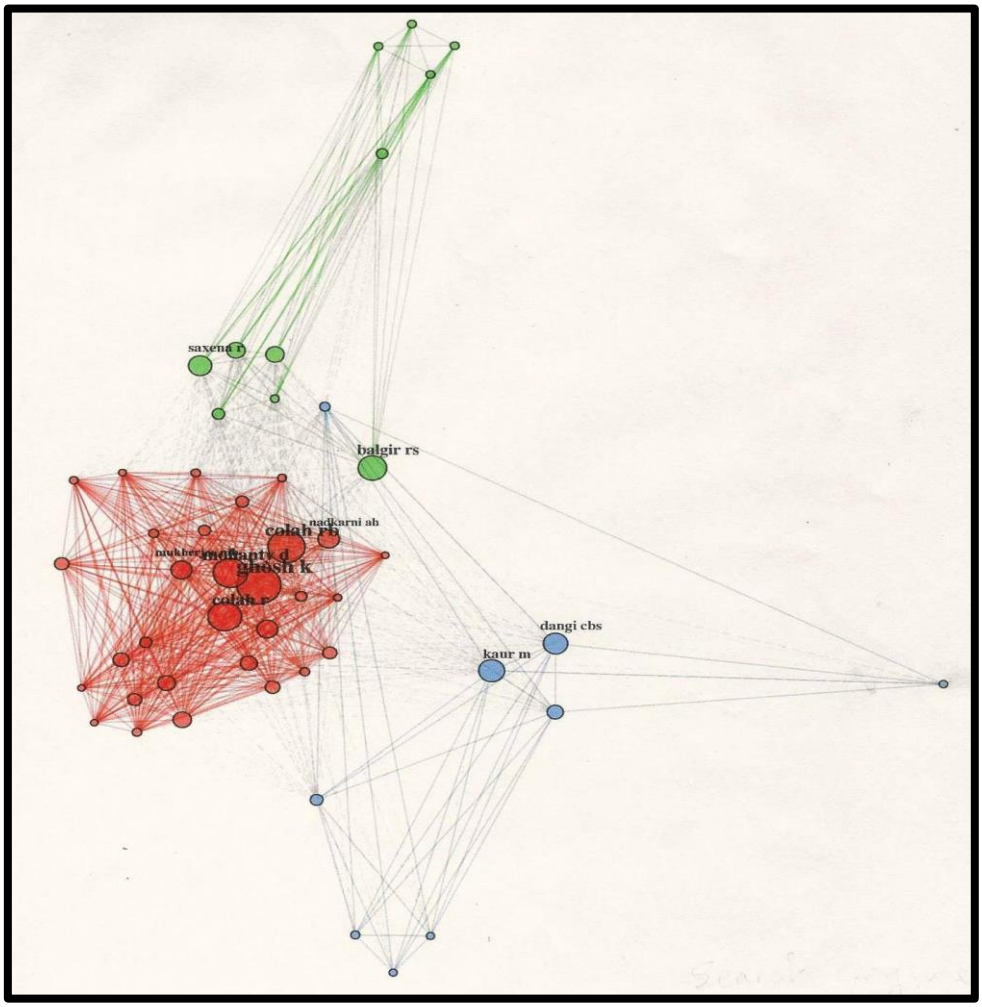
Disease	Arti-cles	Sour-ces	Period	Average citation per article	Authors Total	Single author articles	Multi author articles	Authors per article	Annual % growth rate
DS	77	61	1991-2014	23.75	303	7 (9.09)	70 (90.91)	3.94	11.64
vWD	13	09	2003-2014	17.31	30	0 (0.00)	13 (100.00)	2.31	6.50
SCA	40	34	1991-2014	24.57	145	5 (12.50)	35 (87.50)	3.62	4.50
HpA	78	46	1998-2014	13.86	218	3 (3.84)	75 (96.66)	2.79	1.78
G6PD	14	12	1992-2014	9.21	50	1 (7.14)	13 (92.86)	3.57	0.00
HpB	43	28	1998-2014	17.07	133	1 (2.33)	42 (97.67)	3.09	4.50

Source: SCOPUS database

Figures in brackets indicate percentage values

DS = Down syndrome
vWD = von Willebrand disease
SCA = Sickle cell anemia
HpA = Hemophilia A
G6PD = Glucose 6 Phosphate Dehydrogenase (G6PD) deficiency
HpB = Hemophilia B

Figure 4.19: Coupling or collaboration of authors of the articles published Sickle cell anemia without molecular biology from India during 1991-2014.



Source: SCOPUS database

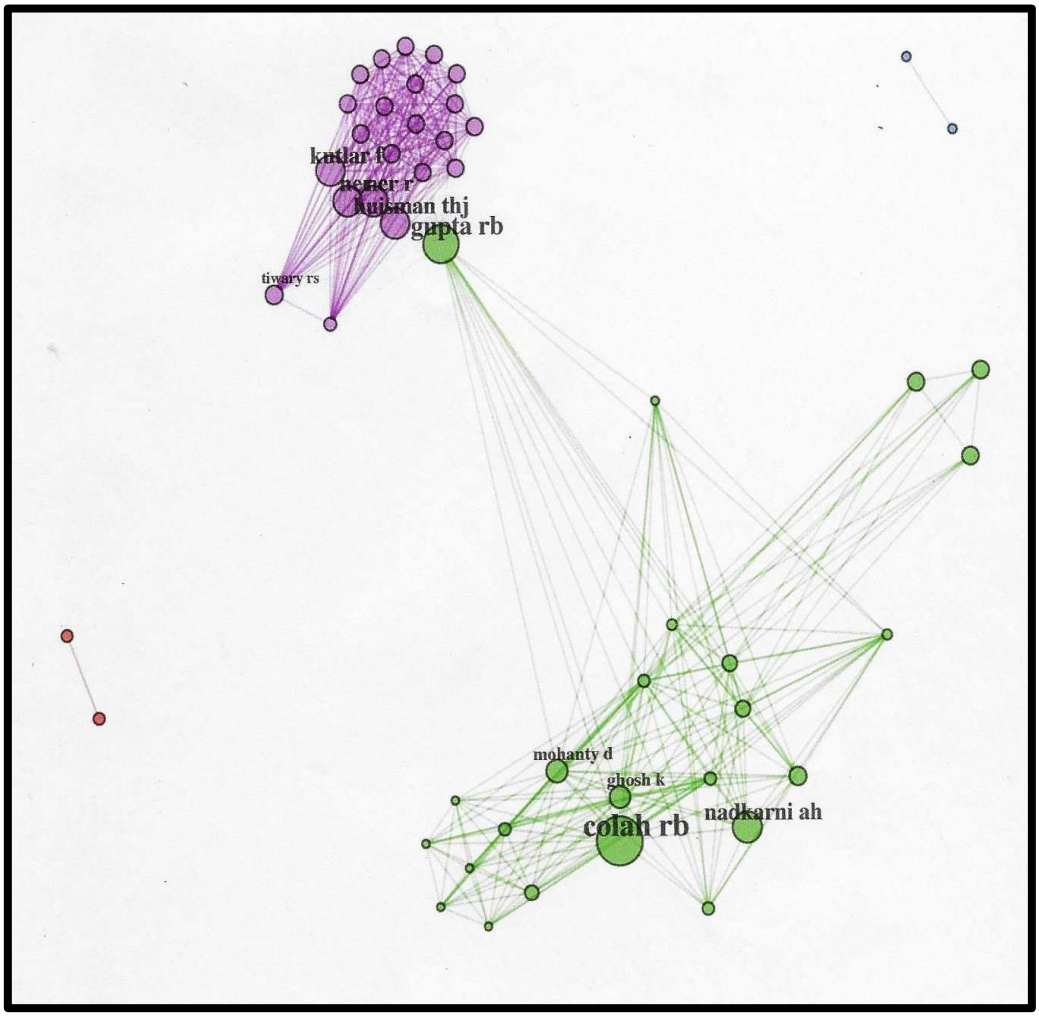
Observation - Interconnected three clusters which are showing some collaboration are seen.

Cluster 1-Total 30 nodes **Main nodes – K. Ghosh, R. Colah, R. B. Colah, A.H. Nadkarni, M.Mukherjee.**

Cluster 2- Total 11 nodes **Main nodes- R. S. Balgir, R.Saxena**

Cluster 3- Total 9 nodes **Main nodes- M. Kaur, C.B. S. Dangi**

Figure 4.20: Coupling or collaboration of authors of the articles published on Sickle cell anemia with molecular biology from India during 1991-2014.



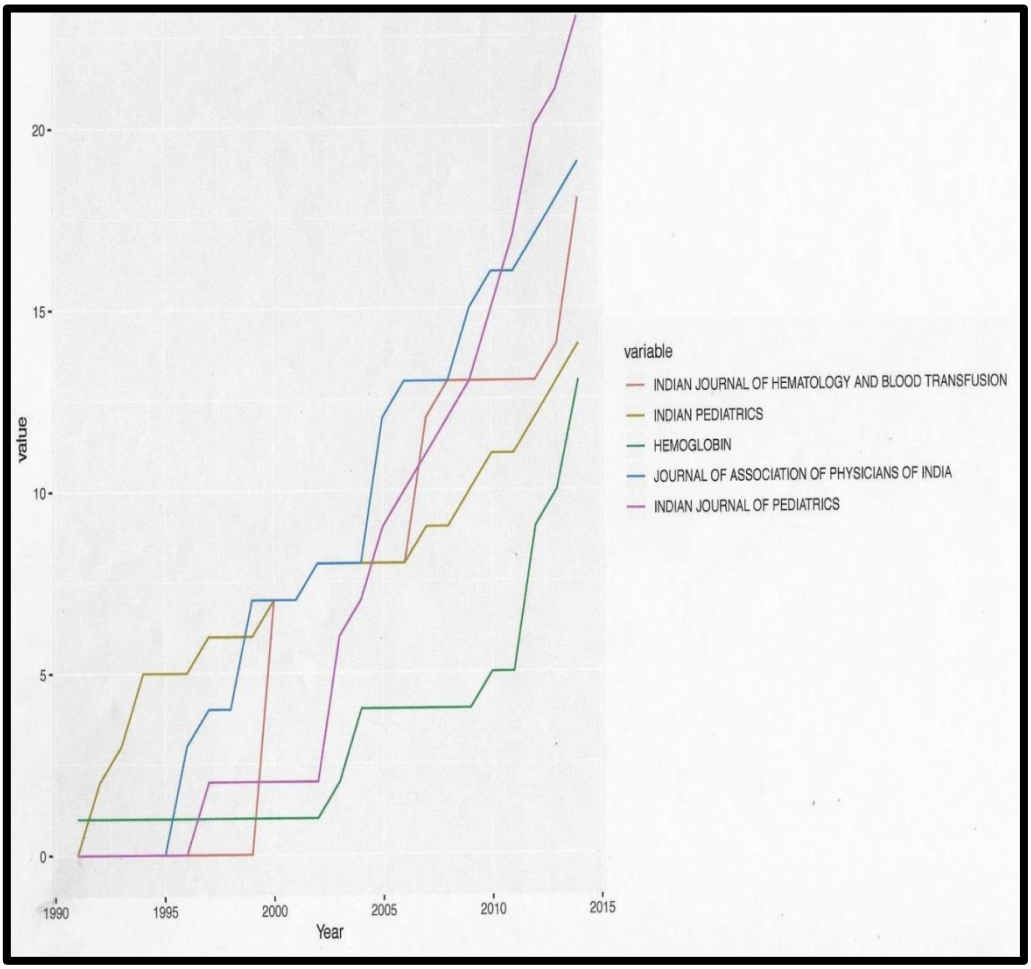
Source: SCOPUS database

Observation- Two interconnected clusters of which the first cluster (purple) very compact one showing good collaboration and two independent small clusters are seen.

Cluster 1 – Total 23 nodes **Main nodes - R. B. Colah, R.B. Gupta, A.H. Nadkarni, K. Ghosh, D. Mohanty.**

Cluster 2 – Total 22 nodes **Main nodes – R.B.Gupta,T.H.J. Huisman, F.Kutlar, R. Nemer.**

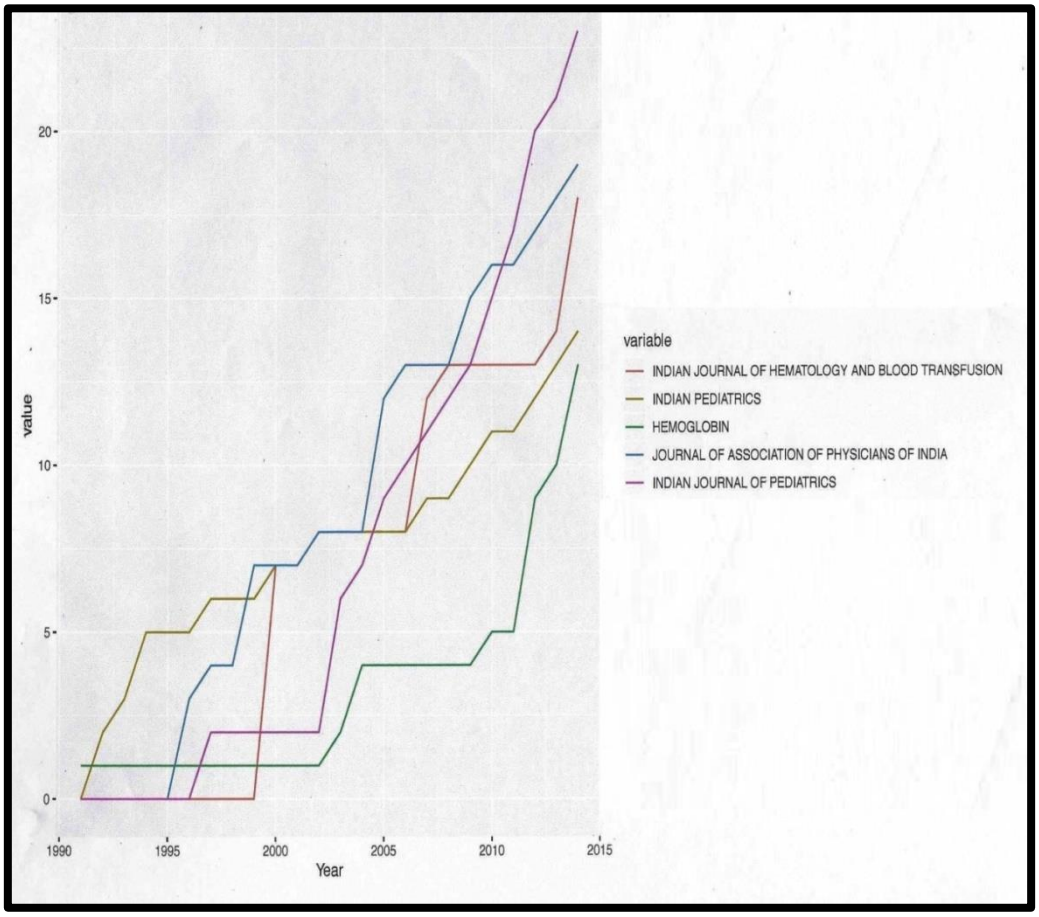
Figure 4.21: Top five journals showing trend of articles published on Sickle cell anemia without molecular biology from India during 1991 -2014 .



Source: SCOPUS database

Observation- This figure shows increasing trend of articles published on Sickle cell anemia without molecular biology from India during 1991-2014.

Figure 4.22: Top five journals showing trend of articles published on Sickle cell anemia with molecular biology from India during 1991 -2014.



Source: SCOPUS database

Observation- This figure shows increasing trend of articles published on Sickle cell anemia with molecular biology in above mentioned top five journals from India during 1991-2014.

Figure 4.23 presents average article citations per year on articles published on sickle cell anemia from India during 1991-2014. A sharp spike showing increase in number of articles published during 2011-2013 was seen.

Figure 4.24 presents average article citations per year on sickle cell anemia involving molecular biology aspects from India during 1991-2014. Here the sharp spike was seen during 2003-2005.

Average total citations per year on articles published on sickle cell anemia from India during 1991-2014 are shown in Figure 4.25. Sharp spikes showing increase in total citations were seen during the years 1991, 1995, 2005 and 2013.

Average total citations per year on articles published on sickle cell anemia involving molecular biology aspects are shown in Figure 4.26. The spike showing increase in total citations was seen during the year 2005.

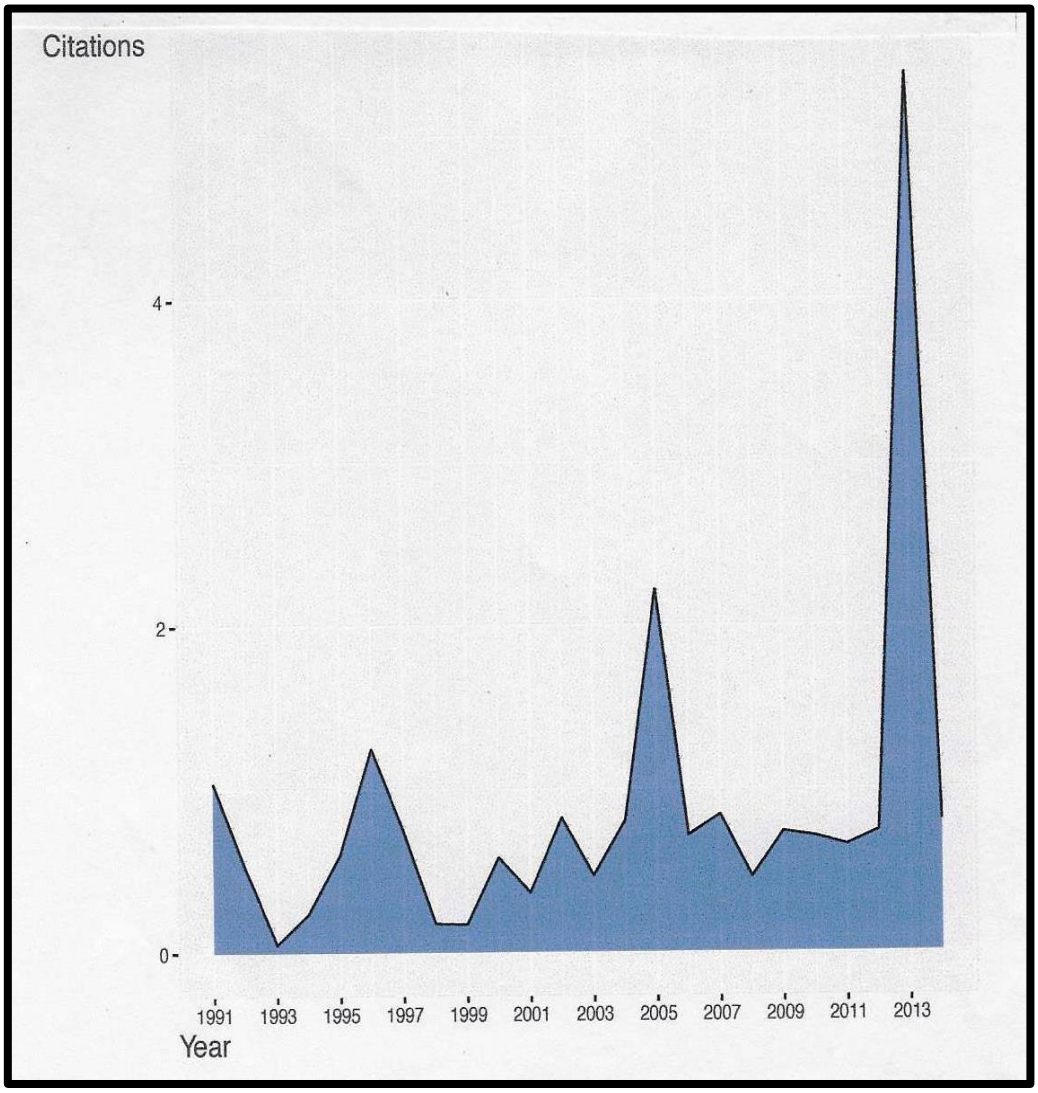
Table 4.34 presents a list of most productive top ten authors expressed on the basis of fractionalized published articles on sickle cell anemia from India during 1991-2014. R.S. Balgir ranked first in this table with total weightage of 18.67 of the total number of articles published by him on this topic.

Table 4.34 reveals a list of to ten articles published on sickle cell anemia from India during 1991-2014 and receiving maximum citations. An article by Simonneau et al (2013) published in the journal 'American College of Cardiology' ranked first in this table receiving 1,558 total citations during this period.

Table 4.36 presents a list of the most productive top ten authors expressed on the basis of fractionalized published articles on sickle cell anemia involving molecular biology aspects from India during 1991-2014. R.B. Colah ranked first in this table with total weightage of 12.64 of the total number of articles published by her on this topic.

Table 4.37 illustrates list of top 10 articles published on sickle cell anemia involving molecular biology aspects from India during 1991-2014 and receiving maximum citations. An article by Kumar et al (2005) published in the journal 'Toxicology letters' ranked first in this table receiving 437 total citations during the above mentioned period.

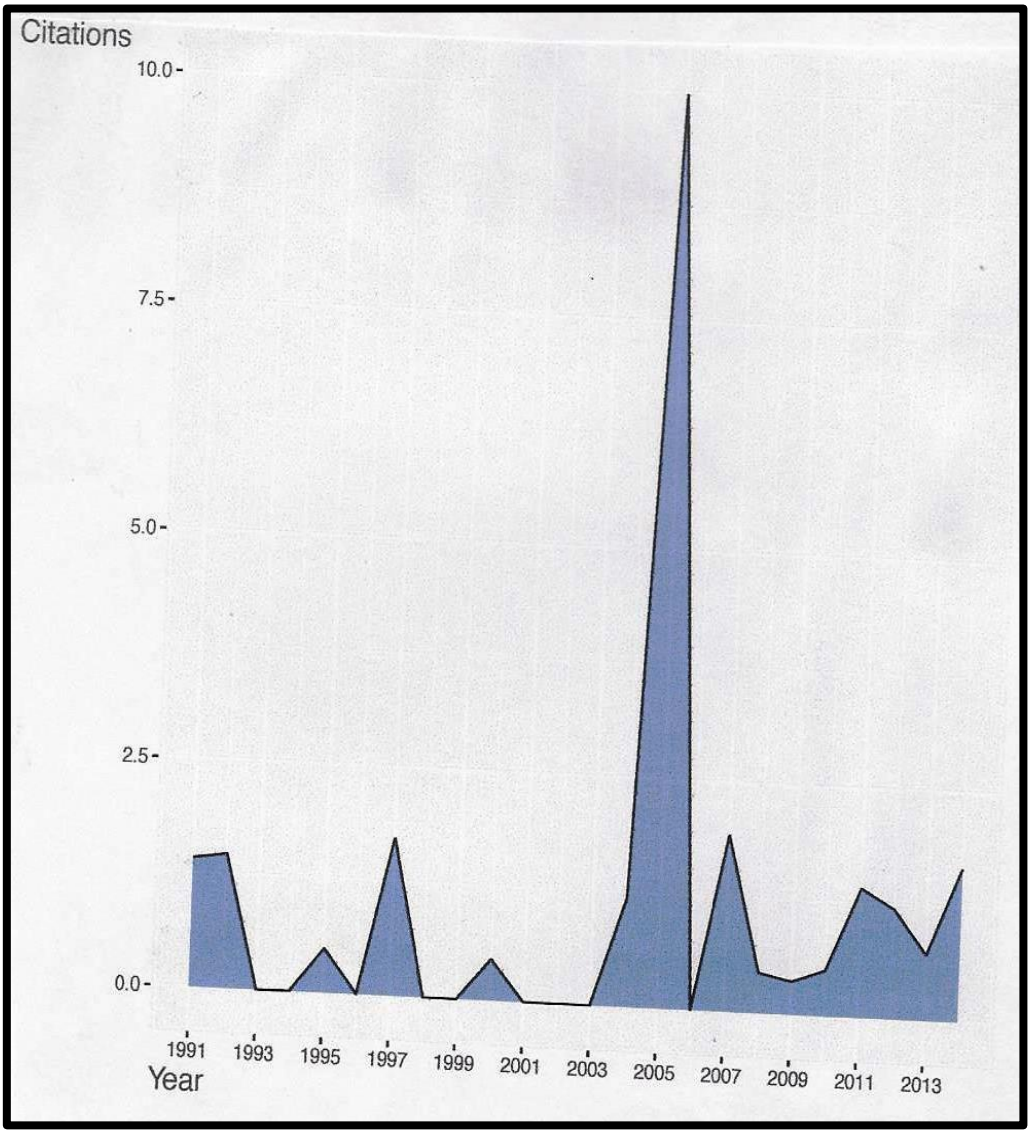
Figure 4.23: Average article citations per year received on articles published on Sickle cell anemia without molecular biology from India 1991 -2014.



Source: SCOPUS database

Observation- This figure shows maximum articles were published on sickle cell anemia without molecular biology aspects were cited during 2013 followed by those cited during 2005 from India during 1991-2014.

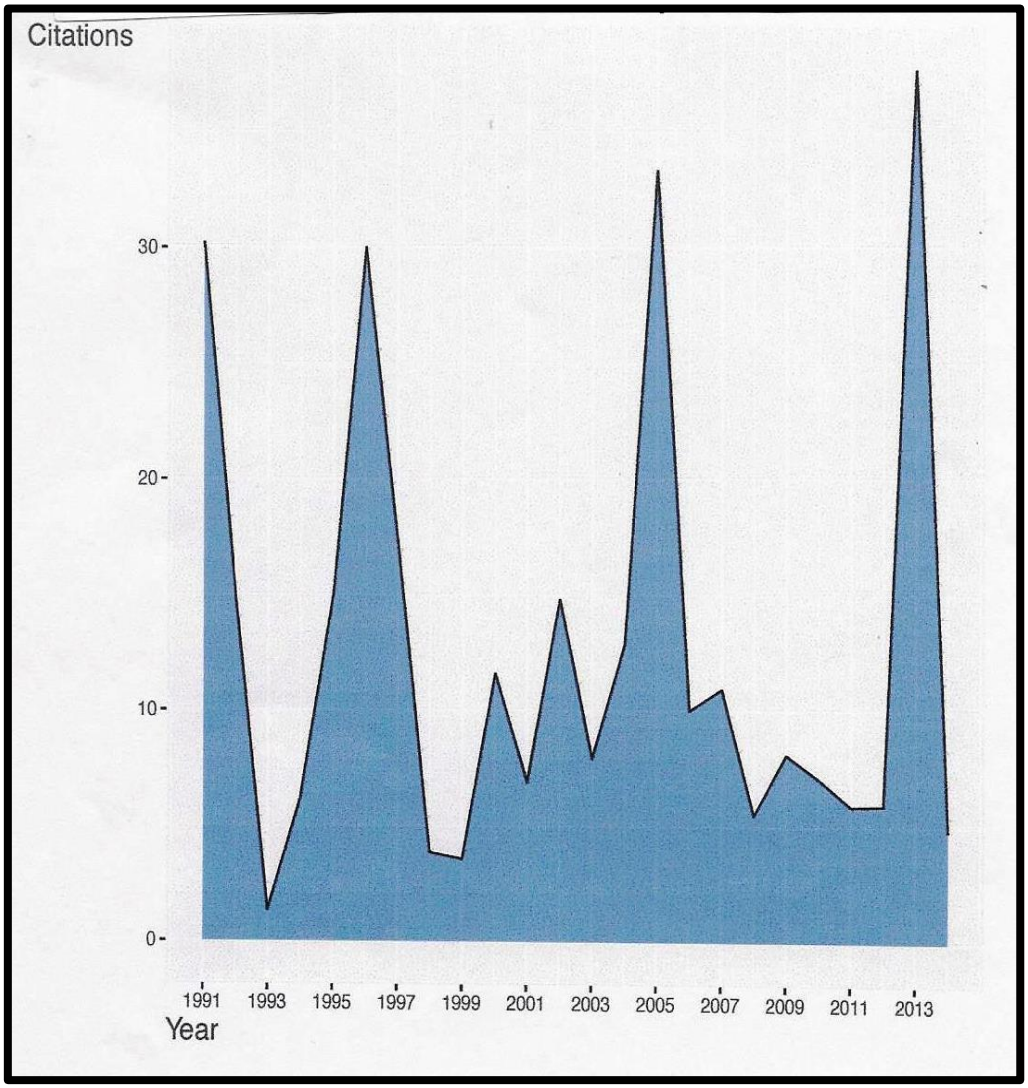
Figure 4.24: Average article citations per year received on articles published on Sickle cell anemia with molecular biology from India during 1991 -2014.



Source: SCOPUS database

Observation- Maximum articles were cited during 2005 from India on sickle cell anemia with molecular biology during 1991-2014.

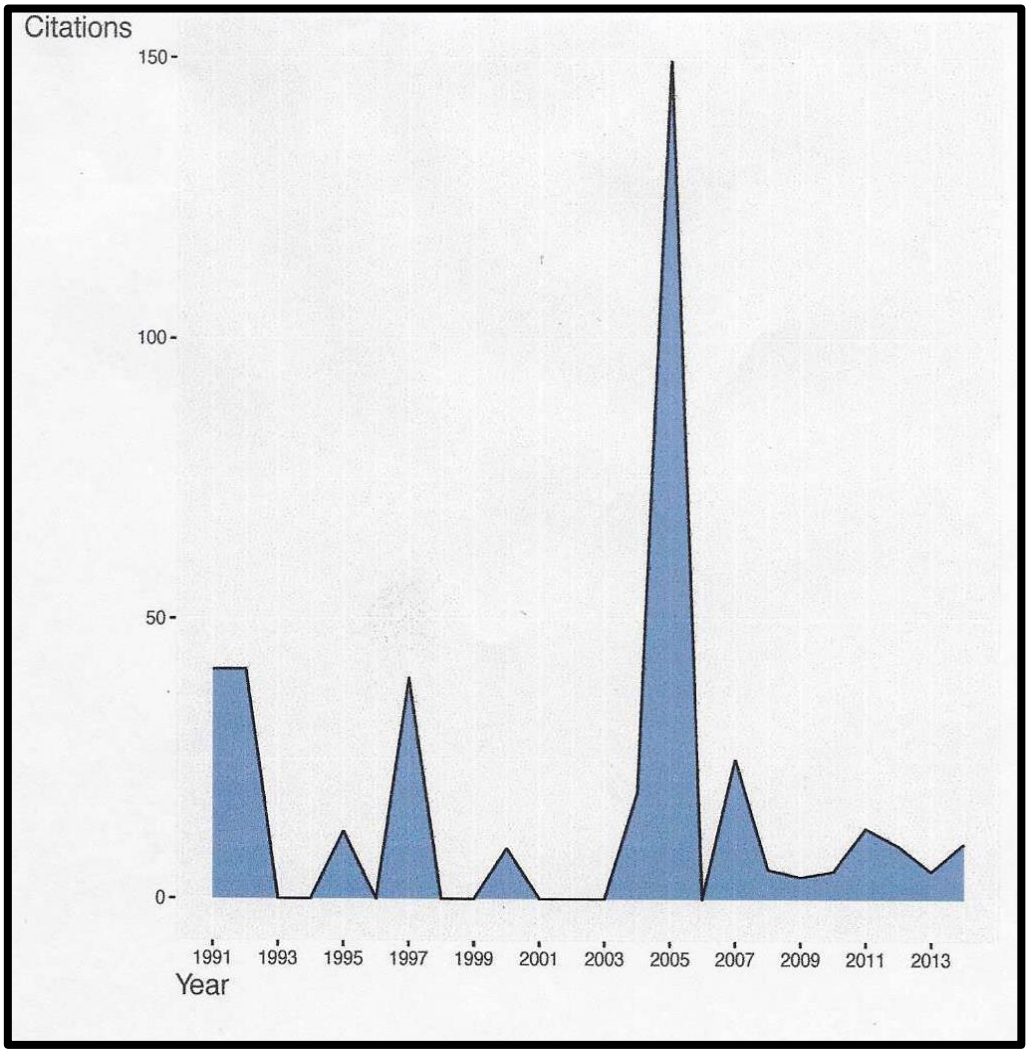
Figure 4.25: Average total citations per year received on articles published on Sickle cell anemia without molecular biology from India during 1991 -2014.



Source: SCOPUS database

Observation- Maximum total citations were received for the articles during 2013 followed by those received during 2005,1995 and 1991 from India published on sickle cell anemia without molecular biology during 1991-2014.

Figure 4.26: Average total citations per year received on articles published on Sickle cell anemia with molecular biology from India during 1991 -2014.



Source: SCOPUS database

Observation- Maximum total citations were received for the articles during 2005 from India published on sickle cell anemia with molecular biology during 1991-2014.

Table 4.34: Most productive top ten authors expressed as fractionalized published articles on Sickle cell anemia without molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Balgir RS	18.67
2	Kar BC	4.95
3.	Mohanty D	4.45
4.	Ghosh K	4.00
5.	Colah RB	3.87
6.	Saxena R	3.48
7.	Mukherjee MB	3.35
8.	Pandey S	3.10
9.	Dash BP	2.79
10.	Shah A	2.70

Source : SCOPUS database

Explanation- Contribution of top ten most productive authors in published articles on sickle cell anemia without molecular biology from India during 1991-2014.

Table 4.35: Top ten articles published on Sickle cell anemia without molecular biology from India during 1991-2014 receiving maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Simonneau G, 2013, J Am Coll Cardiol	1,558	194.75
2	Kumar S, 2005, Toxicol Lett	437	27.31
3	Pandian JD, 2007, Int J Stroke	99	7.07
4	Balgir RS, 2000, Current Science	84	4.00
5	Balgir RS, 1996, J Assoc Phys Ind	78	3.12
6	Ner C, 1992, Human Genetics	76	2.62
7	Mukherjee MB, 1997, Am J Hematol	69	2.88
8	Kalantri SP, 2006, Natl Med J Ind	62	4.13
9	Colah RB, 2007, Ind J Paediatr	53	3.79
10	Mohanty D, 2002, Curr Opin Hematol	53	2.79

Source: SCOPUS database

J Am Coll Cardiol - Journal of American College of Cardiology
Toxicol Lett - Toxicology Letters
Int J Stroke - International Journal of Stroke
J Assoc Phy Ind - Journal of Association of Physicians of India
Am J Hematol - American Journal of Hematology
Natl Med J Ind - National Medical Journal of India
Ind J Pediatr - Indian Journal of Paediatrics
Curr Opin Hematol - Current Opinion in Hematology

Table 4.36: Most productive top ten authors expressed on the basis of fractionalized published articles on Sickle cell anemia with molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1	Colah RB	12.64
2	Gupta RB	8.92
3	Nadkarni AH	5.21
4	Huisman THJ	3.98
5	Kutlar F	3.50
6	Mohanty D	3.41
7	Ghosh K	3.15
8	Mukherjee MB	2.98
9	Tiwary RS	2.98
10.	Balgir RS	2.90

Source : SCOPUS database

Explanation- Contribution of top ten most productive authors in publishing on Sickle cell anemia with molecular biology from India during 1991-2014.

Table 4.37: Top ten articles published on Sickle cell anemia with molecular biology from India during 1991 -2014 received maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Kumar S, 2005, Toxicol Lett	437	27.31
2	Ner C 1992, Human Genetics	76	2.62
3	Mukherjee MB, 1997,Am J Hematol	69	2.88
4	Colah RB, 2007, Ind J Paediatr	53	3.79
5	Gupta RB, 1991, Hemoglobin	41	1.37
6	Ramlingam S, 2014, Curr Gene Ther	33	4.71
7	Colah RB, 2011, Ind J Med Res	25	2.50
8	Mukherjee MB, 2004, Hemoglobin	19	1.12
9	Upadhye DS, 2012, J Clin Patho	17	1.89
10	Balgir R, Ind J Med Sc	16	1.23

Source : SCOPUS database

Toxicol Lett - Toxicology Letters
Am J Hemat - American Journal of Hematology
Ind J Padiatr - Indian Journal of Paediatrics
Curr Gene Ther - Current Gene Therapy
Ind J Med Res - Indian Journal of Medical Research
J Clin Patho - Journal of Clinical Pathology
Ind J Med Sc - Indian Journal of Medical Science

4.2.2 Down syndrome

Figure 4.27 depicts the network analysis diagram of coupling of authors of the articles published on Down syndrome from India during 1991-2014. A big compact cluster with 49 nodes was seen. All the nodes were of the same size and with equal prominence.

Figure 4.28 reveals the network analysis diagram of coupling of authors of the articles published on Down syndrome involving molecular biology aspects from India during 1991-2014. Totally 8 clusters were seen. The first compact cluster showed 13 nodes with equal prominence. The second cluster revealed ten nodes again all were showing equal prominence. The third cluster showed nine nodes. The fourth cluster was presented with six nodes out of which one node was bit prominent than others. The fifth and the sixth cluster depicted five nodes in each one; while the remaining two revealed only single independent node in each one.

Figure 4.29 shows trend of articles published on Down syndrome in top five journals from India during 1991-2014. Since this is a disease seen mainly in young children, many articles on this subject were published in 'Indian Pediatrics' and 'Indian Journal of Pediatrics' during the above mentioned time period.

Figure 4.30 represents trend of articles published on Down syndrome involving molecular biology aspects from India during 1991-2014.

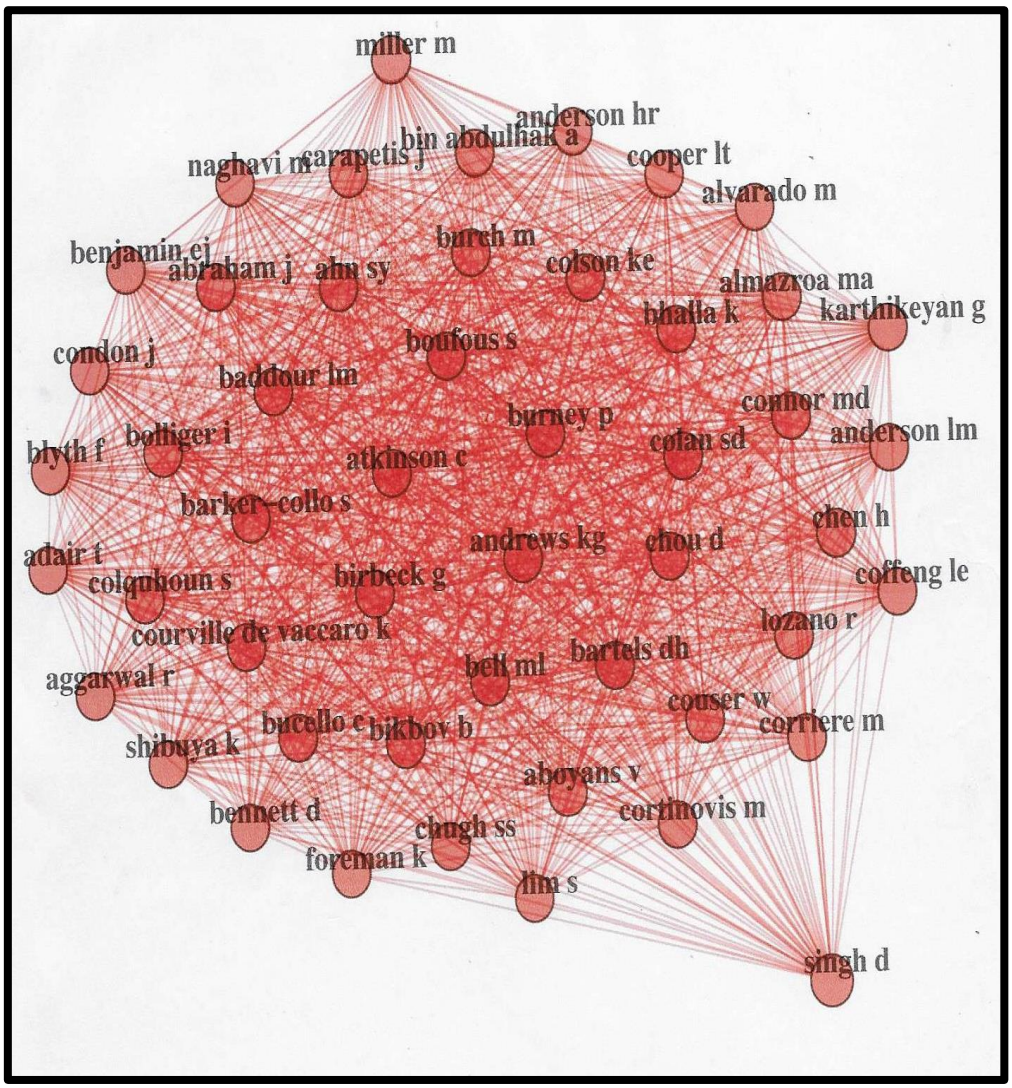
Figure 4.31 describes average article citations per year on articles published on Down syndrome from India during 1991-2014. A sharp spike showing increase in average citations was seen in the year 2011.

Figure 4.32 depicts average article citations per year on articles published on Down syndrome involving molecular biology aspects from India during 1991-2014. During 1999-2002 no citations were recorded; however sharp peaks were seen during 2003 and 2013.

Figure 4.33 shows average total citations per year on articles published on Down syndrome from India during 1991-2014. A sharp spike showing increase in average total citations was seen during the year 2013.

Figure 4.34 reveals average total citations per year on articles published on Down syndrome involving molecular biology aspects from India during 1991-2014. No citations were recorded during the years 1991-2002. A sharp spike was seen during the year 2003.

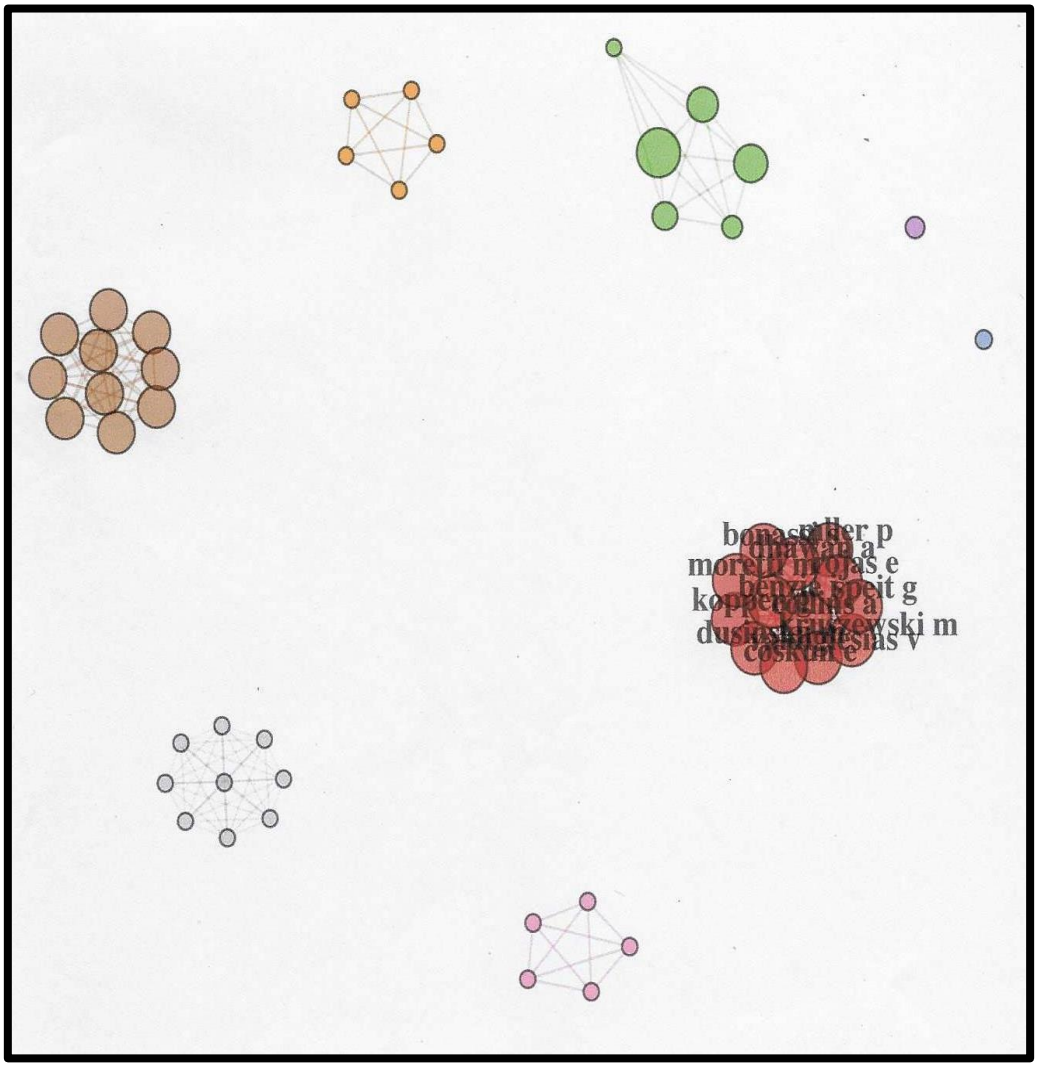
Figure 4.27: Coupling or Collaboration of authors of the articles published on Down syndrome without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- A compact Cluster with 49 nodes of same size and equal prominence.

Figure 4.28: Coupling or Collaboration of authors of the articles published on Down syndrome with molecular biology from India during 1991 -2014.

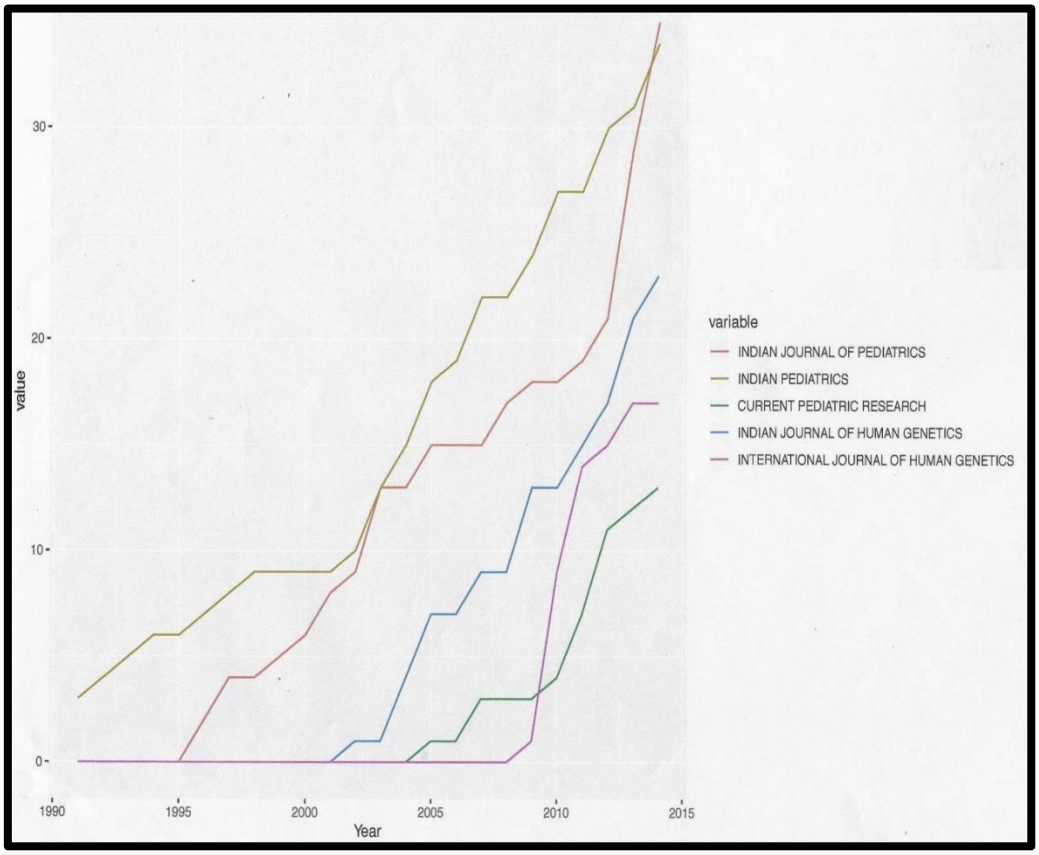


Source : SCOPUS database

Observation- Totally 8 clusters are seen

The biggest cluster with 13 nodes, equal prominence
The second biggest cluster with 10 nodes, equal prominence
The third cluster with 9 nodes, equal prominence
Remaining clusters with 1 – 6 nodes

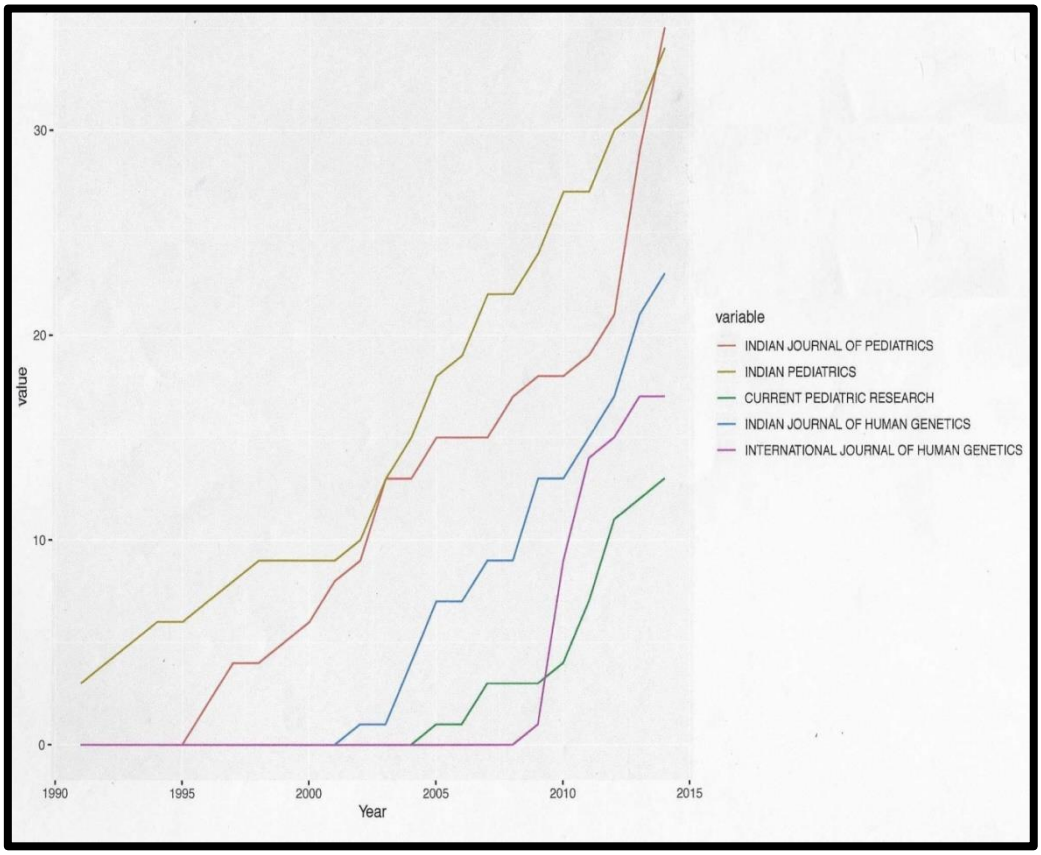
Figure 4.29: Top five journals showing trend of articles published on Down syndrome without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- This figure shows rising trend of articles published on Down syndrome without molecular biology from India during 1991-2014 in the above mentioned five journals.

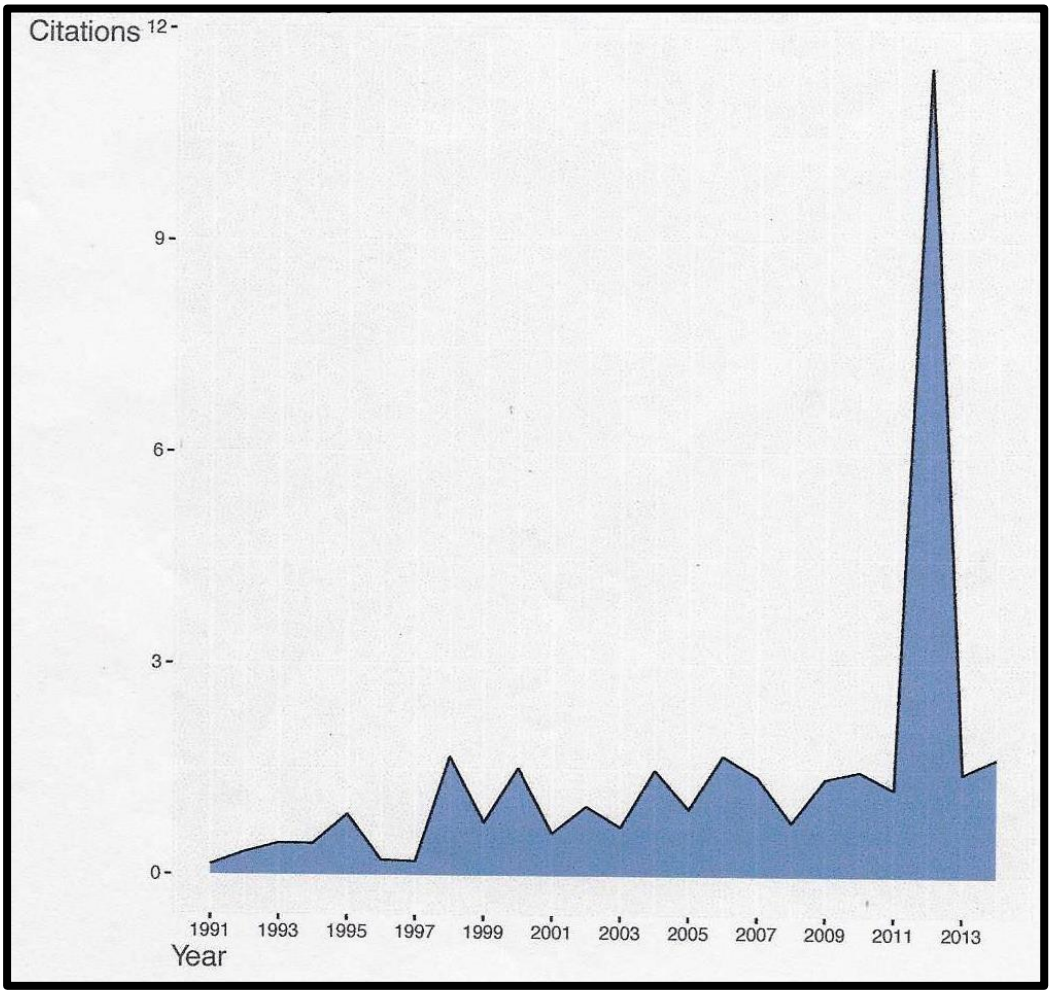
Figure 4.30: Top five journals showing trend of articles published on Down syndrome with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- This figure shows rising trend of articles published on Down syndrome with molecular biology from India during 1991-2014 in the above above mentioned five journals.

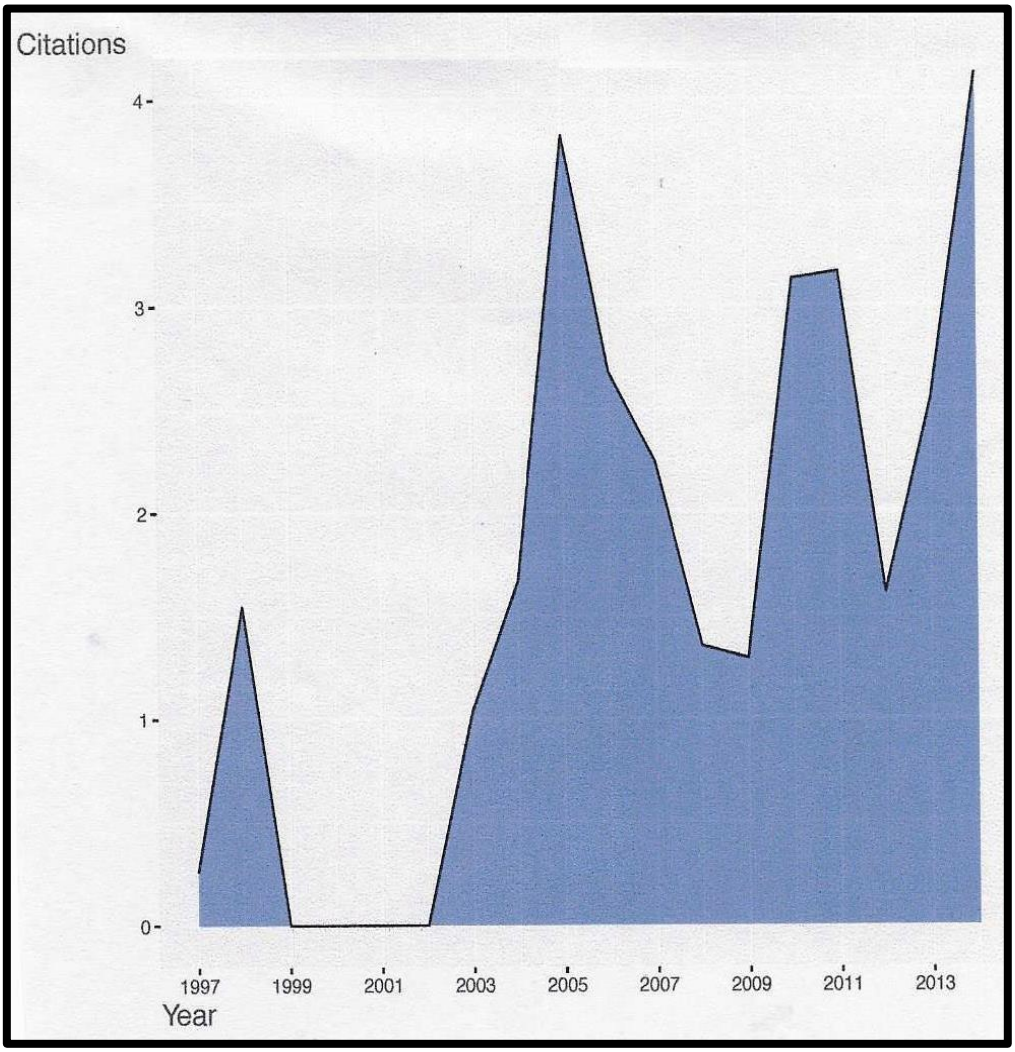
Figure 4.31: Average article citations per year received on articles published on Down syndrome without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- Maximum articles were cited during 2013 published on Down syndrome without molecular biology from India during 1991-2014.

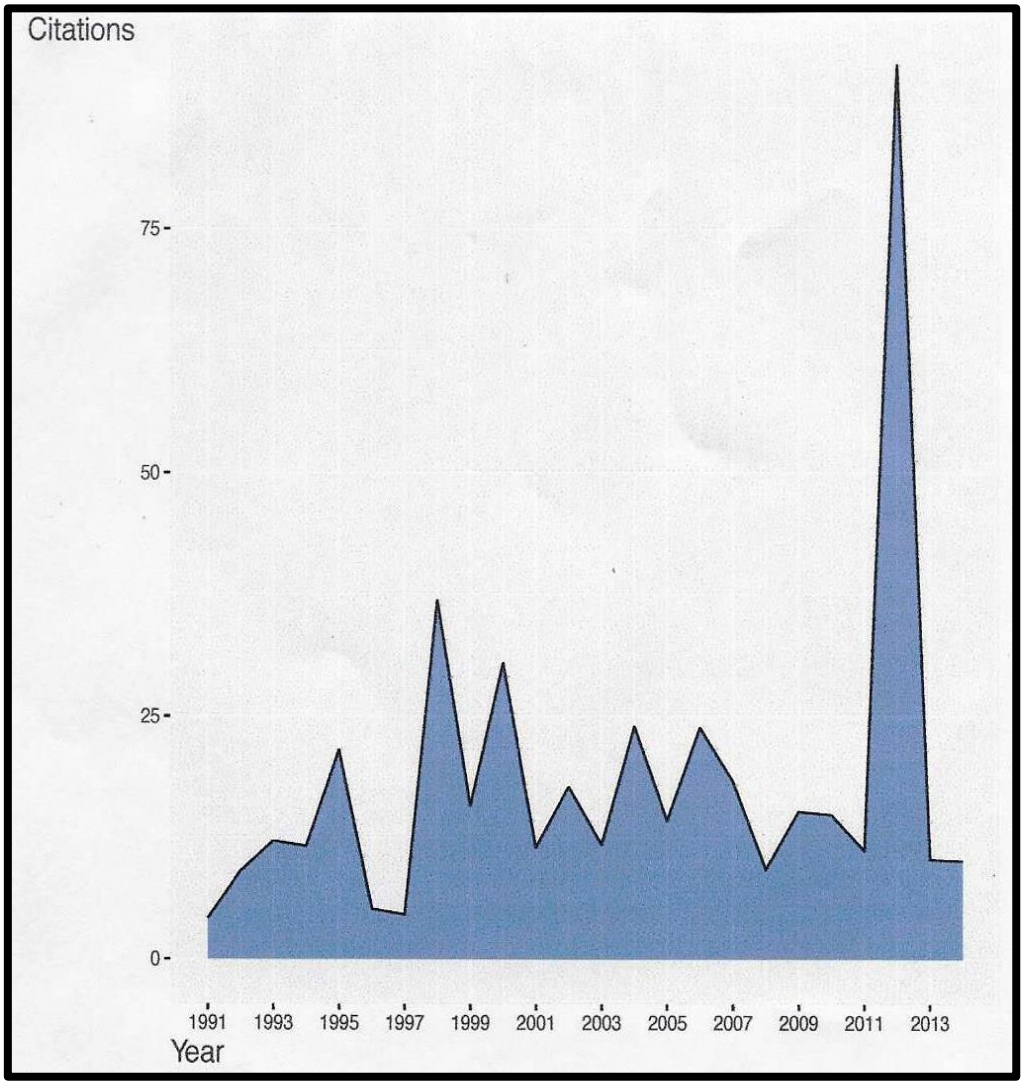
Figure 4.32: Average total citations per year received on articles published on Down syndrome with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum total citations were received during 2013 followed by those received during 2003 for articles published on Down syndrome with molecular biology from India during 1991-2014.No citations were received during the years 1999 – 2003.

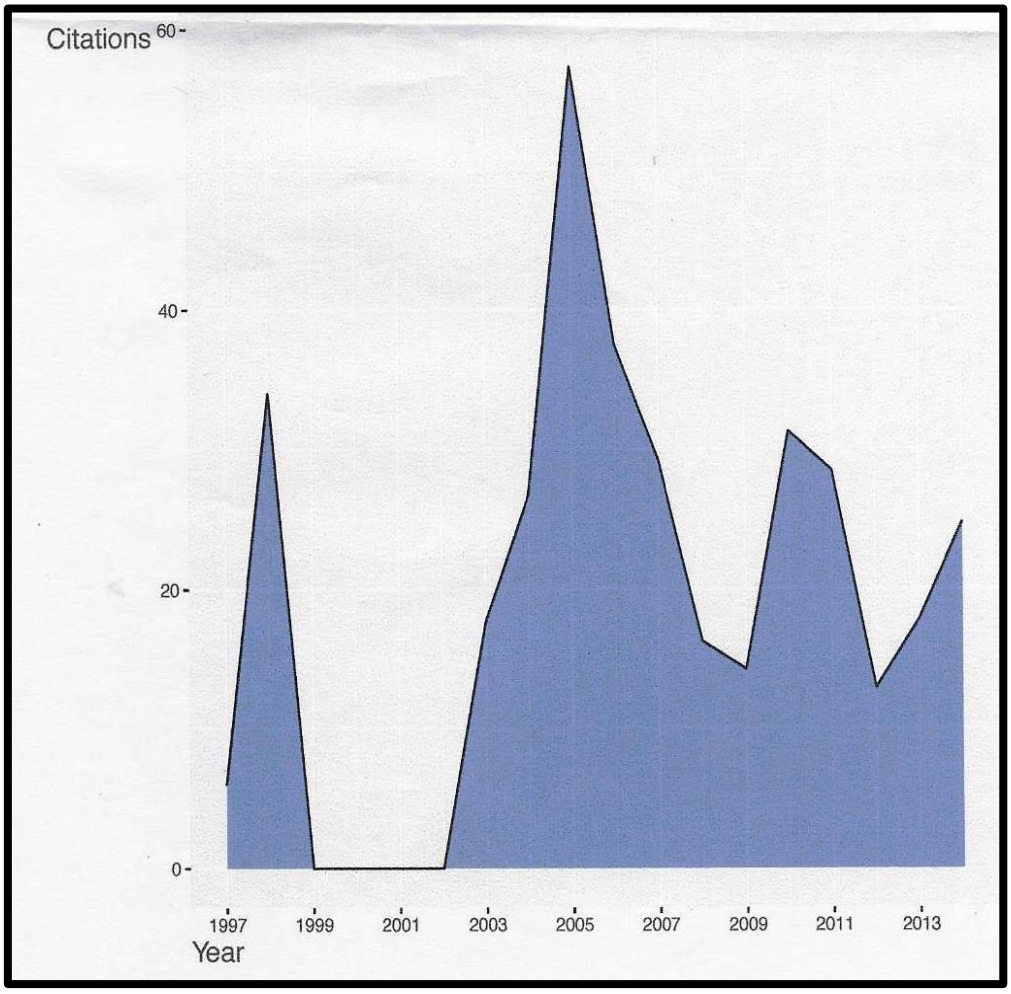
Figure 4.33: Average total citations per year received on articles published on Down syndrome without molecular biology from India during 1991-2014.



Source : SCOPUS database

Observation- Maximum total citations for articles published on Down syndrome without molecular biology from India during 1991-2014 was received during 2013.

Figure 4.34: Average total citations per year received on articles published on Down syndrome with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- Maximum total citations for articles published on Down syndrome with molecular biology from India during 1991-2014 was received during the year 2005. During the years 1999 – 2003 no articles received any citations.

Table 4.38 illustrates the most productive top ten authors expressed on the basis of fractionalized published articles on Down Syndrome from India during 1991-2014. M. Kabra was on the top position with weightage of 4.89 of the total articles published by her on Down syndrome during this period.

Table 4.39 reveals the list of top ten articles published on Down syndrome from India during 1991-2014 and receiving maximum citations. An article by Lezano et al (2012) published in the journal 'Lancet' received 7,128 total citations during this period and ranked first.

Table 4.40 depicts the list of most productive top ten authors expressed on the basis of fractionalized published articles on Down syndrome involving molecular biology aspects from India during 1991-2014. K.S. Rao ranked first with the weightage of 1.5 of the total articles published by him during the above mentioned period.

Table 4.41 reveals the list of top ten articles published on Down syndrome involving molecular biology aspects from India during 1991-2014 and receiving maximum citations. An article by Nazki et al (2014) published in the journal 'Gene' received 144 total citations during this period and ranked first.

4.2.3 G6PD deficiency

Figure 4.35 depicts the network analysis diagram of coupling of authors of the articles published on G6PD deficiency from India during 1991-2014. Two clusters were seen. The first cluster was a major one and very compact cluster with 44 nodes. Amongst these, K. Ghosh occupied the most prominent node. R.B. Colah and D. Mohanty also occupied the prominent nodes but smaller than that of K. Ghosh. M.B. Mukherjee was the fourth author who occupied a prominent node. His node was smaller than the above mentioned three authors but bigger than rest of nodes. The second cluster was a small cluster which revealed seven nodes with equal prominence.

Figure 4.36 reveals the network analysis of coupling of the authors of the articles published on G6PD deficiency involving molecular biology aspects from India during 1991-2014. Totally 5 clusters were seen. The biggest cluster revealed 14 nodes with two prominent nodes. . The most prominent node was shown by R.B. Colah followed by K. Ghosh. The second cluster showed 10 nodes with

prominent nodes occupied by M.B. Mukherjee followed by D. Mohanty. The third, fourth and fifth clusters revealed 9, 7 and 5 nodes respectively with equal prominence

Figure 4.37 reveals the trend of articles published from India in top five journals on G6PD deficiency during 1991-2014. Maximum articles were published in 'Indian Journal of Pediatrics' followed by the journal 'Anthropologist'.

Figure 4.38 describes the trend of articles published from India in top five journals on G6PD deficiency involving molecular biology aspects during 1991-2014. Very small number of articles were published under this category. Maximum number of articles were published in the journal 'Annals of Human Biology' from this category during the above mentioned period.

Figure 4.39 shows average article citations per year received on articles published on G6PD deficiency from India during 1991-2014. A sharp spike indicating increase in total citations was seen during the period 1998-2000.

Figure 4.40 demonstrates average article citations per year received on articles published on G6PD deficiency involving molecular biology aspects from India during 1991-2014. Here the sharp spike was seen during the period 2008-2010.

Figure 4.41 depicts average total citations per year received on articles published on G6PD deficiency from India during 1991-2014. A sharp spike indicating increase in total citations per year was seen during the period 1998-2000.

Figure 4.42 represents average total citations per year received on articles published on G6PD deficiency involving molecular biology aspects from India during 1991-2014. No citations were received during the four year period (1996-2000) while in the remaining years five sharp peaks with more than 10 citations per year were seen. Amongst these, the biggest peak was seen during the period 2008-2010.

Table 4.42 presents the most productive top ten authors who published on G6PD deficiency from India during 1991-2014. R.S. Balgir ranked first with the total weightage of 9.00 of the total articles published by him during this period.

Table 4.43 reveals top ten articles published on G6PD deficiency from India during 1991-2014 and receiving maximum citations. An article published by Beutler (2007) in the journal 'American Journal of Tropical Medicine and Hygiene' ranked first receiving 138 total citations during this period.

Table 4.38: Most productive top ten authors expressed on the basis of fractionalized published articles on Down syndrome without molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles Fractionalized
1	Kabra M	4.89
2	Rajangam S	4.59
3	Ramchandra Rao K	4.11
4	Sharma S	3.53
5	Verma IC	3.00
6	Sinha S	2.94
7	Vishnu Bhat B	2.91
8	Ghosh S	2.83
9	Gupta S	2.81
10.	Kumar A	2.63

Source : SCOPUS database

Explanation- Contribution of top ten most productive authors in articles on Down syndrome without molecular biology from India during 1991-2014.

Table 4.39: Top ten articles published on Down syndrome without molecular biology from India during 1991-2014 receiving maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Lezano R, 2012 Lancet	7,128	792.00
2	Goel A, 1998, J Neurosurg	237	10.30
3	Walker AK, 2010, Genes Dev	199	18.09
4	Graessler J, 2009, PLOS one	198	16.50
5	Noor R, 2002, Med Sci Monit	1,922	10.11
6	Nazki FH, 2014, Gene	144	20.57
7	Gupta V, 2010, Protein Science	140	12.73
8	Collins A, 2014, Mutat Res Rev	122	17.43
9	Mohankumar K, 2006, Virus Research	188	7.87
10	Narayanan S. 2005, FEBS Lett	112	7.00

Source : SCOPUS database

J Neurosurg - Journal of Neurosurgery
Genes Dev - Genes and Development
Med Sci Monit - Medical Science Monitor
Mutat Res Rev - Mutation Research Review
FEBS Lett - Federation of European Biochemical Societies Letters Journal

Table 4.40: Most productive top ten authors expressed on the basis of fractionalized published articles on Down syndrome with molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles Fractionalized
1	Rao KS	1.50
2	Nandgopal K	1.42
3	Sinha S	1.30
4	Datta S	1.22
5	Banerjee D	1.17
6	Mukhopadhyay K	1.12
7	Agarwal R	1.00
8	Bhat M	1.00
9	Ranhotra HS	1.00
10.	Saha B	1.00

Source : SCOPUS database

Explanaation- Contribution of top ten most productive authors in published on Down syndrome with molecular biology from India during 1991-2014.

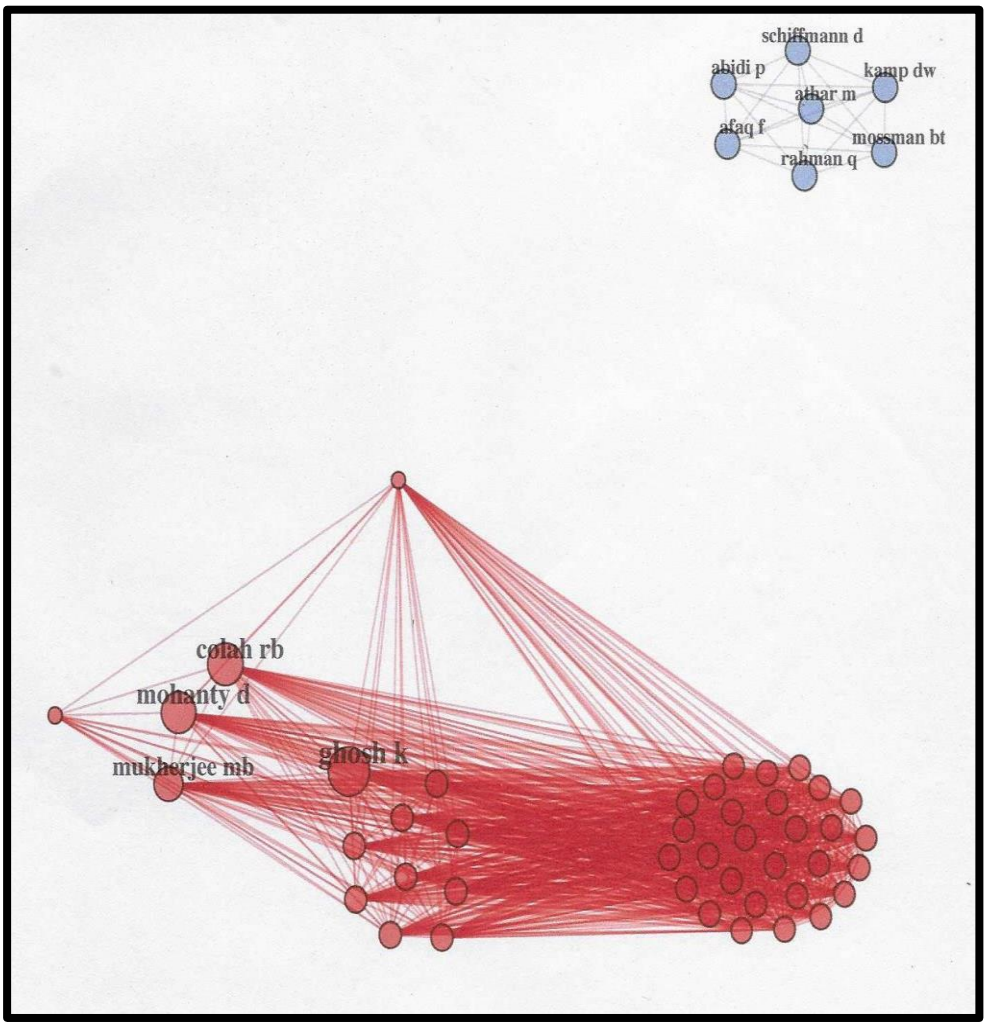
Table 4.41: Top ten articles published on Down syndrome with molecular biology from India during 1991-2014 receiving maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Nazki FH, 2014, Gene	144	20.57
2	Collins A, 2014, Mutat Res Rev Mutat Res	122	17.43
3	Narayanan S, 2005, FEBS Lett	122	7.00
4	Dhayalan A, 2011, Hum Mol Genet	98	9.80
5	Rao KS, 2007, Nat Clin Pract Neurol	81	5.79
6	Surjit M, 2006, J Biol Chem	75	5.00
7	Sodhi RK, 2010, Vasc Pharmacol	74	6.73
8	Ren J, 2011, Cell Signal	72	7.20
9	Sengupta S, 2005, Oncogene	59	3.69
10	Karthigeyan D, 2011, Med Res Rev	51	5.10

Source : SCOPUS database

Mutat Res Rev Mutat Res - Mutation Research : Reviews in Mutation Research
FEBS Lett - Federation of European Biochemical Societies Letters Journal
Hum Mol Genet - Human Molecular Genetics
Nat Clin Pract Neurol - Nature Clinical Practice: Neurology
J Biol Chem - Journal of Biological Chemistry
Vasc Pharmacol - Vascular Pharmacology
Med Res Rev - Medical Research Review

Figure 4.35: Coupling or Collaboration of authors of the articles published on G6PD deficiency without molecular biology from India during 1991 -2014.



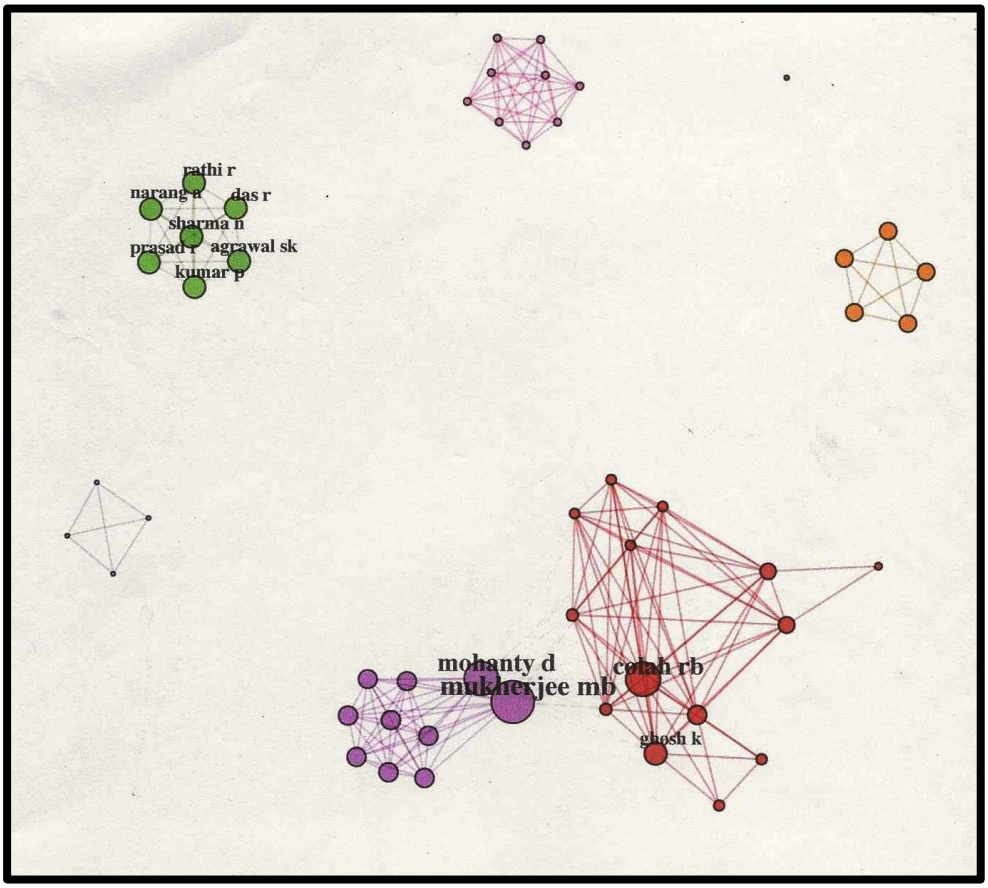
Source : SCOPUS database

Observation- Two clusters are seen.

Cluster 1 – Very compact, 44 nodes, Main nodes – K. Ghosh, R. B. Colah, D. Mohanty, M.B. Mukherjee

Cluster 2 – 7 nodes with equal prominence

Figure 4.36: Coupling or Collaboration of authors of the articles published on G6PD deficiency with molecular biology from India during 1991 -2014.



Source : SCOPUS database

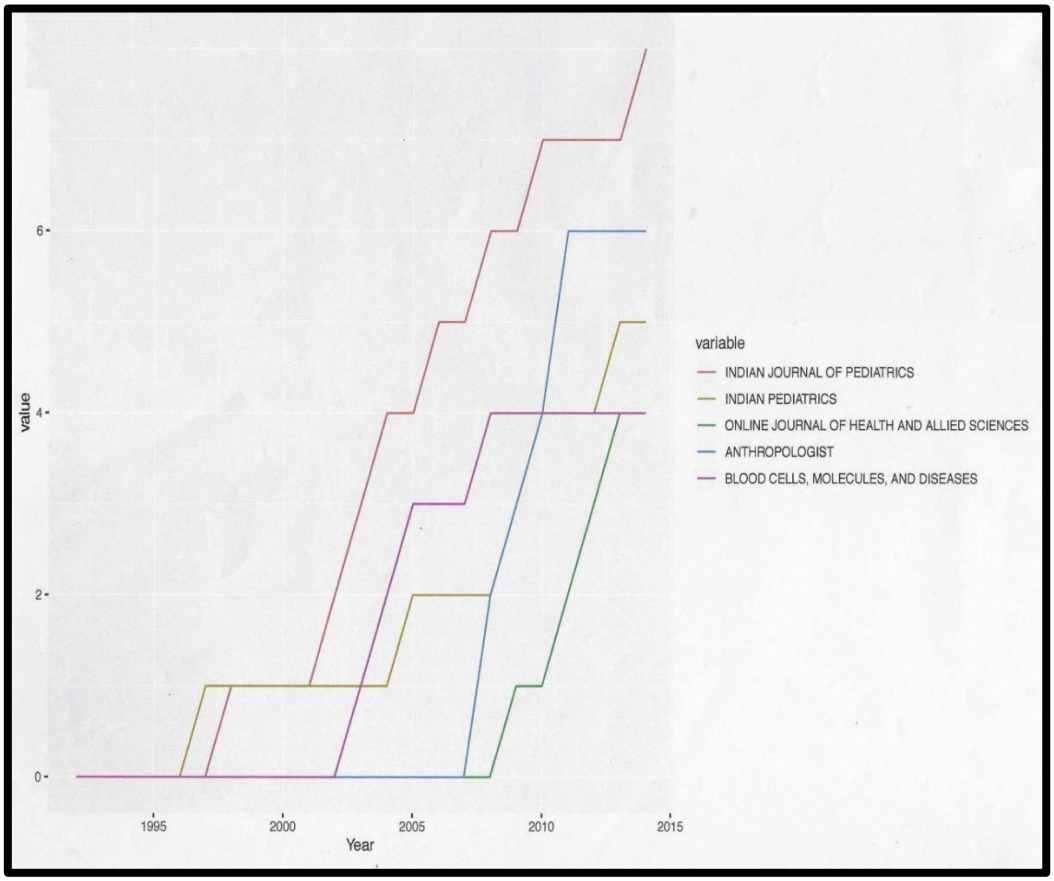
Observation- Totally five clusters are seen.

Cluster 1 – 14 nodes, Main nodes - R.B. Colah, K. Ghosh

Cluster 2 – 10 nodes, Main nodes - M. Mukherjee, D. Mohanty

Remaining clusters – 1-9 nodes

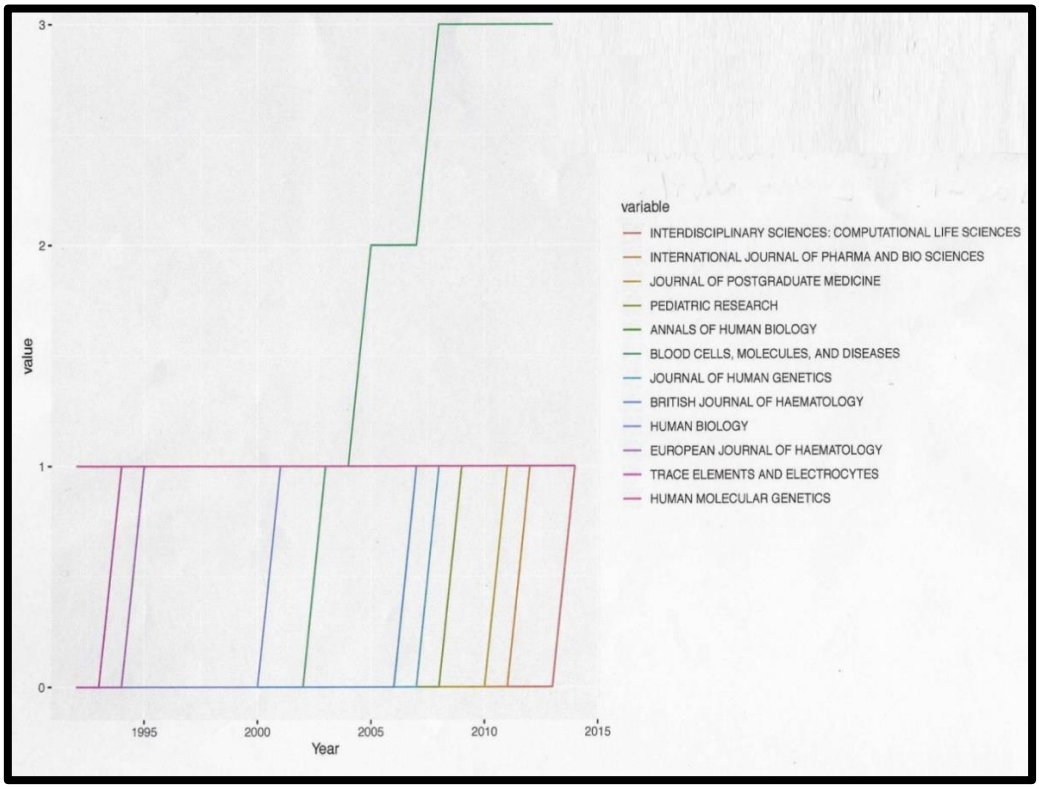
Figure 4.37: Top five journals showing trend of articles published on G6PD deficiency without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- This figure shows increasing trend of articles published on G6PD deficiency without molecular biology from India during 1991-2014.

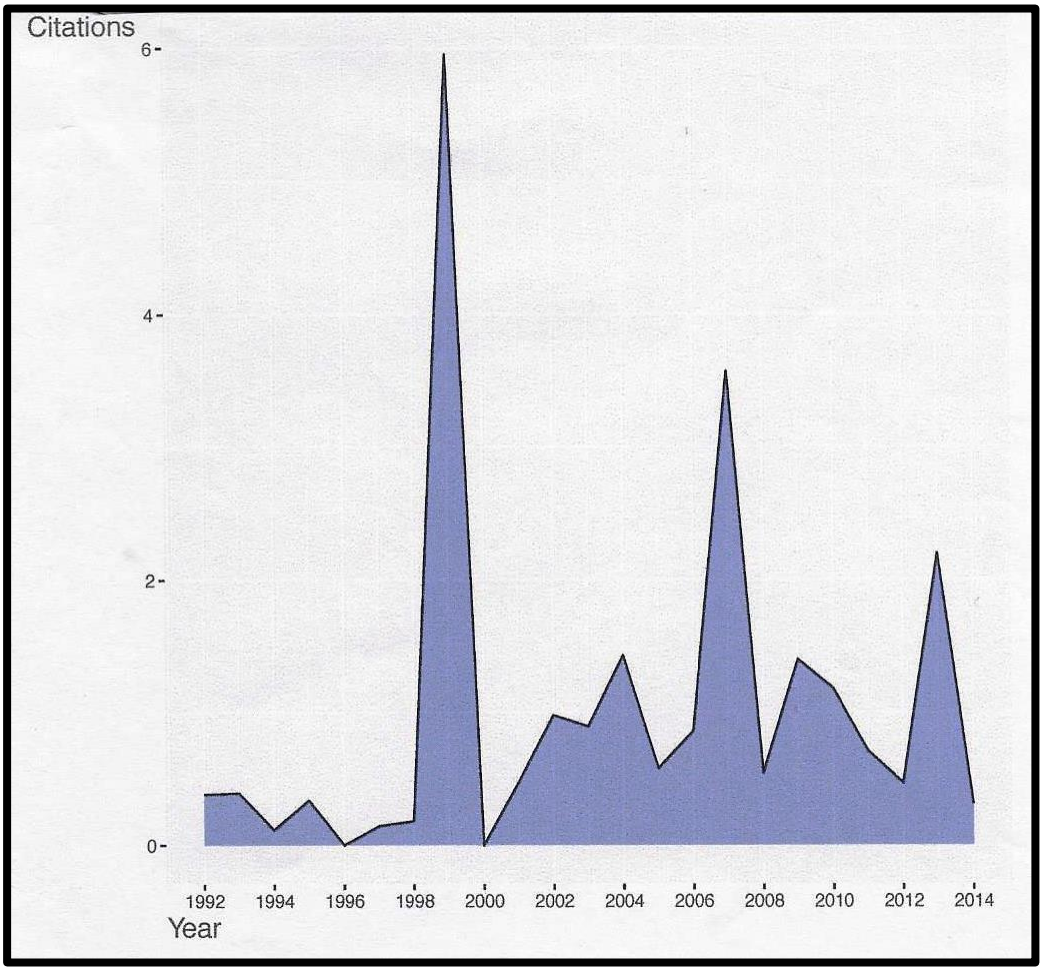
Figure 4.38: Trend of articles published in different journals on G6PD deficiency with molecular biology from India 1991 -2014.



Source : SCOPUS database

Observation- Maximum articles on G6PD deficiency with molecular biology from India during 1991-2014 have been published in the journal 'Annals of Human Biology'.

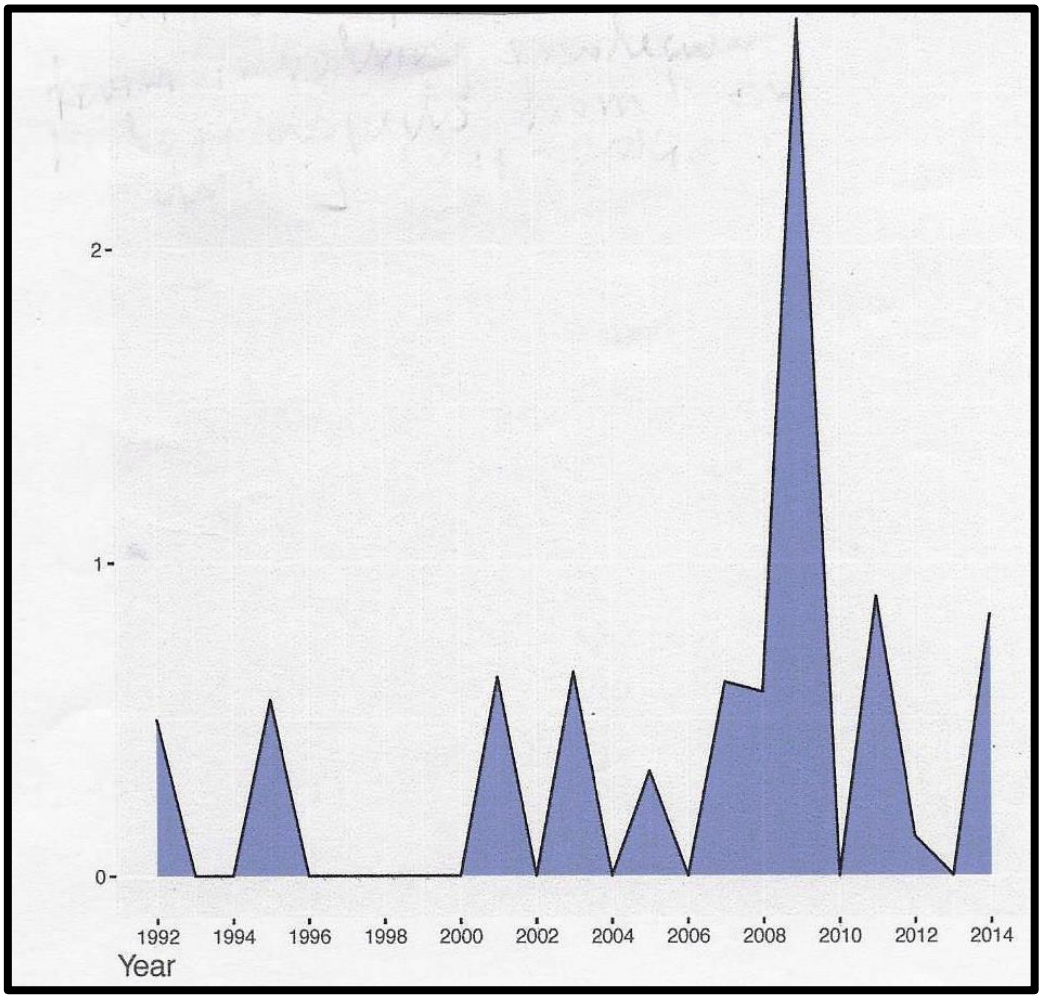
Figure 4.39: Average article citations per year received on articles published on G6PD Deficiency without molecular biology from India during 1991 -2014 .



Source: SCOPUS database

Observation- Maximum articles were cited during 1998 published on G6PD deficiency without molecular biology from India during 1991-2014.

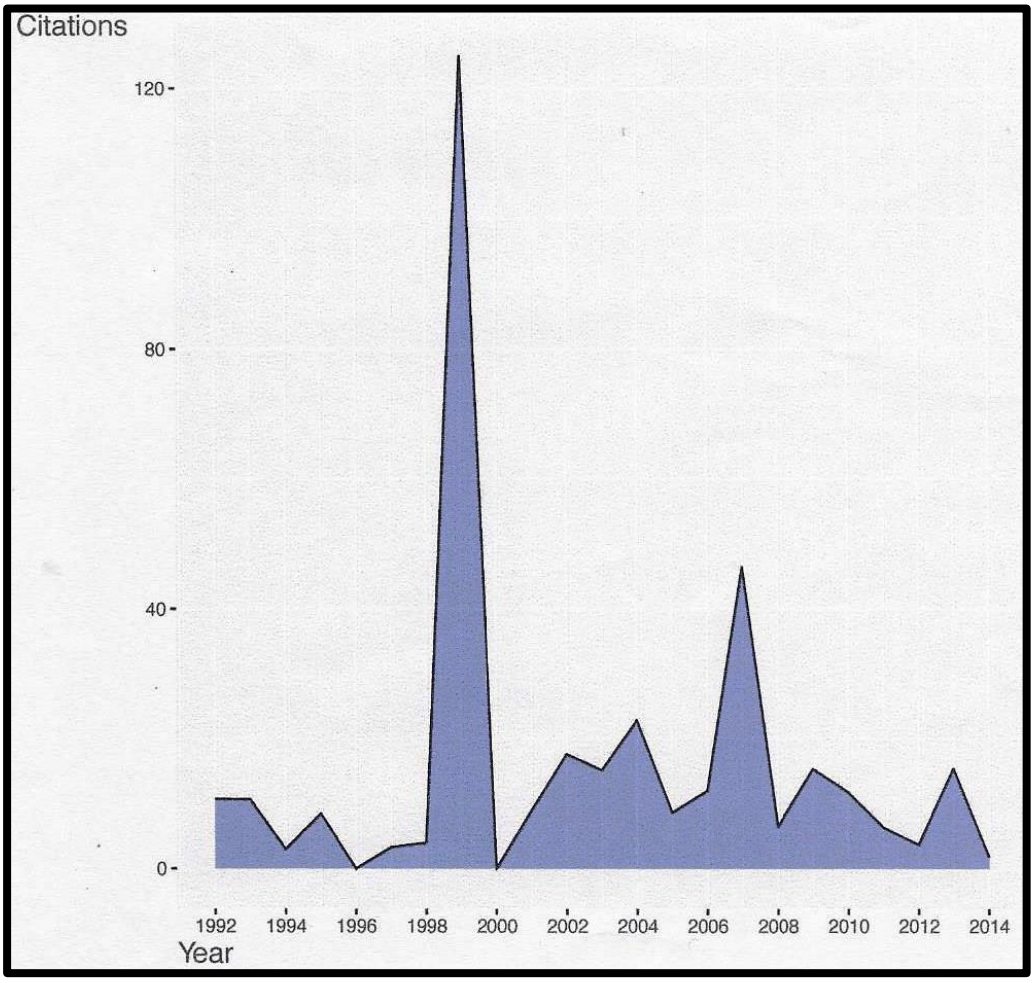
Figure 4.40: Average article citations per year received on articles published on G6PD deficiency with molecular biology from India during 1991 -2014.



Source: SCOPUS database

Observation- Maximum articles were cited during 2008 published on G6PD deficiency with molecular biology from India during 1991-2014.

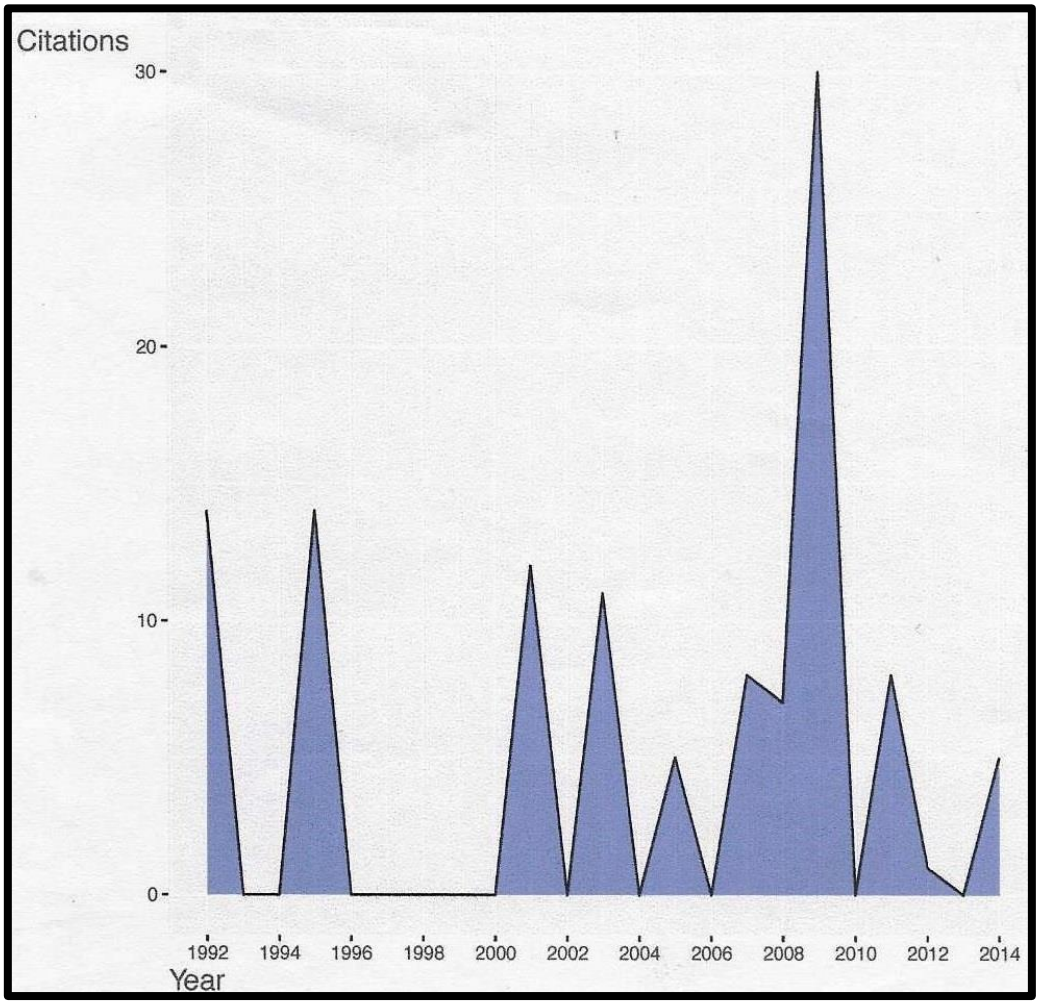
Figure 4.41: Average total citations per year received on articles published on G6PD deficiency without molecular biology from during 1991 -2014.



Source: SCOPUS database

Observation- Maximum total citations were received by articles 1998 published on G6PD deficiency without molecular biology aspects from India during 1991-2014.

Figure 4.42: Average total citations per year received on articles published on G6PD deficiency with molecular biology from India during 1991 -2014.



Source: SCOPUS database

Observation- Maximum total citations were received by articles during 1998 published on G6PD deficiency with molecular biology from India during 1991-2014.No articles received any citations during 1996 – 2000.

Table 4.42: Most productive top ten authors expressed on the basis of fractionalized published articles on G6PD deficiency without molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Balgir RS	9.00
2.	Mohanty D	3.37
3.	Colah RB	3.33
4.	Ghosh K	2.70
5.	Mukherjee MB	2.33
6.	Sukumar S	1.68
7.	Sachdeva MP	1.42
8.	Kedar PS	1.35
9.	Jamal F	1.08
10.	Qidwai T	1.08

Source : SCOPUS database

Explanation - Contribution of top ten most productive authors in publishing articles on G6PD deficiency without molecular biology from India during 1991-2014.

Table 4.43: Top ten articles published on G6PD deficiency without molecular biology from India during 1991-2014 received maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Beutler E, 2007, Am J Trop Med Hyg	138	9.86
2	Rahman Q, 1999, Crit Rev Toxicol	125	5.68
3	Domingo GJ, 2013, Malaria Journal	66	8.25
4	Sukumar S, 2004, Blood Cells Mol Dis	53	3.12
5	Kedar PS, 2003 Clin Lab Haematol	52	2.89
6	Verma IC, 2002, Community Genetics	52	2.74
7	Ghosh K, 2007, Parasitology Research	43	3.07
8	Galappaththy Gnl, 3013 Cochrane Database Syst Rev 2	40	5.0
9.	Tripathy V, 2007, J Postgrad Med	39	2.79
10.	Mohanty D, 2004, Ind J Paediatr	38	2.24

Source : SCOPUS database

Am J Trop Med Hyg - American Journal of Tropical Medicine and Hygiene
Crit Rev Toxicol - Critical Reviews in Toxicology
Blood Cells Mol Dis - Blood Cells Molecules and Diseases
Clin Lab Haematol - Clinical Laboratory Haematology
Cochrane Database Syst Rev 2 - Cochrane Database of Systematic Reviews 2
J Postgrad Med - Journal of Postgraduate Medicine
Ind J Pediatr - Indian Journal of Paediatrics

Table 4.44: Most productive top ten authors expressed on the basis of fractionalized published articles on G6PD deficiency with molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Colah RB	1.100
2.	Choudhary D	1.000
3.	Mukherjee MB	1.000
4.	Jamal F	0.833
5.	Qidwai T	0.833
6.	Mohanty D	0.800
7.	Ghosh K	0.600
8.	Sukumar S	0.500
9.	Chalvam R	0.400
10.	Kedar PS	0.400

Source : SCOPUS database

Explanation - Contribution of top ten most productive authors in publishing articles on G6PD deficiency with molecular biology from India during 1991-2014.

Table 4.44 represents top ten authors expressed on the basis of fractionalized published articles on G6PD deficiency involving molecular biology aspects from India during 1991-2014. R.B. Colah occupied the first rank with the weightage of 1.1 of the total articles published by her during this period.

Table 4.45 reveals top ten articles published on G6PD deficiency involving molecular biology aspects from India during 1991-2014 and receiving maximum citations. An article published by Agarwal et al (2007) in the journal 'Pediatric Research' was on the top of the list receiving 30 citations during this period.

4.2.4 von Willebrand disease

Figure 4.43 reveals the network analysis diagram showing coupling of authors of the articles published on von Willebrand disease from India during 1991-2014. Two clusters were seen which were interconnected with each other indicating some collaboration between two groups. The first compact cluster depicted 35 nodes with equal prominence while the second cluster revealed 15 nodes with A.Srivastava occupying the prominent node. K. Ghosh and R. Saxena occupied the nodes of the same size but smaller than that of A. Srivastava.

Figure 4.44 presents the network analysis diagram showing coupling of authors of the articles published on von Willebrand disease involving molecular biology aspects from India during 1991-2014. Only one cluster with 29 nodes was seen,. This shows that there has been a good collaboration among the scientists in conducting research in this aspect. R. Saxena, F. Cyen, V. Budde, R. Schnepenhrim occupying nodes with equal strength. F. Ahmad and M. Kannan occupied nodes with equal size but smaller than the other nodes.

Figure 4.45 shows trend of articles published on von Willebrand disease in different journals from India during 1991-2014. 'Hemophilia' was the main journal where maximum articles on this disorder were published.

Figure 4.46 depicts the trend of articles published on von Willebrand disease involving molecular biology aspects from India during 1991-2014. Number of articles published during this time period was small and were published mainly in four journals viz. 'Hemophilia', 'PLOS ONE', 'Thrombosis and Hemostasis' and 'Blood Cells Molecules and Diseases'.

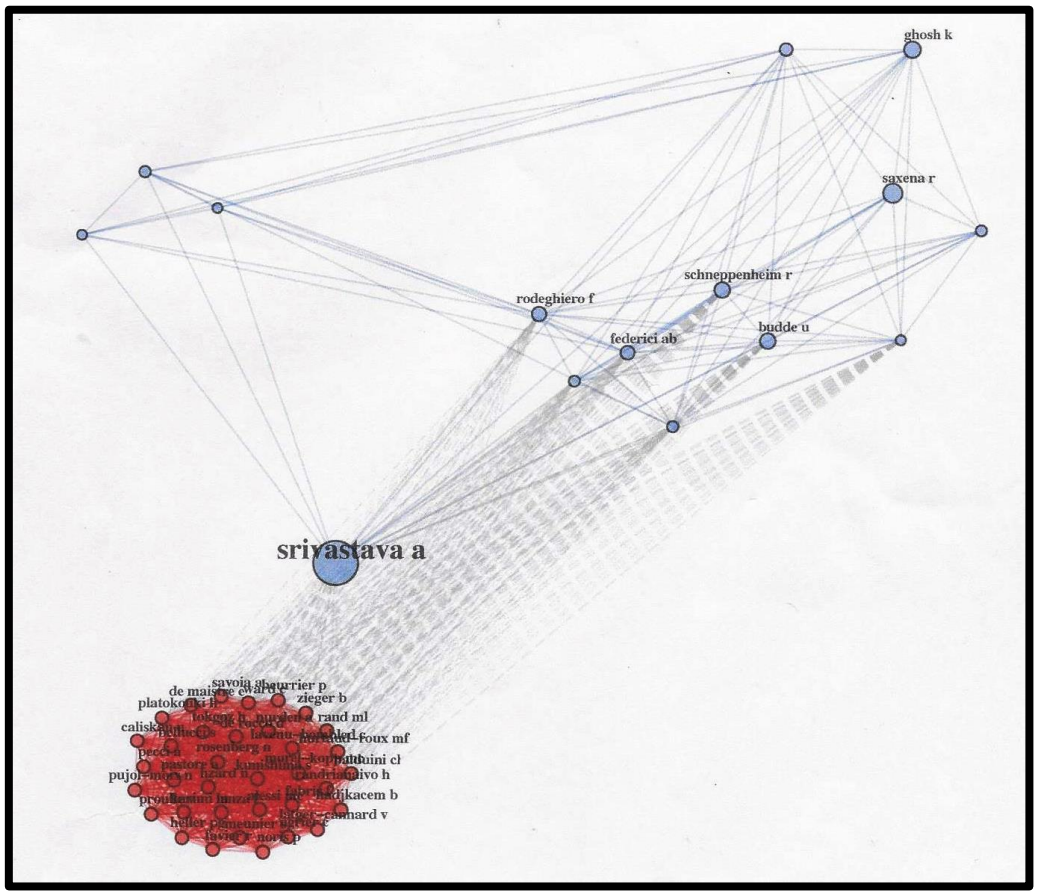
Table 4.45: Top ten articles published on G6PD deficiency with molecular biology aspects from India during 1991-2014 received maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Agarwal SK 2009 Pediatric Research	30	2.500
2	Reddy PH, 1995, Eur J Haematol	14	0.538
3	Ahluwalia A, 1992, Hum Mol Genet	14	0.483
4	Murhekar KM, 2001,Human Biology	12	0.600
5	Sukumar S, 2003, Blood Cells Mol Dis	11	0.611
6	Chalvam R, 2008, J Hum Genet	09	0.692
7	Bhatia S, 2011, J Post grad Med	08	0.800
8	Chalvam R, 2007, BrJ Haematol	08	0.571
9	Nishank SS, 2008, Ann Hum Biol	06	0.462
10	Kedar PS, 2008, Blood Cells Mol Dis	06	0.462

Source : SCOPUS database

Eur J Haematol - European Journal of Haematology
Hum Mol Genet - Human Molecular Genetics
Blood Cells Mol Dis - Blood Cells Molecules and Diseases
J Hum Genet - Journal of Human Genetics
J Postgrad Med - Journal of Postgraduate Medicine
Br J Haematol - British Journal of Haematology
Ann Hum Biol - Annals of Human Biology

Figure 4.43: Coupling or collaboration of authors of the articles published on von Willebrand disease without molecular biology from India during 1991- 2014.



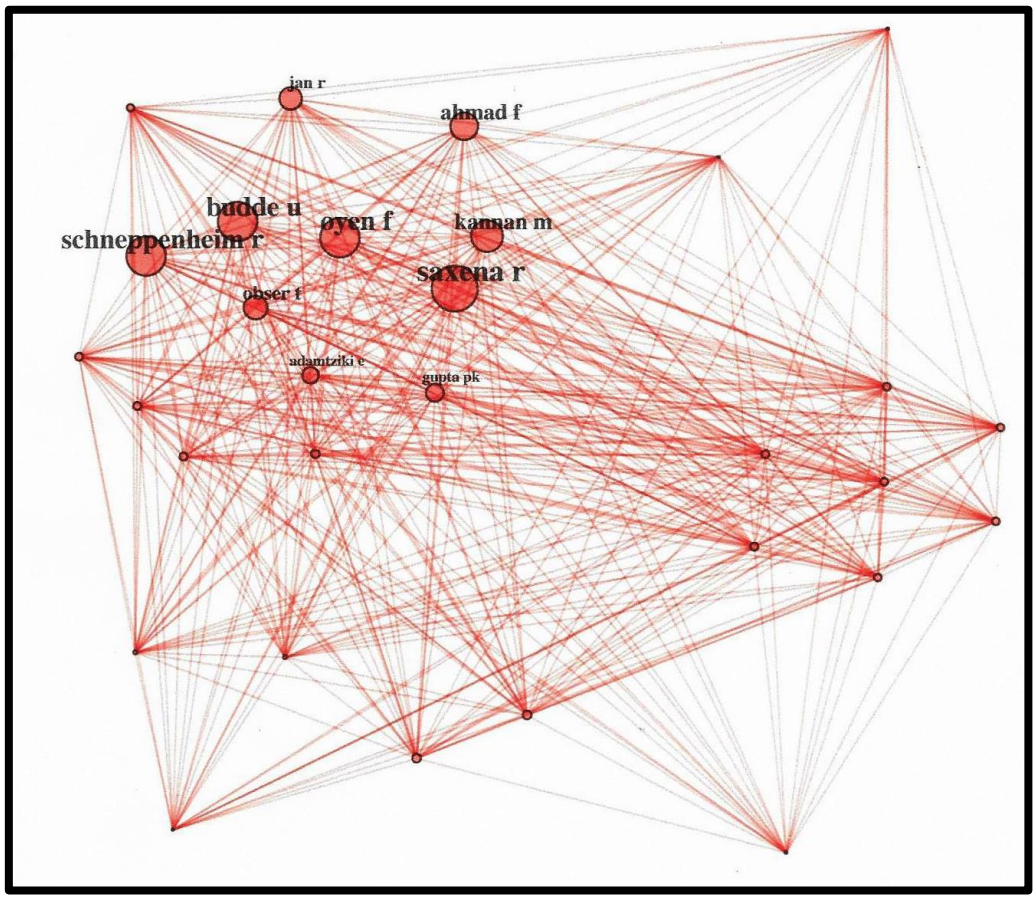
Source : SCOPUS database

Observation- Two clusters are seen

Cluster 1 – Very compact, 35 nodes with equal prominence

Cluster 2 – 15 nodes Main nodes – A. Srivastva, R. Saxena, K. Ghosh

Figure 4.44: Coupling or collaboration of authors of the articles published on von Willebrand disease with molecular biology from India during 1991-2014.

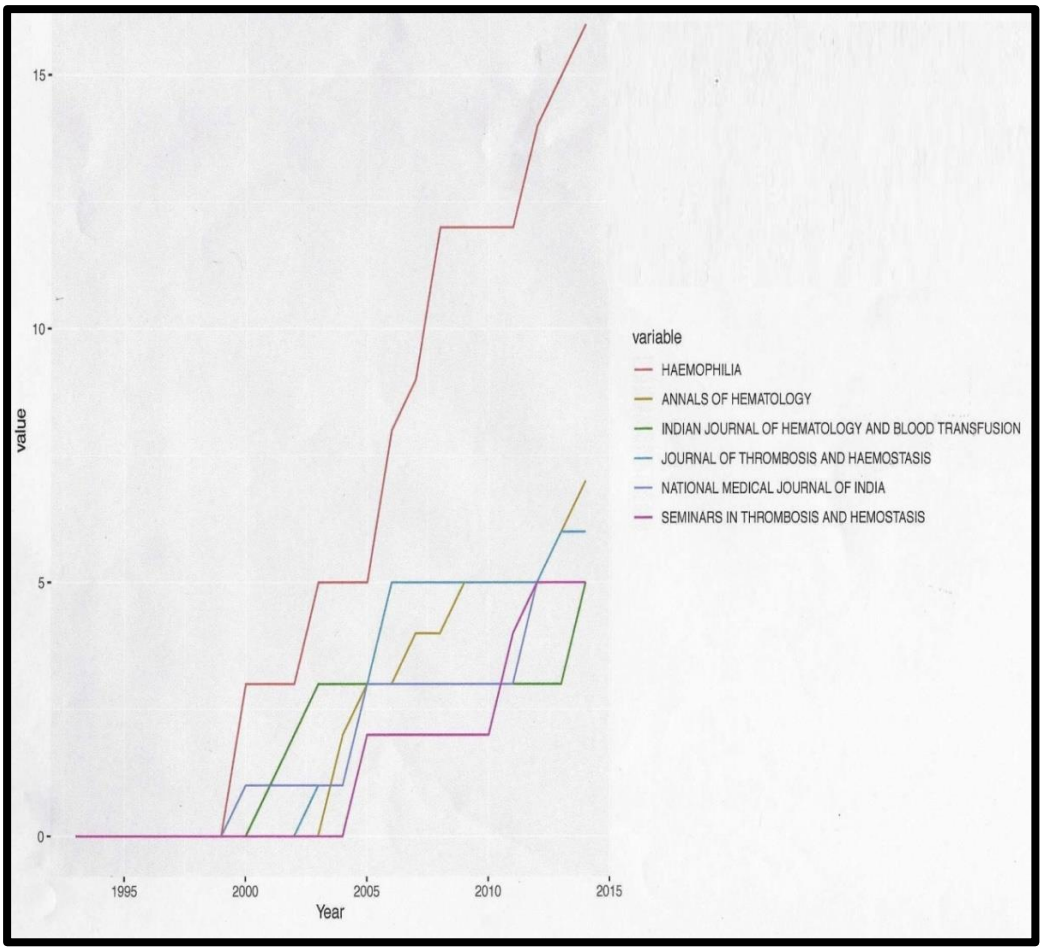


Source : SCOPUS database

Observation- Only one cluster is seen. It indicates good collaboration

Totally 29 nodes, Main nodes – R. Saxena, F. Cyen, V. Budde, R. Schneppenheim

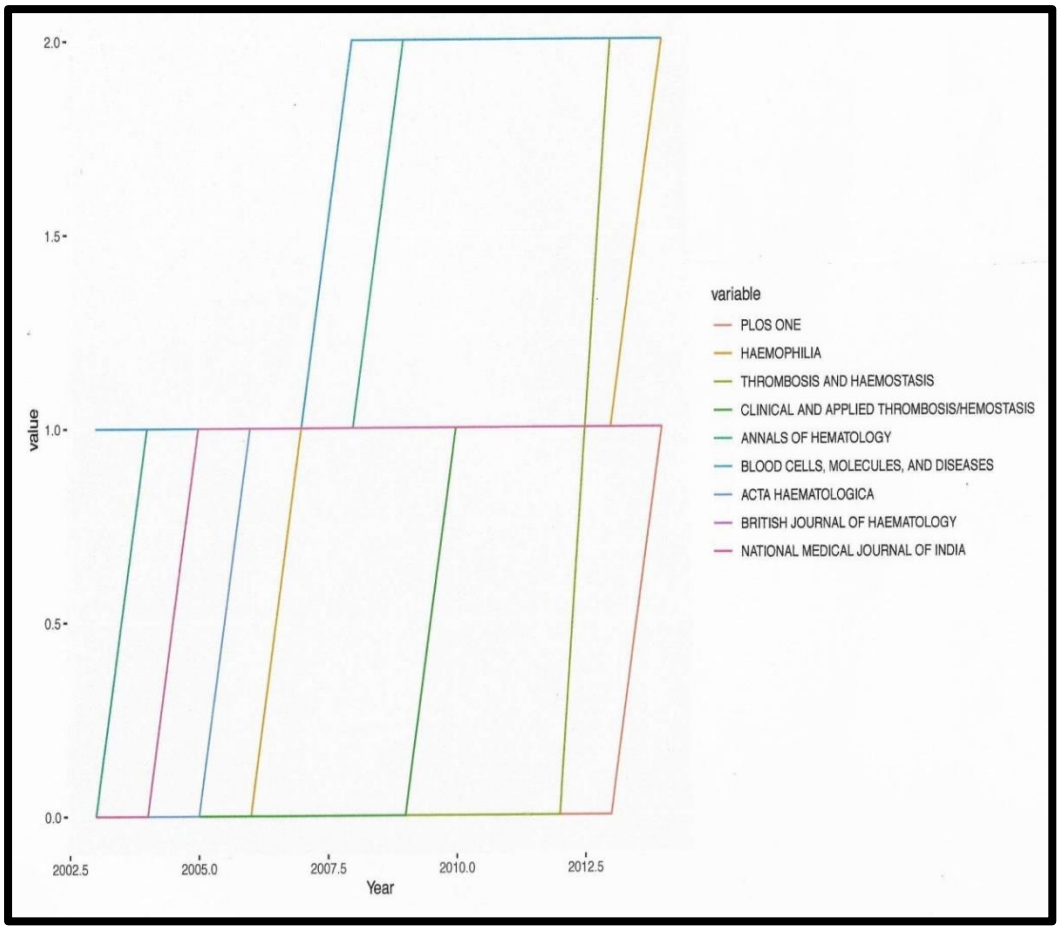
Figure 4.45: Top five journals showing trend of articles published on von Willebrand disease without molecular biology from India during 1991 -2014 .



Source : SCOPUS database

Observation - Regular articles on von Willebrand disease started appearing since 1999 onwards maximum articles on von Willebrand disease without molecular biology from India during 1991-2014 have been published in the journal 'Hemophilia'.

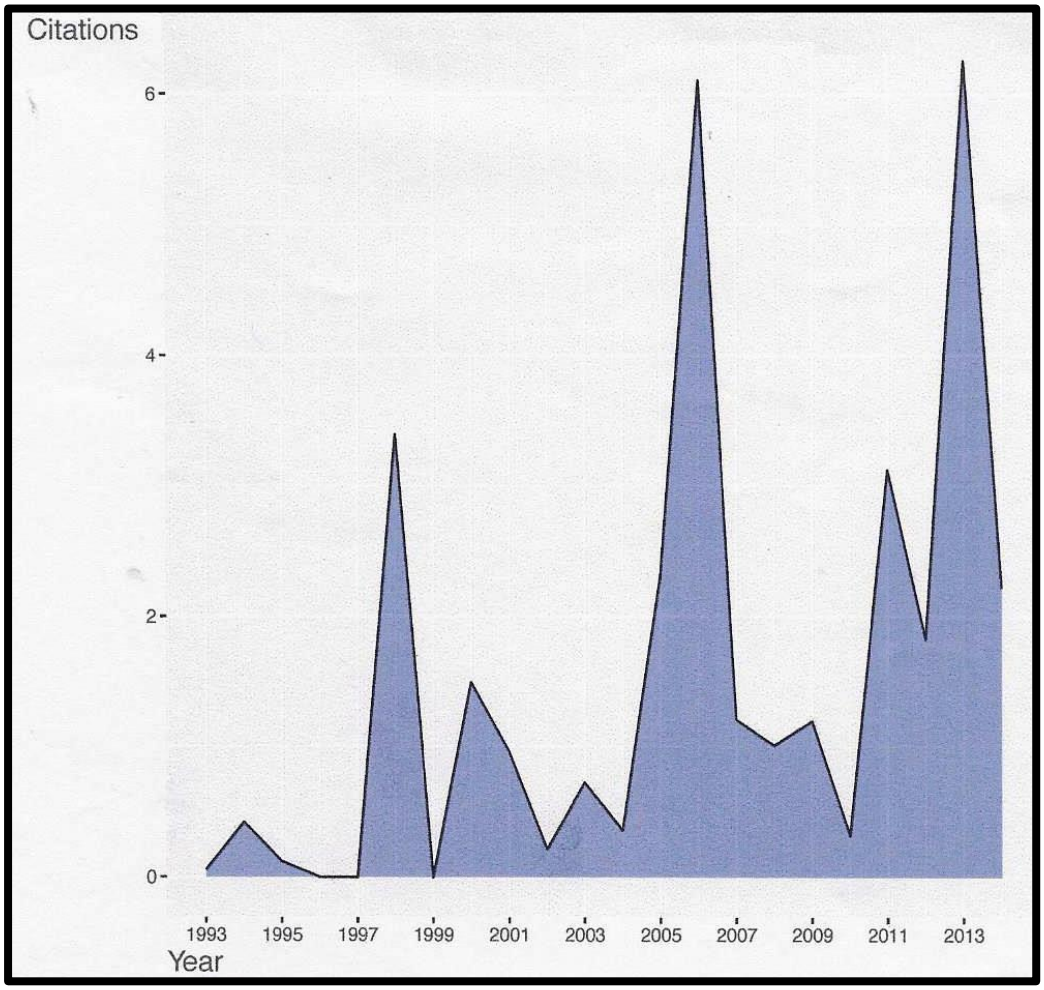
Figure 4.46: Top nine journals showing trend of articles published von Willebrand disease with molecular biology from during 1991 – 2014.



Source: SCOPUS database

Observation- Regular articles on von Willebrand disease with molecular biology started appearing regularly in the journals since 2003 onwards. Maximum articles on this disease have been published in journal 'Clinical and Applied Thrombosis and Hemostasis'.

Figure 4.47: Average article citations per year received on articles published on von Willebrand disease without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- Maximum articles were cited during 2013 followed by those cited during 2005 published on von Willebrand disease without molecular biology from India during 1991-2014.

Figure 4.47 reveals average article citations per year on articles published on von Willebrand disease from India during 1991-2014. Even though less number of articles were published during 2004-2007 an 2011-2013, more than 6.0 average article citations were received.

Figure 4.48 demonstrates average article citations per year on articles published von Willebrand disease involving molecular biology aspects from India during 1991-2014. A sharp spike was seen during the period 2003-2004.

Figure 4.49 depicts average total citations per year on articles published on von Willebrand disease from India during 1991-2014. Sharp spikes indicating increase in average total citations were seen during the periods 1997-1999 and 2004-2007.

Figure 4.50 illustrates average total citations per year on articles published on von Willebrand disease involving molecular biology aspects from India during 1991-2014. A sharp spike was seen receiving more than 80 total citations during 2003-2004.

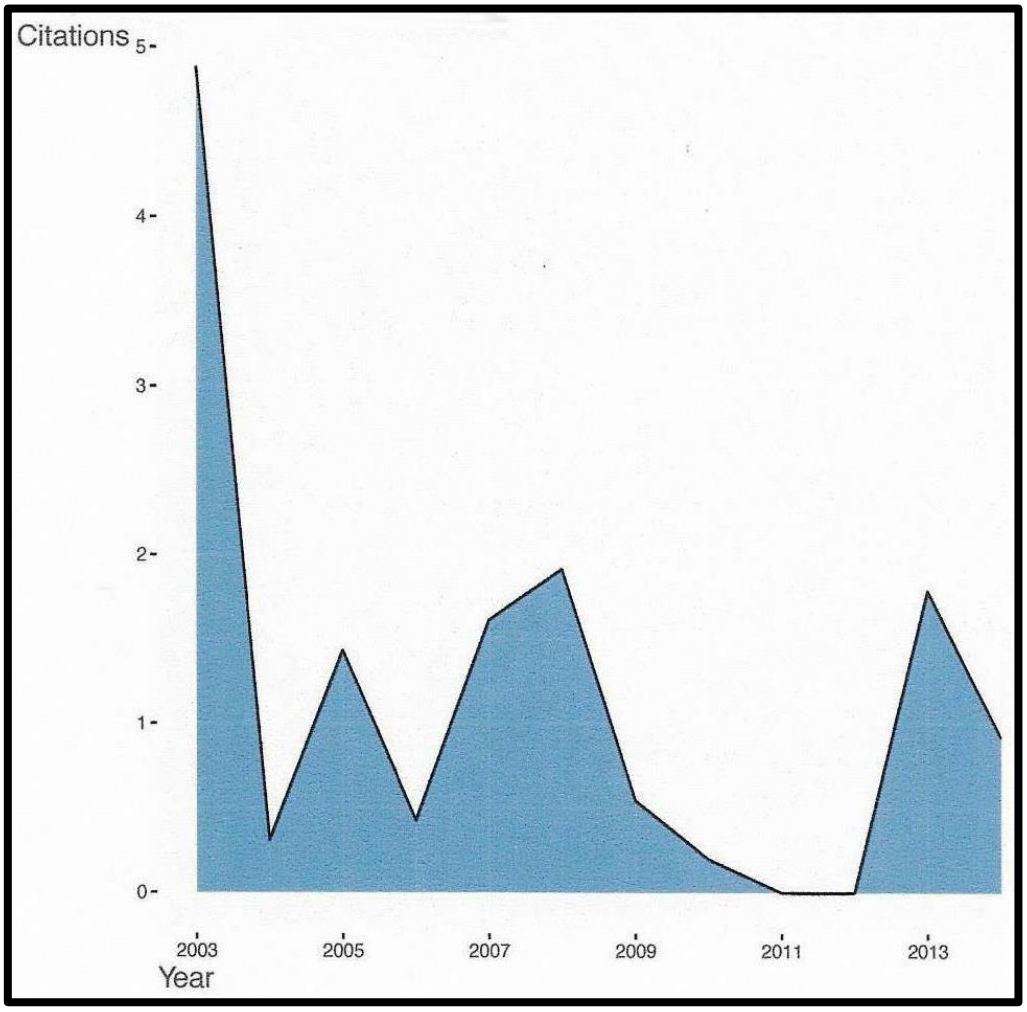
Table 4.46 shows most productive top ten authors expressed on the basis of fractionalized published articles on von Willebrand disease from India during 1991-2014. K. Ghosh occupied the top position with the weightage of 9.75 of the total articles published by him during this period.

Table 4.47 reveals the most cited articles on von Willebrand disease from India during 1991-2014. An article by Srivastava et al (2013) published in the journal 'Hemophilia' received 810 total citations during this period and ranked first in the table.

Top ten authors expressed on the basis of fractionalized published articles on von Willebrand disease involving molecular biology aspects from India are depicted in table 4.48. R. Saxena was on the top position with the weightage of 1.48 of the total articles published by her during the above mentioned period.

Table 4.49 reveals top ten articles on von Willebrand disease involving molecular biology aspects from India during 1991-2014 and receiving maximum citations. An articles published by Baronciani et al (2003) in the journal 'Blood Cells Molecules and Diseases' received 83 total citations during this period and ranked on the top position.

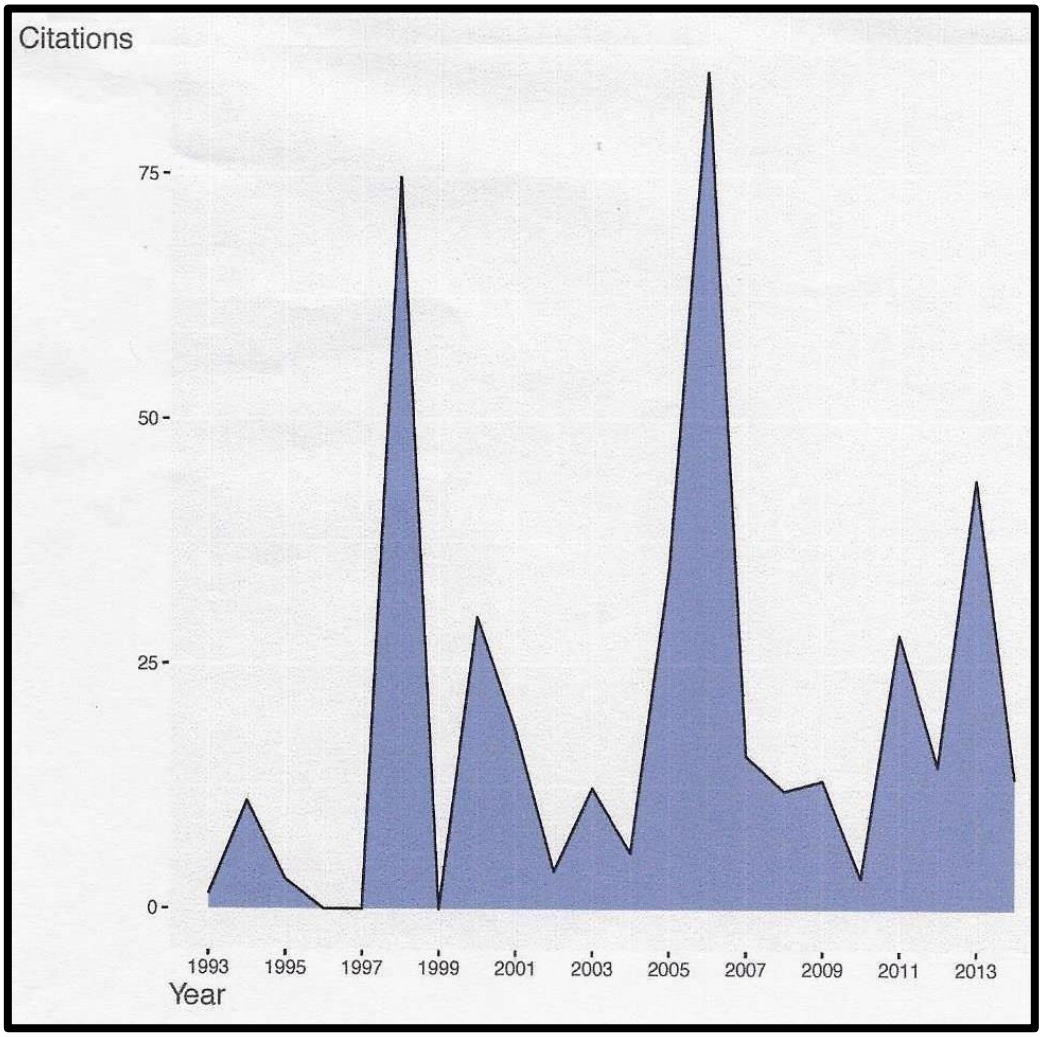
Figure 4.48: Average article citations per year received on articles published on von Willebrand disease with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum articles were cited during 2003 published on von Willebrand disease with molecular biology from India during 1991-2014.

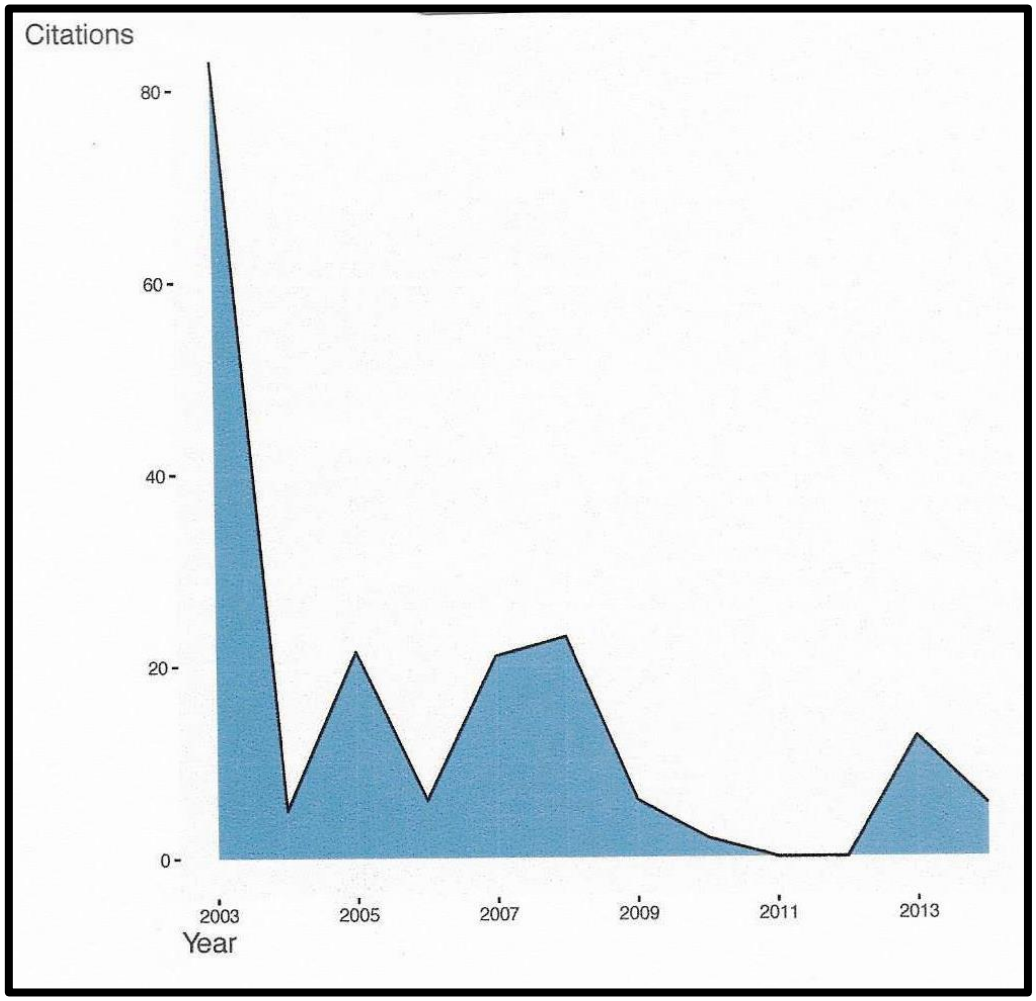
Figure 4.49: Average total citations per year received on articles published on von Willebrand disease without molecular biology from India during 1991 -2014



Source : SCOPUS database

Observation - Maximum articles were cited during 2005 followed by those cited during 1997 published on von Willebrand disease without molecular biology from India during 1991-2014.

Figure 4.50: Average total citations per year received on articles published on von Willebrand disease with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- Maximum total citation were received during 2003 for articles published on von Willebrand disease with molecular biology from India during 1991-2014.No articles received any citations during 2011-2012.

Table 4.46: Most productive top ten authors expressed on the basis of fractionalized published articles on von Willebrand disease without molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Ghosh K	9.75
2.	Shetty S	7.01
3.	Saxena R	5.39
4.	Srivastava A	3.35
5.	Mmohanty D	3.31
6.	Choudhary VP	2.85
7.	Kasatkar P	2.73
8.	Kannan M	2.36
9.	Das UN	2.00
10.	Gupta PK	1.96

Source : SCOPUS database

Explanation- Contribution of top ten most productive authors in publishing articles on von Willebrand disease without molecular biology from India during 1991-2014.

Table 4.47: Top ten articles published on von Willebrand disease without molecular biology from India during 1991-2014 received maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Srivastava A, 2013, Hemophilia	810	101.25
2	Sadler JE, 2006, J Thromb Hemost	721	48.07
3	Rodeghiero F, 2005, J Thromb Hemost	248	15.50
4	Kadir RA, 1998, BJOG, Int J Obster Gynecol	199	8.65
5	Jain S, 2011, J Pharm Bioallied sci	168	16.80
6	Ghoshal K, 2014, Science World Journal	85	12.14
7	Baronciani L, 2003, Blood Cells Mol Dis	83	4.61
8	Peyandi F, 2006, Hemophilia	81	5.40
9	Kodavanti UP, 2011, Environ Health Perspect	70	7.00
10	Savoia A 2014, Human Mutation	68	9.71

Source : SCOPUS database

J Thromb Hemost - Journal of Thrombosis and Hemostasis
BJOG, Int J Obster Gynecol - BJOG International Journal of Obstetrics and Gynaecology
J Pharm Bioallied sci - Journal of Pharmacy and Bioallied Science
Blood Cells Mol Dis - Blood Cells Molecules and Diseases
Environ Health Perspect - Environmental Health Perspectives

Table 4.48: Most productive top ten authors expressed on the basis of fractionalized published articles on von Willebrand disease with molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Saxena R	1.48
2.	Ghosh K	1.28
3.	Shetty S	1.28
4.	Kannan M	1.17
5.	Ahmad F	0.92
6.	Budde U	0.64
7.	Oyen F	0.46
8.	Schneppenheim R	0.64
9.	Gupta PK	0.56
10.	Mohanty D	0.54

Source : SCOPUS database

Explanation - Contribution of top ten most productive authors in published articles on von Willebrand disease with molecular biology from India during 1991-2014.

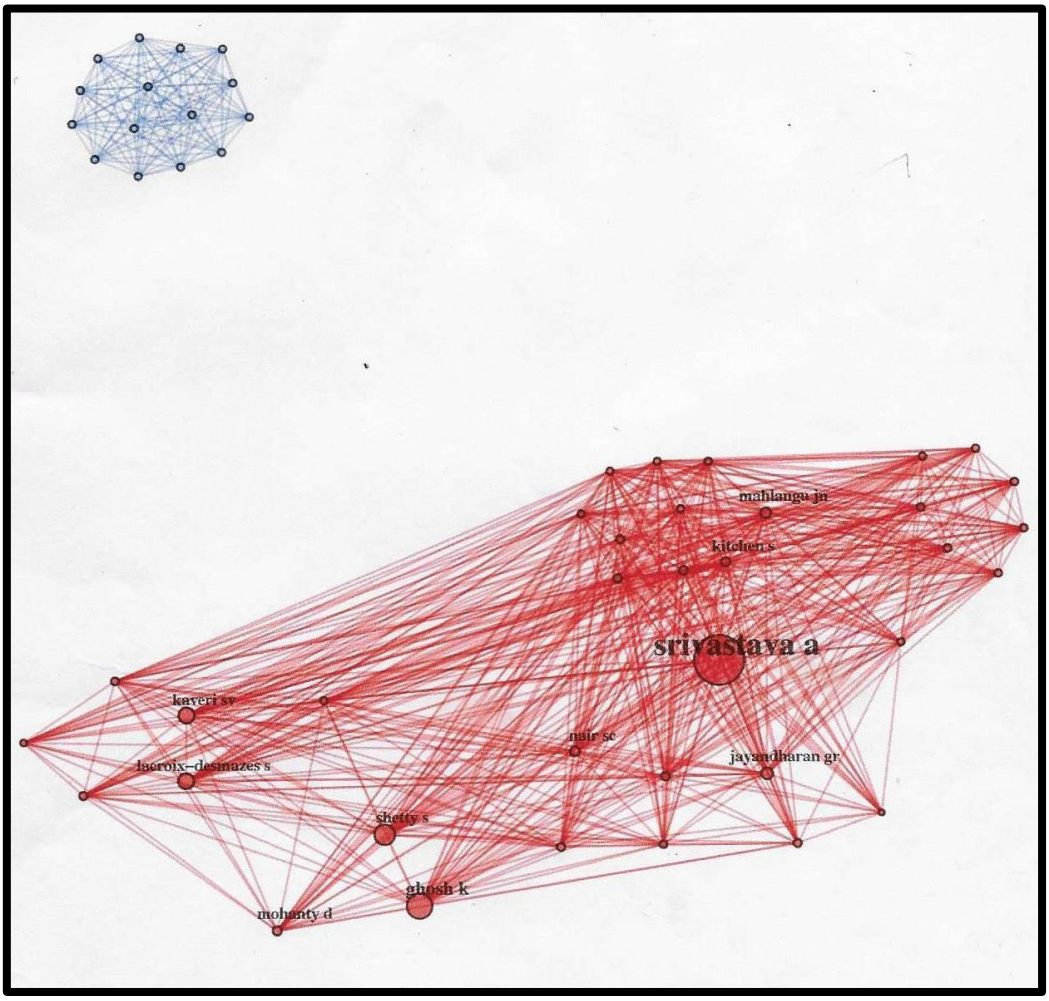
Table 4.49: Top ten articles published on von Willebrand disease with molecular biology from India during 1991-2014 received maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Baronciani L,2003, Blood Cells Mol Dis	83	4.61
2	Gupta PK, 2005, Br J Haematol	38	2.37
3	Gupta PK, 2008, Blood Cells Mol Dis	23	1.76
4	Shetty S, 2007 Haemophilia	21	1.50
5	Ahmad F, 2013, Thromb Haemost	16	2.00
6	Ahmad F, 2013, Thromb Haemost	09	1.12
7	Kasatkar P, 2014, PLOS one	08	1.1
8	Ahmad F, 2009, Annals of Hematology	06	0.50
9	Trasi S, 2006, Acta Haematologica	06	0.40
10	Trasi S, 2005, Natl Med J Ind	05	0.31

Source : SCOPUS database

Blood Cells Mol Dis - Blood Cells Molecules and Diseases
Br J Haematol - British Journal of Haematology
Thromb Haemost - Thrombosis and Haemostasis
Natl Med J Ind - National Medical Journal of India

Figure 4.51: Coupling or collaboration of authors of the articles published on Hemophilia A without molecular biology from India during 1991-2014.



Source : SCOPUS database

Two clusters are seen

Cluster 1 - 35 nodes Main nodes - A. Srivastava, K. Ghosh, S. Shetty

Cluster 2 - 15 nodes with equal prominence

4.2.5 Hemophilia A

Figure 4.51 shows the network analysis diagram of coupling of authors of the articles published on hemophilia A from India during 1991-2014. Two clusters were seen. The main cluster showed 35 nodes. Amongst these the most prominent node was occupied by Hemophilia A.

A. Srivastava. K. Ghosh and S. Shetty occupied the nodes of the same size but smaller than that of A. Srivastava. D. Mohanty, and G.R. Jayandharan occupied very small nodes. The second cluster was a small one showing 15 nodes with equal size

Figure 4.52 reveals the network analysis diagram of coupling of authors of articles published on hemophilia A involving molecular biology aspects from India during 1991-2014. Totally 5 clusters were seen. However no prominent cluster was not seen indicating lack of collaboration in the research carried out on this disorder. The first cluster showed 17 nodes with K. Ghosh and S. Shetty occupying prominent nodes. The second cluster showed 13 nodes with G.R. Jayandharan and A. Srivastava occupying prominent nodes. S.C. Nair occupied a node with smaller size than the nodes occupied by two scientists. The third cluster revealed 8 nodes. M. Chandy occupied the prominent node. The fourth cluster showed 7 nodes with equal prominence while the fifth cluster showed 5 nodes.

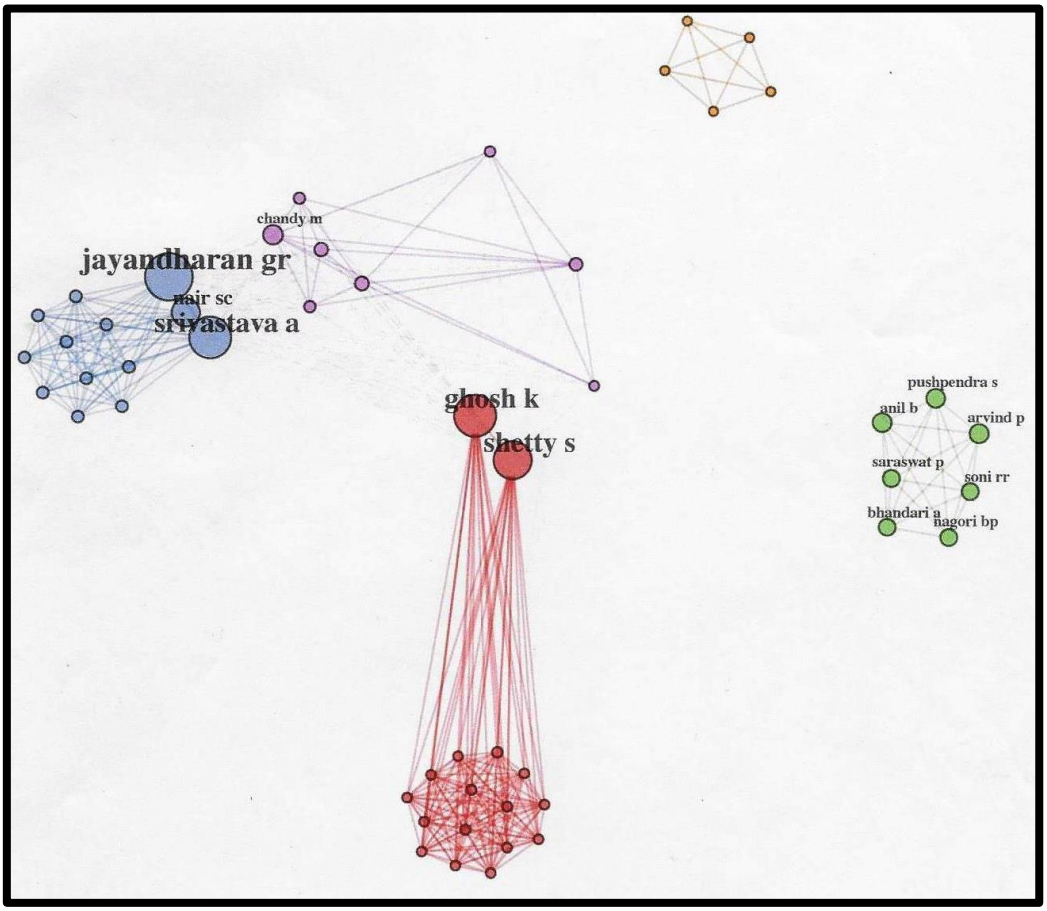
Figure 4.53 depicts trend of articles published on hemophilia A from India during 1991-2014. The majority of the articles on this disorder have been published in the journal 'Hemophilia'.

Figure 4.54 reveals trend of articles published on hemophilia A involving molecular biology aspects from India during 1991-2014. Here also main journal which published maximum articles on this disorder was 'Hemophilia'; but articles also have been published in other journals even though the number of articles published was relatively small as compared to other set.

Figure 4.55 describes average article citations per year on articles published on hemophilia A involving molecular biology aspects from India during 1991-2014. A sharp spike was seen during 2011-2013.

Figure 4.56 shows average article citations per year on articles published on hemophilia A involving molecular biology aspects from India during 1991-2014. Two sharp spikes during the period 2009-2011 and 2012-2014 were seen.

Figure 4.52: Coupling or collaboration of authors of the articles published on Hemophilia A with molecular biology from India during 1991-2014.

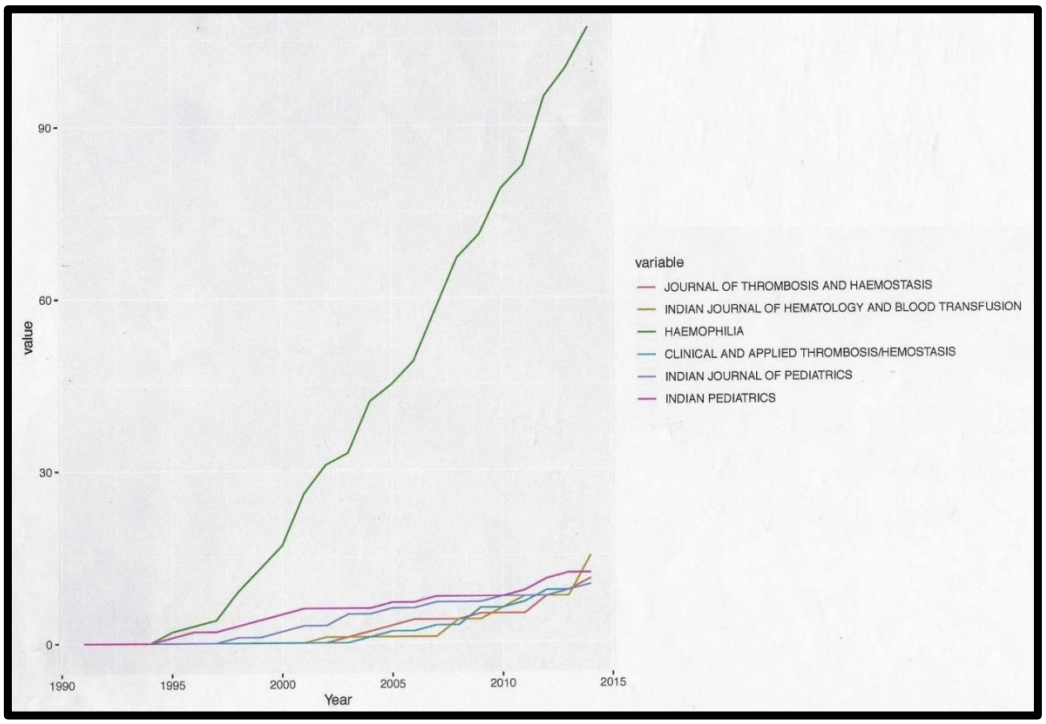


Source : SCOPUS database

Observation - Totally five clusters are seen

Cluster 1- 16 nodes **Main nodes – K. Ghosh, S. Shetty**
Cluster 2 - 13 nodes **Main nodes – A. Srivasatava, G. R. Jayandharan**
Cluster 3 - 8 nodes **Main node – M. Chandy**
Cluster 4 and 5 - Nodes with equal prominence

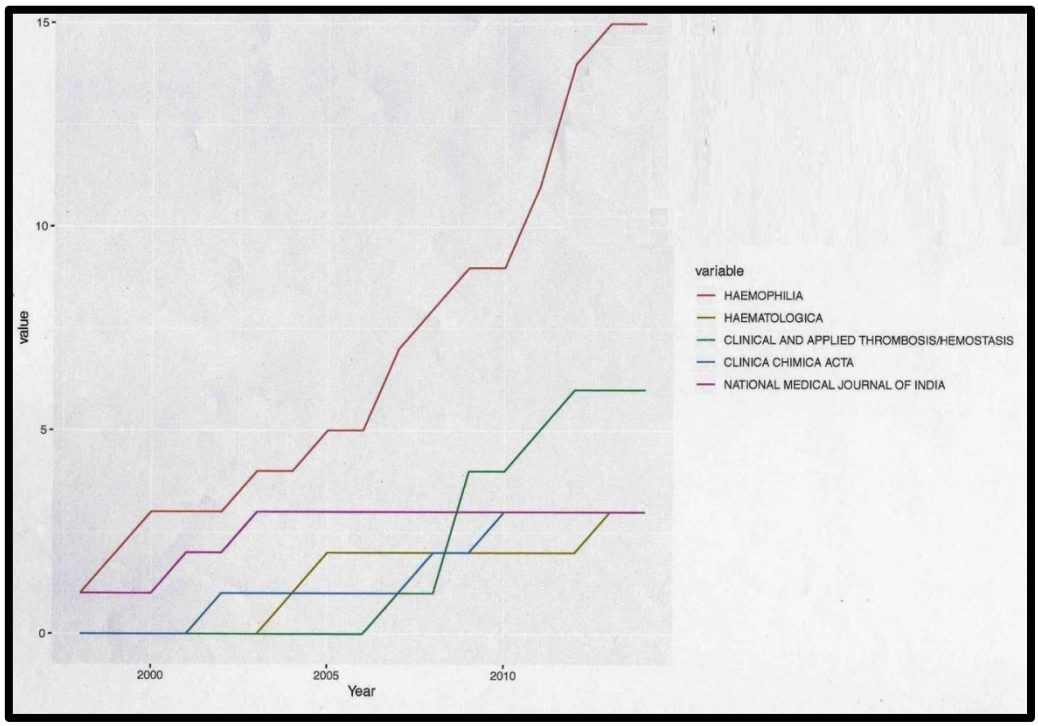
Figure 4.53: Top six journals showing trend of articles published Hemophilia A without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum articles on Hemophilia A without molecular biology from India during 1991-2014 were published in journal 'Hemophilia'.

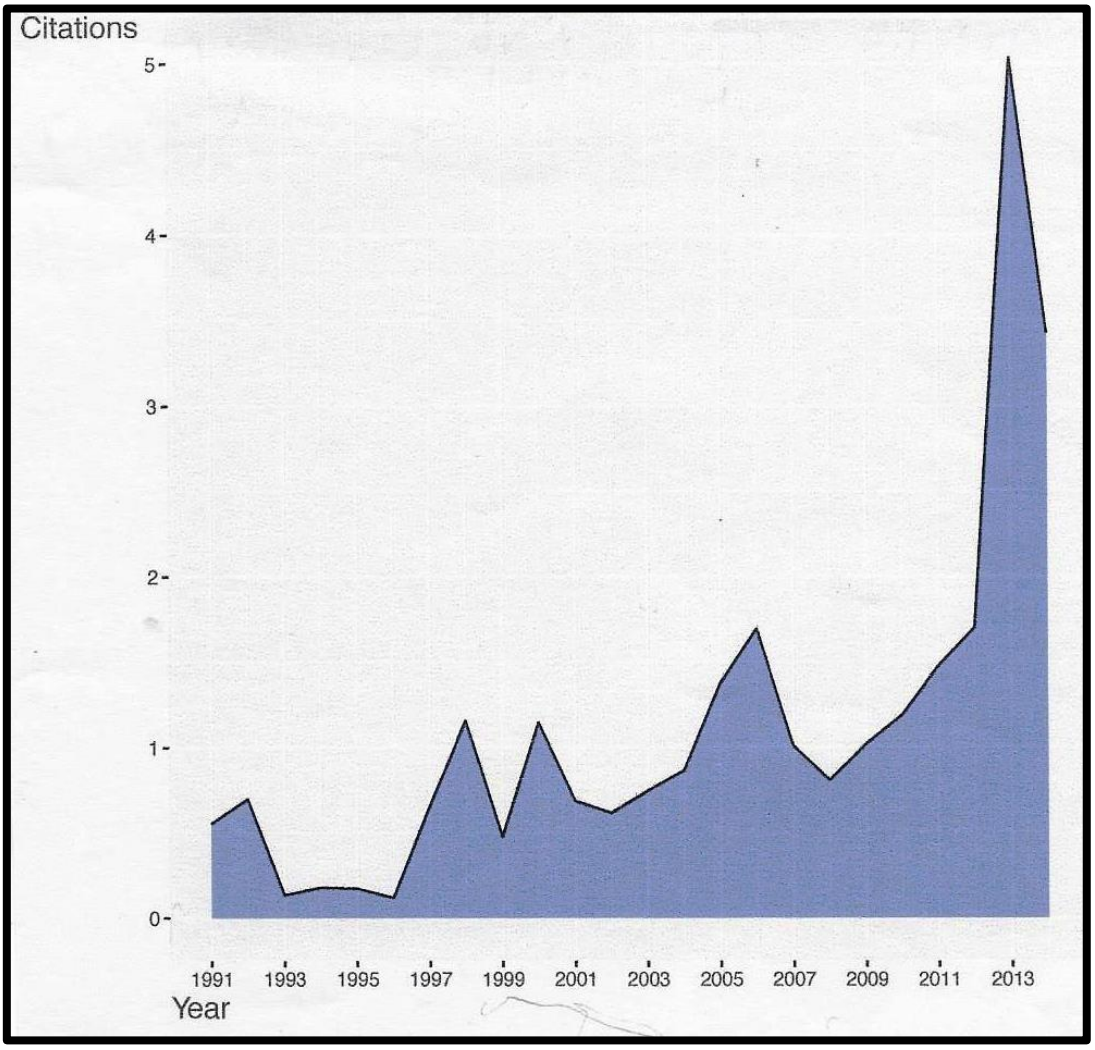
Figure 4.54: Trend of articles published in different journals on Hemophilia A with molecular biology from India during 1991 -2014 .



Source : SCOPUS database

Observation - Maximum articles on Hemophilia A with molecular biology from India during 1991-2014 were published in the journal 'Hemophilia'.

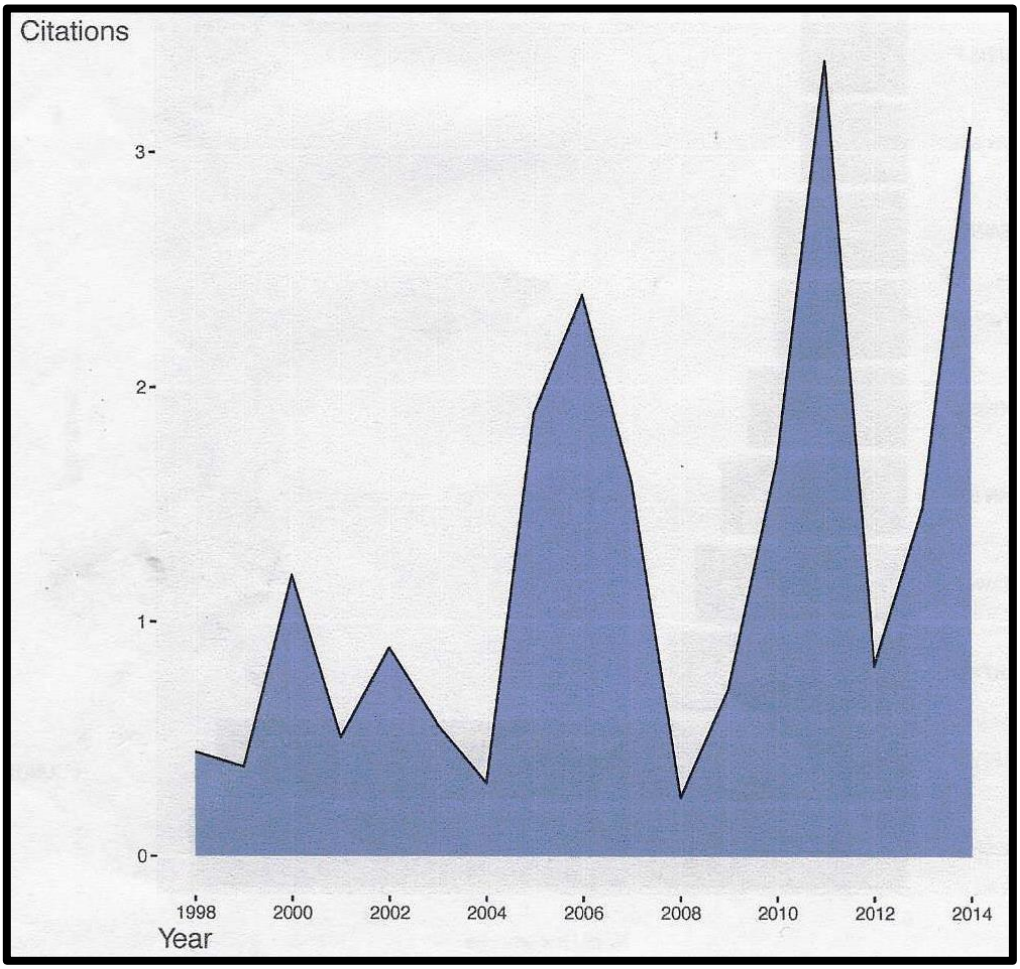
Figure 4.55: Average article citations per year received on articles published on Hemophilia A without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum articles cited during 2013 published on Hemophilia A without molecular biology from India during 1991-2014.

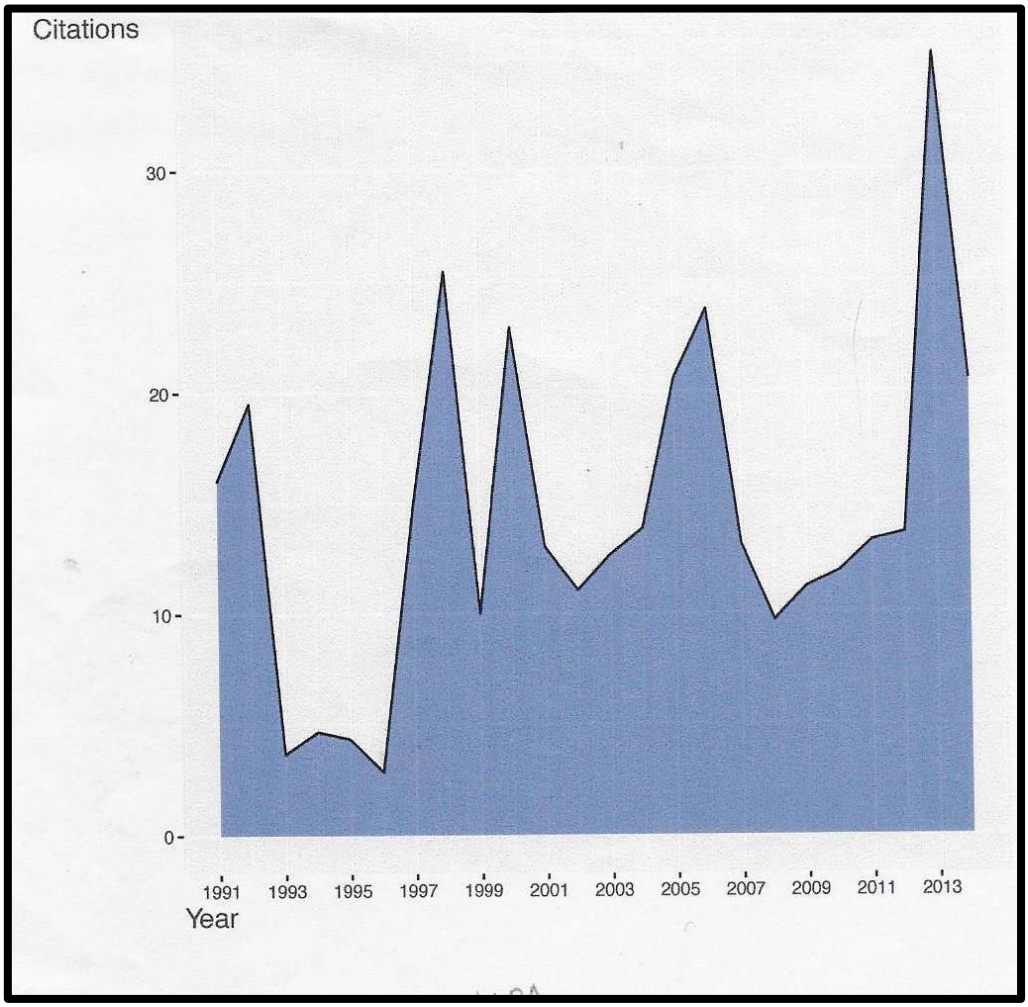
Figure 4.56: Average article citations per year received on articles published on Hemophilia A with molecular biology from India during 1991 -2014.



Source: SCOPUS database

Observation - Maximum articles were cited during 2010 followed by those cited during 2014 and 2006 published Hemophilia A with molecular biology from India during 1991-2014.

Figure 4.57: Average total citations per year received on articles published on Hemophilia A without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum total citations were received by articles during 2013 followed by those during 1997 and 2005 without molecular biology from India during 1991-2014.

Figure 4.57 depicts average total citations per year on articles published hemophilia A from India during 1991-2014. Totally 5 spikes were seen during different time periods. The major spike was seen during the period 2012-2014.

Figure 4.58 reveals average total citations per year on articles published on hemophilia A involving molecular biology aspects from India during 1991-2014. Totally 4 spikes indicating increase in total citations was seen during different time periods. Amongst these, the major peak was seen during 2004-2008.

Table 4.50 represents the list of most productive top ten authors expressed on the basis of fractionalized published articles on hemophilia A from India during 1991-2014. K. Ghosh ranked number one with the total weightage of 34.39 of the total articles published by him during this period.

Table 4.51 shows top ten articles published on hemophilia A from India during 1991-2014 and receiving maximum citations. An article published by Srivastava et al (2017) in the journal 'Hemophilia' occupied top position in the list as it received 810 total citations during the above mentioned period.

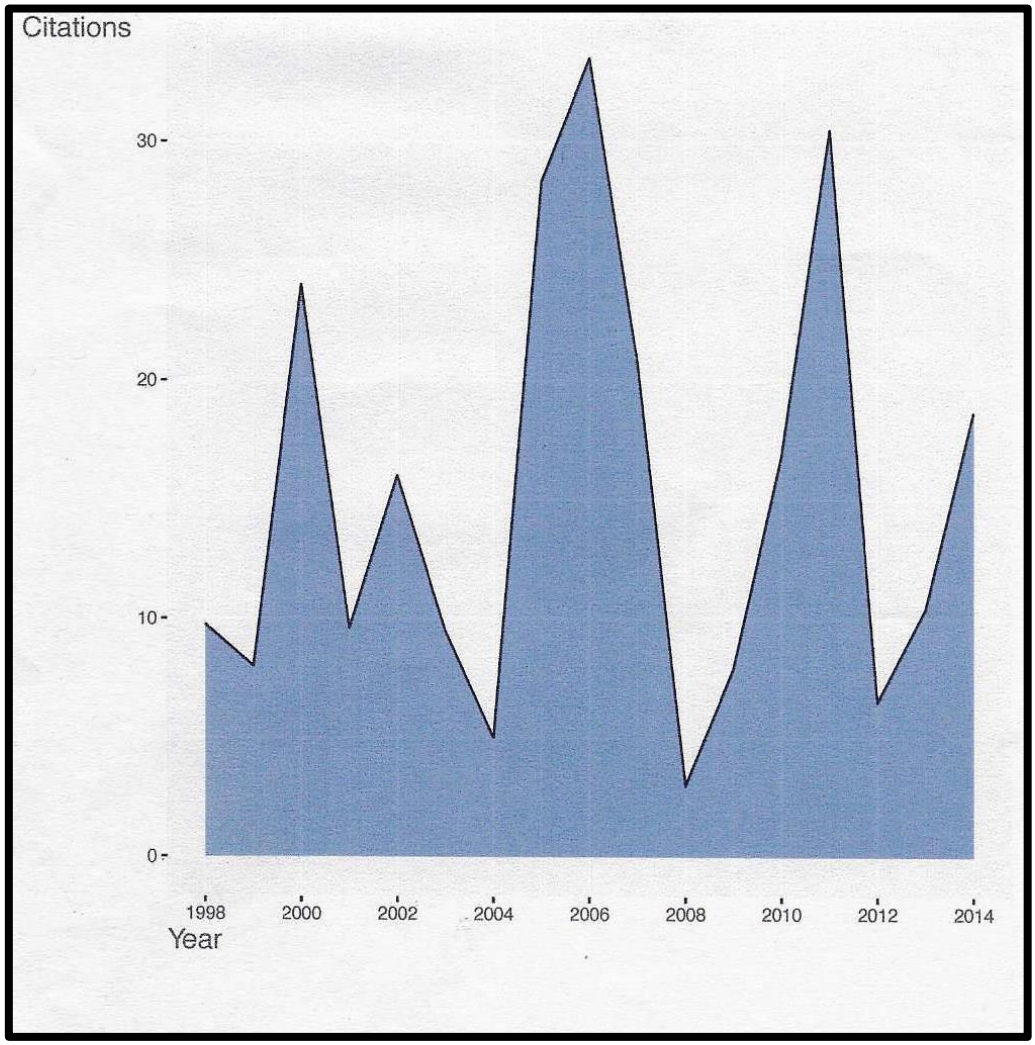
Table 4.52 reveals the list of most productive top ten authors expressed on the basis of fractionalized published articles on Hemophilia A involving molecular biology aspects from India during 1991-2014. In this list also K. Ghosh occupied number one with the total weightage of 7.70 of the total articles published by him during this period.

Table 4.53 represents top ten articles published on hemophilia A involving molecular biology aspects from India during 1991-2014 and receiving maximum citations. An article published by Saranya et al (2011) in the journal 'International Journal of Biological Macromolecules' ranked first in the list receiving 164 total citations during the above mentioned period.

4.2.6 Hemophilia B

Figure 4.59 depict network analysis diagram of coupling of authors of the articles published on Hemophilia B from India during 1991-2014. Two clusters were seen. The first cluster was a very compact cluster showing about 25 nodes. Amongst these showed nodes with equal prominence. These nodes were placed very close to each other indicating good collaboration among them. The second cluster represented 21 nodes with A. Srivastava occupying the prominent node.

Figure 4.58: Average total citations per year received on articles published on Hemophilia A with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation- Maximum total citations were received by articles during 2006 followed by those received during 2010 and 2000 with molecular biology from India during 1991-2014.

Table 4.50: Most productive top ten authors expressed on the basis of fractionalized published articles on Hemophila A without molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Ghosh K	34.39
2.	Shetty S	23.69
3.	Srivastava A	17.86
4.	Mohanty D	10.02
5.	Kar A	04.31
6.	Jijina F	04.51
7.	Chandy M	04.09
8.	Choudhry VP	03.96
9.	Saxena R	03.90
10.	Kabra M	02.86

Source : SCOPUS database

Explanation - Contribution of top ten most productive authors in publishing articles on HemophiliaA without molecular biology from India during 1991-2014.

Table 4.51: Top ten articles published on Hemophilia A without molecular biology from India during 1991-2014 received maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Srivastava A, 2013, Hemophilia	810	101.25
2	Mahlangu J, 2014, Blood	223	31.86
3	Kadir RA, 1998, BJOG Int J Obster Gynaecol	199	08.65
4	Blanchette VS, 2014, J Thromb Haemost	186	26.57
5	Powell JS, 2013, New Engl J Med	178	22.25
6	Saranya N, 2011, Int J Biol Macromol	164	16.40
7	Omata M, 2012, Hepatology International	133	14.78
8	Palta S, 2014, Indian J Anaesth	126	18.00
9.	Ghosh SS 2006, Appl Biochem Biotechnol	100	6.67
10.	Lacroix-Desmazes S, 2002, New Engl J Med	95	5.00

Source : SCOPUS database

BJOG, Int J Obster Gynecol - BJOG International Journal of Obstetrics and Gynaecology
J Thromb Hemost - Journal of Thrombosis and Hemostasis
New Engl J Med - New England Journal of Medicine
Int J Biol Macromol - International Journal of Biological Macromolecules
Indian J Anaesth - Indian Journal of Anaesthesia

Table 4.52: Most productive top ten authors expressed on the basis of fractionalized published articles on Hemophila A with molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Ghosh K	7.70
2.	Shetty S	7.45
3.	Mohanty D	2.19
4.	Husain N	2.12
5.	Quadros L	1.73
6.	Mittal B	1.28
7.	Pandey GS	1.28
8.	Srivastava A	1.10
9.	Basu B	1.00
10.	Chattopadhyay P	1.00

Source : SCOPUS database

Explanation - Contribution of top ten most productive authors in publishing articles on Hemophilia A with molecular biology from India during 1991-2014.

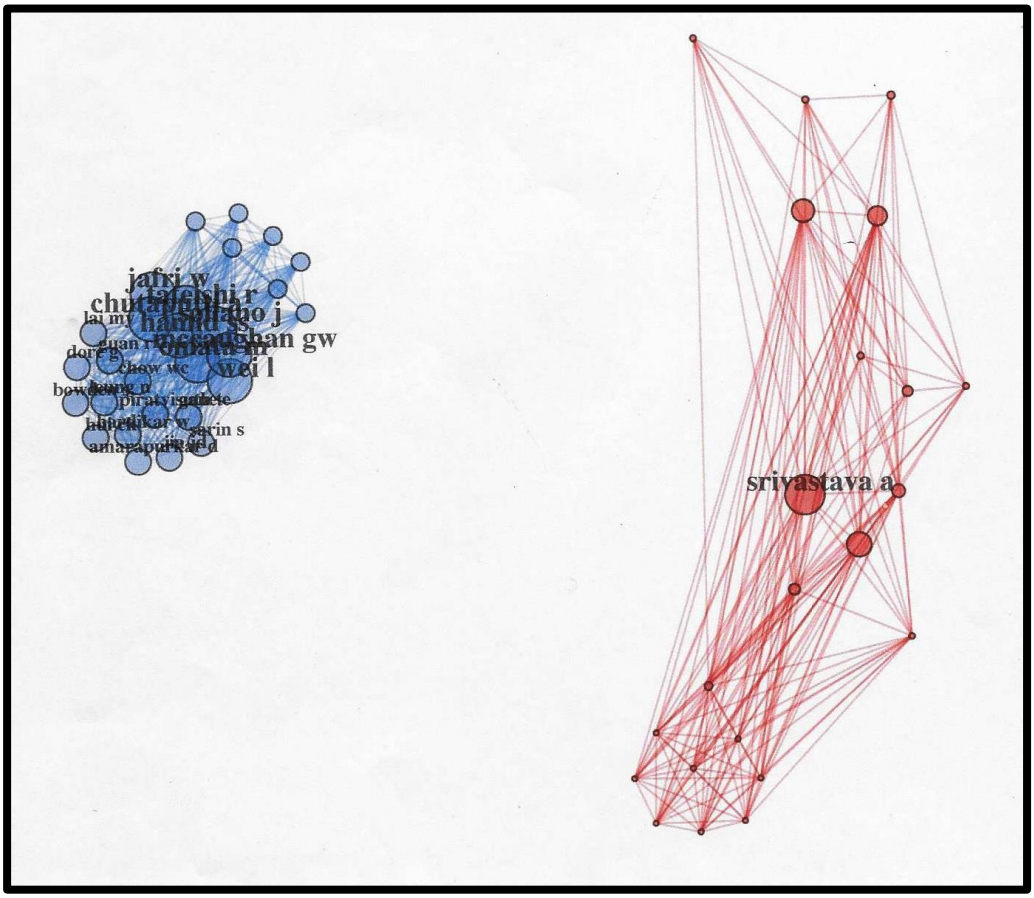
Table 4.53: Top ten articles published on Hemophila A with molecular biology aspects from India during 1991-2014 received maximum citations .

Sr. No.	Article	Total citations	Total citations per year
1	Saranya N, 2011,Int J Biol Macromol	164	16.40
2	Ghosh SS, 2006, Appl Biochem Biotechnol	100	6.67
3	Markusic DM, 2010, Molecular Therapy	79	7.18
4	Shetty S, 2007, Br J Haematol	58	4.14
5	Jayandharan G, 2005, Haemophilia	54	3.38
6	Santosh B, 2014, Biomed Res Int	36	5.14
7	Sen D, 2013,Human Gene Therapy Methods	26	3.25
8	Chowdhury MR, 2000, Haemophilia	24	1.14
9	Ahmed RPH, 2005, Haematologica	22	1.38
10	Brooks MB, 2003, Mammalian Genome	22	1.22

Source : SCOPUS database

Int J Biol Macromol - International Journal of Biological Macromolecules
Appl Biochem Biotechnol - Applied Biochemistry and Biotechnology
Br J Haematol - British Journal of Hematology
Bio Med Res Int - Biomed Research International

Figure 4.59: Coupling or collaboration of authors of the articles Published on Hemophilia B without molecular biology from India during 1991-2014.



Source : SCOPUS database

Observation - Tow clusters are seen. The first one is very compact

Cluster 1- 20 – 25 nodes, 4- 5 nodes show equal prominence indicating good collaboration

Cluster 2 – 22 nodes, Main node- A. Srivastava

Figure 4.60 reveals network analysis diagram of coupling of authors of the articles published on Hemophilia B involving molecular biology aspects from India during 1991-2014. Totally 4 clusters were seen. The first cluster showed 19 nodes with K. Ghosh and S. Shetty occupying main nodes with equal size. The second cluster showed 13 nodes with G.R. Jayandharan occupying main node. Again 13 nodes were seen in the third cluster. S.C. Nair occupied the main node. The fourth cluster was a small cluster with 5 nodes of equal size.

Figure 4.61 represents trend of articles published on hemophilia B from India during 1991-2014. Articles on this disorder were published from 1995 onwards and most of the articles were published in the journal 'Hemophilia'.

Figure 4.62 shows trend of articles published on hemophilia B involving molecular biology aspects from India during 1991-2014. Total number of articles published under this group was very small. Again many articles were published in the journal 'Hemophilia'.

Figure 4.63 reveals average article citations per year on articles published on hemophilia B from India during 1991-2014. Very fewer citations per article were received for the articles on hemophilia B during the above mentioned period. A sharp spike was seen during 2011-2013.

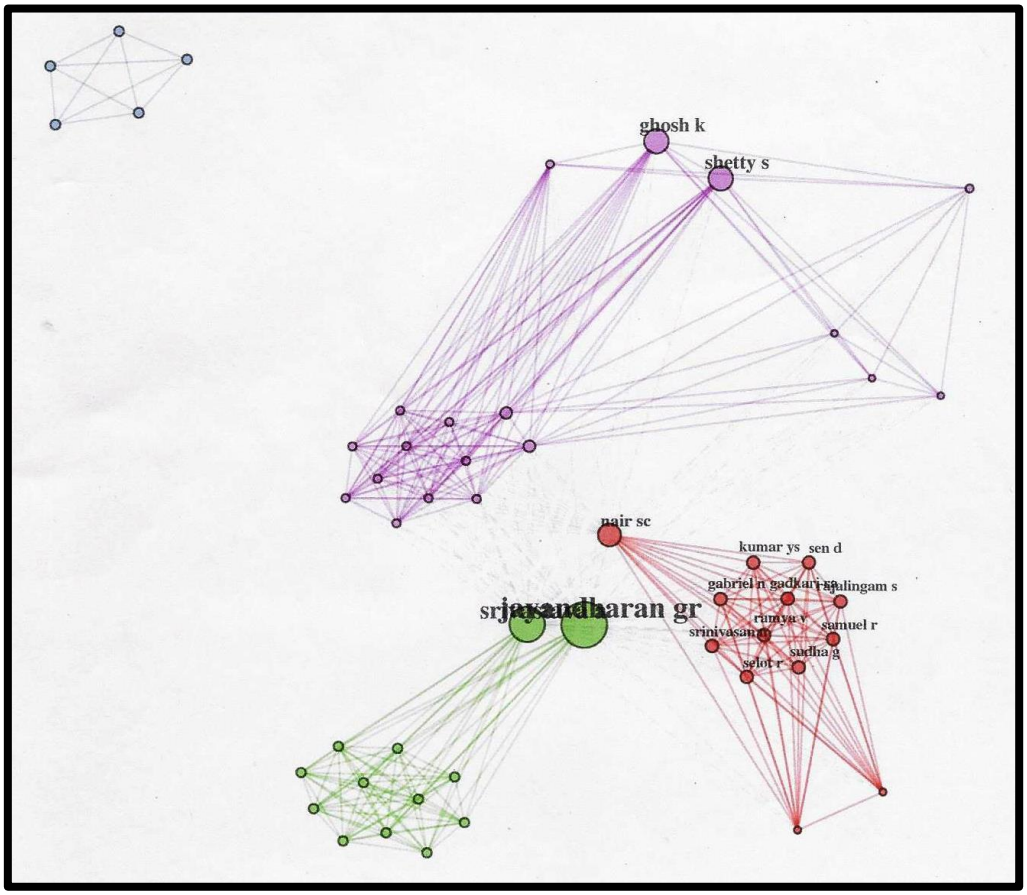
Figure 4.64 reveals average article citations per year on articles published on hemophilia B involving molecular biology aspects from India during 1991-2014, A sharp spike was seen during the period 2009-2012.

Figure 4.65 depicts average total citations per year on articles published on hemophilia B from India during 1991-2014. Several spikes were seen indicating good citations during the above mentioned time period. The biggest spike was seen during 2005-2009.

Figure 4.66 shows average total citations per year on articles published on hemophilia B involving molecular biology aspects from India during 1991-2014. A sharp spike was seen during 2009-2012.

Table 4.54 reveals the list of the most productive top ten authors expressed on the basis of fractionalized published articles on hemophilia B from India during 1991-2014. K. Ghosh occupied the first position with the weightage of 17.35 of the total articles published by him on this disorder during the above mentioned period.

Figure 4.60: Coupling or collaboration of authors of the articles published on Hemophilia B with molecular biology from India during 1991-2014.

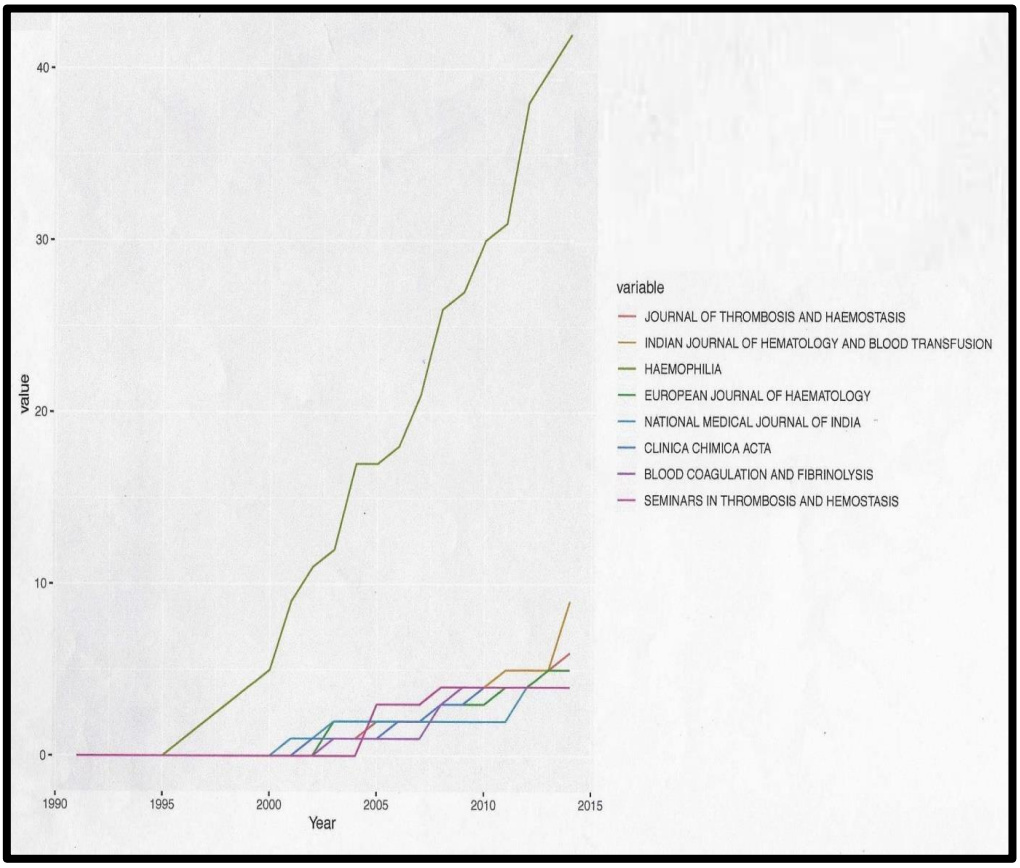


Source : SCOPUS database

Observation - Totally four clusters are seen

Cluster 1 - 19 nodes, Main nodes – K. Ghosh, S. Shetty
Cluster 2 - 13 nodes, Main node – G. R. Jayandharan
Cluster 3 - 13 nodes, Main node – S. C. Nair
Cluster 4 - 0 - 5 nodes with equal prominence

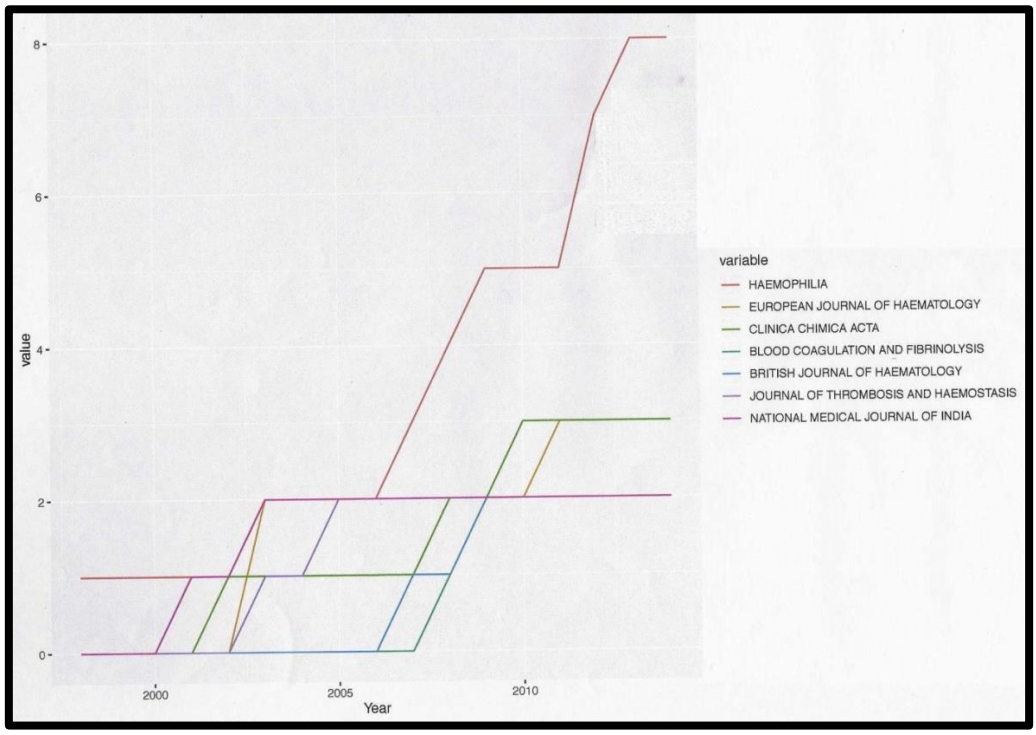
Figure 4.61: Top eight journals showing trend of articles published on Hemophilia B without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum articles on Hemophilia B without molecular biology from India during 1991-2014 were published in the journal 'Hemophilia'.

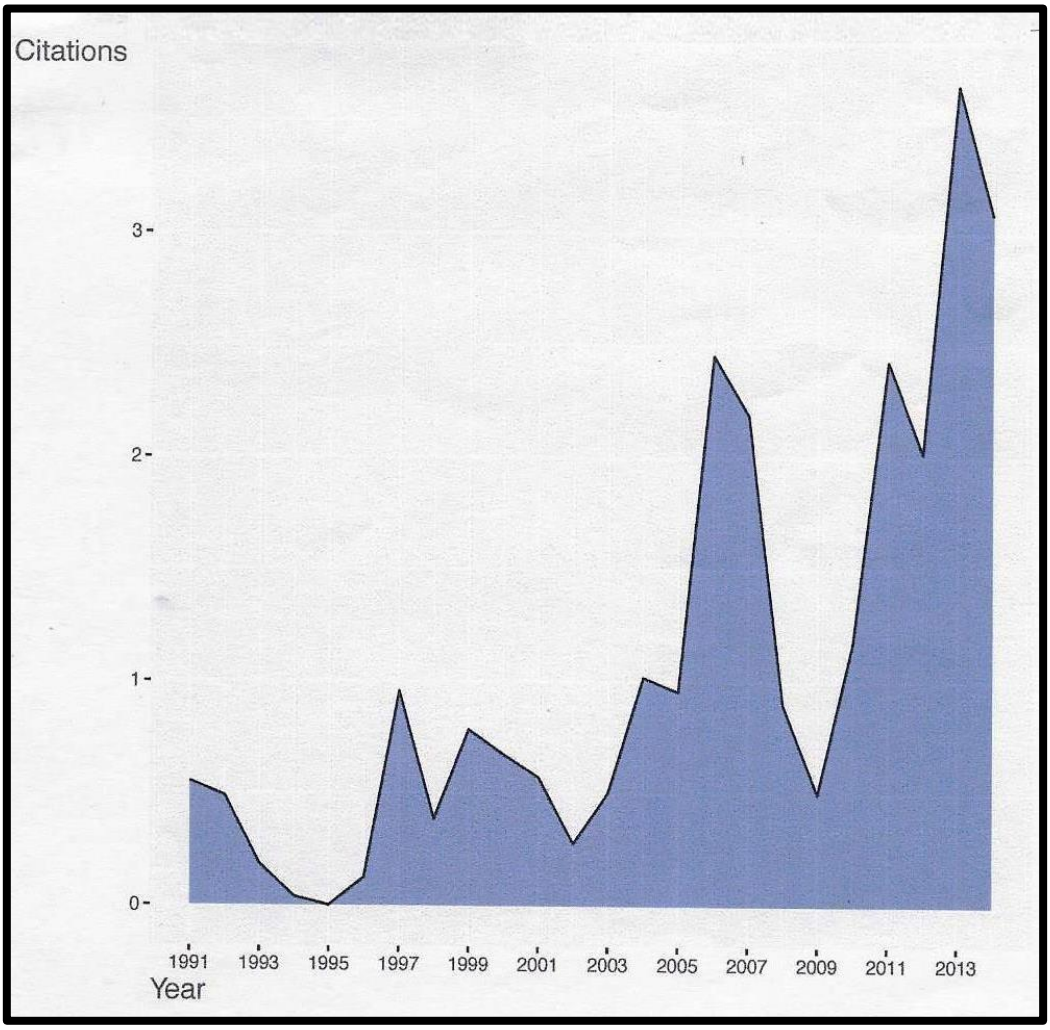
Figure 4.62: Trend of articles published in different journals on Hemophilia B with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum articles on Hemophilia B with molecular biology from India during 1991-2014 were published in the journal 'Hemophilia'.

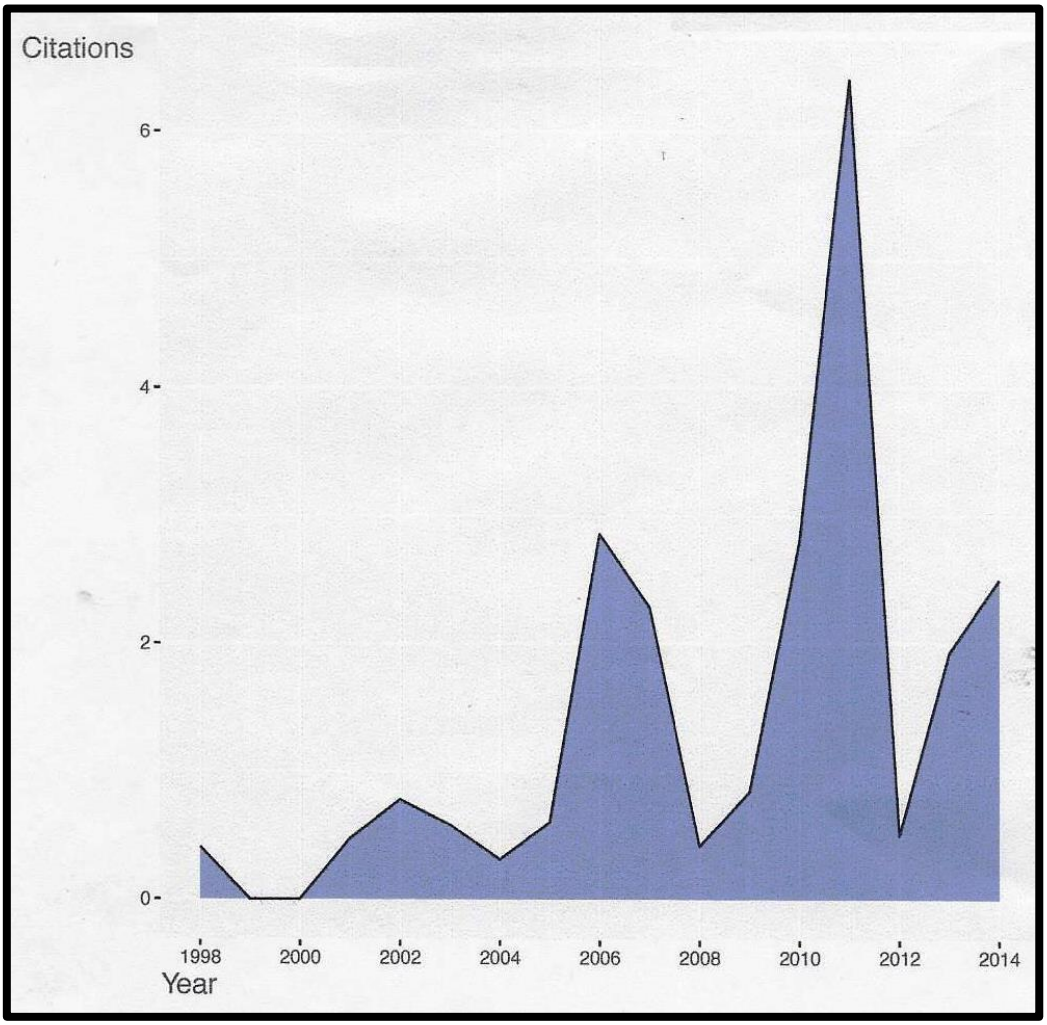
Figure 4.63: Average article citations per year received on articles published on Hemophilia B without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum articles were cited during 2013 followed by those cited during 2005 and 2011 published on Hemophilia B without molecular biology from India during 1991-2014.

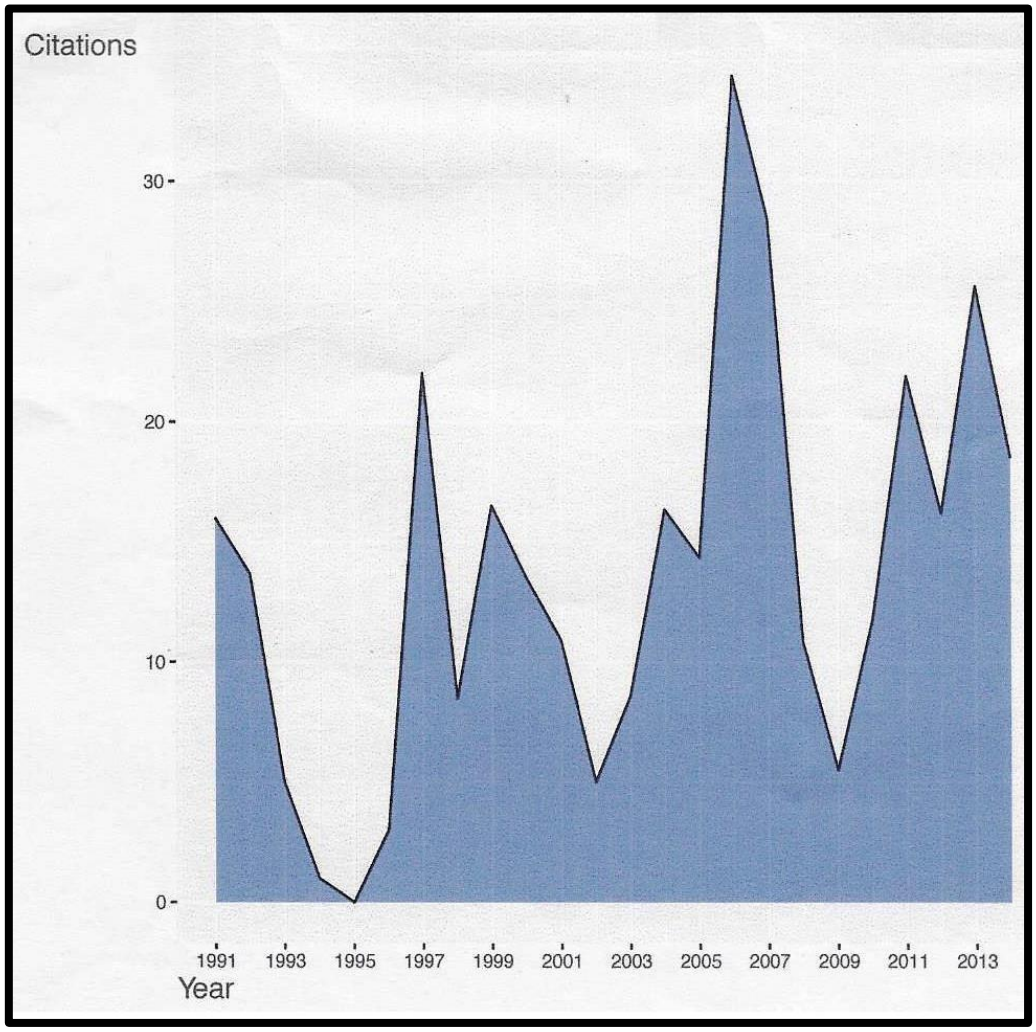
Figure 4.64: Average article citations per year received on articles published on Hemophilia B with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum articles were cited during 2010 published on Hemophilia B with molecular biology from India during 1991-2014.No articles were cited during 1999-2000.

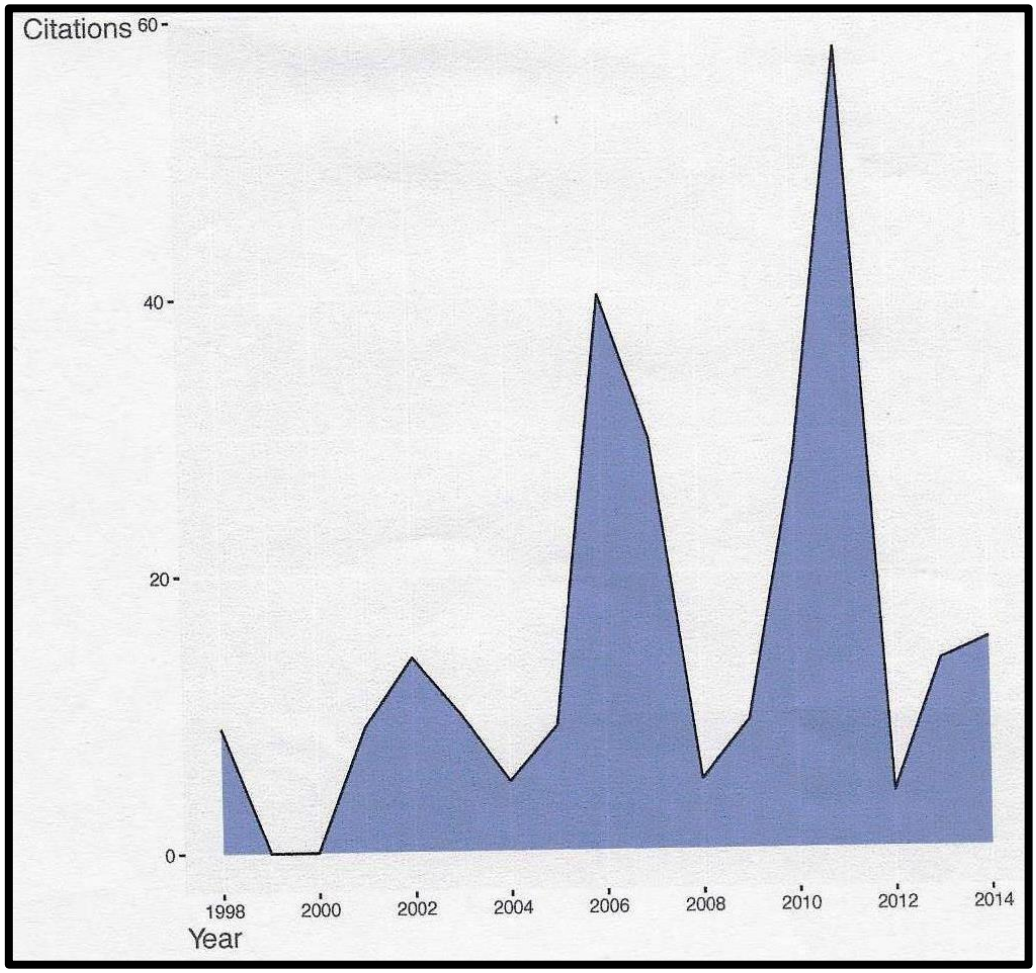
Figure 4.65: Average total citations per year received on articles published on Hemophilia B without molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum total citations were received by articles during 2005 followed by those received during 2013 and 1997 published on Hemophilia B without molecular biology from India during 1991-2014.

Figure 4.66: Average total citations per year received on articles published on Hemophilia B with molecular biology from India during 1991 -2014.



Source : SCOPUS database

Observation - Maximum total citations were received by articles 2010 followed by those received during 2006 published on Hemophilia B with molecular biology from India during 1991-2014.No citations were received for the articles published during 1999 – 2000.

Table 4.54: Most productive top ten authors expressed on the basis of fractionalized published articles on Hemophila B without molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Ghosh K	17.35
2.	Shetty S	11.40
3.	Srivastava A	5.27
4.	Mohanty D	4.28
5.	Chandy M	2.14
6.	Quadros L	2.06
7.	Kar A	2.00
8.	Choudhry VP	1.92
9.	Jijina F	1.88
10.	Jayandharan GR	1.82

Source : SCOPUS database

Explanation - Contribution of top ten most productive authors in publishing articles without molecular biology on hemophilia B from India during 1991-2014.

Table 4.55 depicts top ten articles published on hemophilia B from India during 1991-2014 and receiving maximum citations. An article by Blanchette et al (2014) published in the journal 'Journal of Thrombosis and Hemostasis' received 186 total citations during the above mentioned period and ranked first.

Table 4.56 shows the list of the most productive top ten authors expressed on the basis of fractionalized articles on hemophilia B involving molecular biology aspects from India during 1991-2014. Again K. Ghosh was on the top position with the weightage of 4.96 of the total articles published by him on hemophilia B involving molecular biology aspects during the above mentioned period.

Table 4.57 describes the list of top ten articles published on hemophilia B involving molecular biology aspects from India and receiving maximum citations. An article published by Saranya et al (2011) in the journal 'International Journal of Biological Macromolecules' received 164 total citations during the above mentioned period.

Table 4.55: Top ten articles published on Hemophilia B without molecular biology from India during 1991-2014 received maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Blanchette VS, 2014, J Thromb Hemost	186	26.57
2	Powell JS, 2013, N Engl J Med	178	22.25
3	Saranya N, 2011, Int J Biol Macromol	164	16.40
4	Mccaughan GW, 2007, J Gastroenterol Hepatol	144	10.90
5	Omata M, 2012, Hepatology International	133	14.78
6	Ghosh SS, 2006, Appl Biochem Biotechnol	100	6.67
7	Peyvandi F, 2006, Haemophilia	81	5.40
8	Markusic DM, 2010, Molecular Therapy	79	7.18
9.	Shetty S, 2011, Autoimmunity Reviews	68	6.80
10.	Lundin B, 2012, Haemophilia	61	6.78

Source : SCOPUS database

J Thromb Hemost - Journal of Thrombosis and Hemostasis
N Engl J Med - New England Journal of Medicine
Int J Biol Macromol - International Journal of Biological Macromolecules
J Gastroenterol Hepatol - Journal of Gastroenterology and Hepatology
Appl Biochem Biotechnol - Applied Biochemistry and Biotechnology

Table 4.56: Most productive top ten authors expressed on the basis of fractionalized published articles on Hemophila B with molecular biology from India during 1991-2014.

Sr. No.	Authors	Articles fractionalized
1.	Ghosh K	4.96
2.	Shetty S	4.96
3.	Quadros L	1.73
4.	Mohanty D	1.62
5.	Chattopadhyay P	1.00
6.	Srivastava A	0.84
7.	Jayandharan GR	0.82
8.	Kulkarni B	0.64
9.	Ray K	0.62
10.	Chowdhury MR	0.53

Source : SCOPUS database

Explanation - Contribution of top ten most productive authors in publishing articles with molecular biology on Hemophilia B from India during 1991-2014.

Table 4.57: Top ten articles published on Hemophila B with molecular biology from India during 1991-2014 received maximum citations.

Sr. No.	Article	Total citations	Total citations per year
1	Saranya N, 2011, Int J Biol Macromol	164	16.40
2	Ghosh SS, 2006, Appl Biochem Biotechnol	100	6.67
3	Marksic DM, 2010, Molecular Therapy	79	7.18
4	Shetty S, 2007, Br J Haematol	58	4.14
5	Sen D, 2013, Hum Gene Ther Methods	26	3.25
6	Brooks MB, 2003 Mammalian Genome	22	1.22
7	Shetty S, 2007, Haemophilia	21	1.50
8	Jayandharan GR, 2009, Haemophilia	18	1.50
9.	Shetty S, 2006, Prenatal Diagnosis	18	1.00
10.	Jayandharan G, 2003, J Thromb Hemost	18	1.00

Source : SCOPUS database

Int J Biol Macromol - International Journal of Biological Macromolecules
Appl Biochem Biotechnol - Applied Biochemistry and Biotechnology
Br J Haematol - British Journal of Hematology
Hum Gene Ther Methods - Human Gene Therapy Methods
J Thromb Hemost - Journal of Thrombosis and Hemostasis

Chapter V

Findings Suggestions and Conclusion

5.1 Findings

In the present study the research output on six hematological /genetic disorders from India has been evaluated for the past 60-70 years, (i.e. from the first publication on each disorder recorded in PUBMED up to 2014) specifically to see the impact of molecular biology on the publication of articles on these disorders especially from 1991 onwards. It is the first study of this type which has been carried out on six genetic disorders. So no earlier references are available to compare our data. In all the earlier studies of this type research output on particular disease or particular subject was evaluated using the time interval of 10 or 15 years. However in the present study the articles have been analyzed from the first publication on each disorder which has been listed in the PUBMED database up to 2014. Therefore time interval has been different for each disorder ranging between 60-75 years. No scientometric study has been published on Down syndrome and G6PD deficiency literature from India so far. Vellaichamy and Jayshankar (2018) analyzed hemophilia literature (data on hemophilia A and hemophilia B was not separately considered) from India published during 2003-2017 . Gupta (2012) performed scientometric analysis of research output on five genetic disorders (sickle cell anemia, hemophilia, vonWillebrand disease, thalassemia and thrombopenia) published during 2002-2011 using different parameters. In a vast country like India he found that very few (only 10) research institutes contributed more than 60% of the total number of articles published on the above mentioned five hereditary disorders during 2002-2011. He concluded that there is an urgent need to develop sufficient infrastructure to handle the problems related to these hereditary disorders.

Now we will discuss some reasons regarding the big difference seen in the number of publications on six genetic disorders during two time intervals (i.e. up to 1990 and during 1991-2014). The first publication on G6PD deficiency appeared from India in 1963 while the first publication on sickle cell anemia was reported in 1952. Both these disorders were considered as 'genetic markers' in population genetic studies. So many institutes and scientists engaged in screening of different population groups using various genetic markers also looked for G6PD deficiency and prevalence (presence) of sickle hemoglobin. This data was subsequently published. Therefore 117 publications on G6PD deficiency and 101 publications on sickle cell anemia appeared up to 1990. These are relatively good numbers because during 1991-2014 118 and 243 articles were published from India on G6PD

deficiency and sickle cell anemia respectively. In the case of other disorders the picture is different. First publication on Down syndrome from India appeared in 1963 and only 32 articles were published up to 1990. (i.e. during next 27 years). Because only 3-4 institutes in our country were working on this disorder at that time. Subsequently many medical colleges and institutes started working on this disorder in India and this resulted in increase in the number of publications (192) on Down syndrome considerably during second time interval (1991-2014). Hemophilia A, Hemophilia B and von Willebrand disease are diseases related to blood clotting or coagulation process and very few reports have been published on these disorders up to 1990 (range 7-13 publications). This can be correlated to lack of awareness about these disorders among the people, absence of patient organizations who convince the patient families and their relatives to get tested and lack of interest among various institutes working on these disorders; as these disorders require technically difficult and /or demanding laboratory studies. Similarly laboratory reagents to carry out tests for these disorders were costly and not easily available before 1980. Therefore these disorders wee sparingly studied with few publications up to 1990. During the year 1994, a separate department working on various coagulation disorders (mainly hemophilia A, hemophilia B, von Willebrand disease) was established at ICMR's National Institute of Immunohamatology, Mumbai. Scientists from this department started working on these disorders in a big way. Other eminent scientists like Dr. M. Chandy, Dr. Renu Saxena from our country were also working on this area. Coincidently the journal 'Hemophilia' which is the official journal of World Federation of Hemophilia (WFH) was started at Blackwell Science in 1993 while the journal 'Journal of Thrombosis and Hemostasis' was founded in 2003. So all these scientists got a specific platform to publish their articles on coagulation disorders in these journals. This can be seen in figures 45, 53, 54, 61, 62. Therefore there was a huge increase in the number of articles published on the above mentioned three disorders. Quantitatively it can be seen in Table 4.30 where the growth rate of publications on these three disorders during two time intervals has been calculated. It is 165% for hemophilia B, 209.84% for hemophilia A and 685.7% for von Willebrand disease.

In the present study network analysis of author collaborations in India clearly shows that good collaboration at national level is required to conduct advanced research using molecular biology tools. It has also been observed that whatever

articles have been published by the scientists on the six genetic disorders; more than 90% of the articles are showing collaborative work (Tables 36 and 37) as they are multiauthored articles. This clearly shows the importance of collaborative work. This should be further strengthened to improve the number of publications.

Network analysis also shows that scientists from ICMR-National Institute of Immunohematology, Mumbai (K. Ghosh, S. Shetty, R.B. Colah, D. Mohanty, M.B. Mukherjee, A.H. Nadkarni) were involved in conducting the major research work on five out of six genetic disorders evaluated in the present study. This is clearly seen in figures 19, 20, 35, 36, 43, 44, 51, 52, 59 and 60 where they form the major nodes. Scientists from Christian Medical College, Vellore (A. Srivastava, S.C. Nair, G.R. Jayandharan and M. Chandy) were mainly involved in conducting the major research work on coagulation disorders (Hemophilia A, Hemophilia B and von Willebrand disease). This is clearly seen in figures 43, 44, 51, 52, 59 and 60 where they form the major nodes. Three scientists from Regional Medical Research Centre for Tribals, Jabalpur were involved in conducting major work sickle cell anemia which is seen in figures 19 and 20 where they form the major nodes. One of the scientists from All India Institute of Medical Sciences, New Delhi (R. Saxena) was involved in conducting the major research work on sickle cell anemia and von Willebrand disease. This is seen in figures 19, 43 and 44 where the scientists form the major node. Thus in a big country like India only three to four major research institutes have been working on these genetic disorders. This is one of the reasons for having less number of publications on these disorders from India. This has clearly emerged out from the present study. This situation should be improved.

This type of study provides knowledge about pattern of publications of articles on various research aspects studied on these disorders and also help to understand the lacunae present in the current situation. This will help the scholars to design projects to fill up these lacunae. It will also help the policy makers and funding agencies to sanction grants to such projects. These findings are listed as follows-

1)The present study evaluated the research output of six genetic disorders viz. Sickle cell anemia, G6PD deficiency, Down syndrome, Hemophilia A, Hemophilia B and von Willebrand disease This study has helped us to understand the impact of molecular biology on the publications on these disorders from India as well as from all over the world.

2) This is a very comprehensive study where the research output on six genetic disorders published during last 60-70 years has been analyzed. This type of study has been attempted for the first time in our country.

3) A Scientometric study of research output on Down syndrome and G6PD deficiency was not done earlier. In the present study it is done for the first time. Similarly analysis of the publications on Hemophilia A and Hemophilia B has been attempted separately.

4) This study presents journal-wise , year-wise and category-wise analysis of the articles. Similarly it shows authorship pattern, most productive authors, publication pattern and highly cited articles of six genetic disorders.

5) Totally 1,175 articles from India and 60,826 articles from all over the world on six genetic disorders were evaluated. This accounts for 1.90% of India's share in the global publications on these disorders.

6) 276 articles were published up to 1990 from India on the above mentioned six genetic disorders as compared to 24,710 articles published from all over the world.

7) During 1991-2014, 899 articles were published from India on six genetic disorders as compared to 36,116 articles published from all over the world.

8) Category-wise analysis of the articles published on the six genetic disorders has been done. The articles are separated into following six categories viz. original / research articles, articles based on clinical findings of the patient, case reports, reviews on various aspects of the particular disorder, miscellaneous articles describing non-scientific aspects of the particular disorder and finally articles published using various molecular biology techniques. This has helped us to get clear idea about publication pattern on these disorders over the years. This type of data is useful to the scientists working on these disorders and library professionals who can provide such data to research scholars.

9) The most productive years of publications have been identified during different time intervals. They were different for different disorders.

10) Using network analysis collaborative pattern among the authors from India who published articles on six genetic disorders is established.

11) The most productive top ten authors from India who published maximum articles on each of the six genetic disorders have been identified.

12) Top ten articles published on each of the six genetic disorders from India and received maximum citations have been identified.

5.2 Testing Hypothesis

Hypothesis - "There may be impact of molecular biology on publications of six genetic disorders (viz. Down syndrome, G6PD deficiency, Hemophilia A, Hemophilia B, Sickle cell anemia, von Willebrand Disease) published during 1991 to 2014".

Testing :

Articles published on six genetic disorders during 1991 – 2014 from India as well as all over the world were separated into two groups; articles published during 1991- 2002 and articles published during 2003 – 2014 to see growth rate of publications on these disorders. Articles from both the groups were further separated into six different categories (viz. Original articles, Articles based on clinical findings, Case reports, Reviews, Miscellaneous articles and Articles published using molecular biology tools). Data from category 6 (Articles published using molecular biology tools) was considered to test the hypothesis.

It was observed that 18.23- 42.4% of the articles published on six genetic disorders during 1991-2014 from India as well as from all over the world were under category 6. Chi square test of contingency was applied to see the growth rate and also to test the hypothesis. It was non-significant for all the disorders in India. When articles published from all over the world under category 6 were compared for two time intervals (1991-2002 and 2003-2014), articles published on Down syndrome and Hemophilia B showed statistically significant values. It shows that number of articles published on these two disorders under category 6 has increased significantly during second time interval (2003-2014).

Above results show that growth in number of articles published on six genetic disorders from India is steady during 1991-2014. However in the case of all over the world, articles on Down syndrome and Hemophilia B showed statistically significant increase in numbers.

It proves that there is an impact of molecular biology on publications on six genetic disorders published during 1991-2014.

Table 5.1 - X^2 contingency table to compare articles published under category 6 (Articles published using molecular biology tools) during two time intervals viz. 1991-2002, 2003-2014

1) India

Genetic disorders	X^2	p Value	Significance
G6PD deficiency	6 x 6 = 0.27	p = 0.6050	Non significant
Down syndrome	6 x 6 = 2.332*	p = 0.1268	Non significant
Hemophilia A	6 x 6 = 0.520	p = 0.4707	Non significant
Sickle cell anemia	6 x 6 = 0.042	p = 0.8383	Non significant
Hemophilia B	6 x 6 = 1.823*	p = 0.1769	Non significant
von Willebrand disease	6 x 6 = 0.309	p = 0.5784	Non significant

*- with Yate's correction

X^2 test done for one degree of freedom

2) All over the world

Genetic disorders	X^2	P Value	Significance
G6PD deficiency	6 x 6 = 0.217	p = 0.6414	Non significance
Down syndrome	6 x 6 = 23.6946	p < 0.0001	Significant
Hemophilia A	6 x 6 = 0.0161	p = 0.8991	Non significant
Sickle cell anemia	6 x 6 = 3.6662	p = 0.0557	Non significant
Hemophilia B	6 x 6 = 20.1380	p < 0.0001	Significant
von Willebrand disease	6 x 6 = 1.5440	p = 0.2140	Non significant

X^2 test results are considered statistically significant when p value is < 0.05

5.3 Findings met the objectives of the research

The above mentioned findings clearly show that they met the objectives of this study. Based on these findings following conclusions have been drawn.

5.4 Conclusions

1) India's share in the global publications on individual disorders varies between 1.01% to 3.39%. The maximum share of 3.39% was seen in the case of G6PD deficiency. The articles published during 2015-2020 on these disorders showed that the Indian share in the global publications is increased and varies between 1.92-8.65%.

2) The range of articles published on Down syndrome, G6PD deficiency and sickle cell anemia during 1991-2014 using molecular biology based tools varies between 18.23-21.81%; while in the case of hemophilia A, hemophilia B and von Willebrand disease which are all coagulation related disorders i.e. related to blood clotting, it varies between 28.57- 42.40% suggesting a lot of development in laboratory facilities to generate data involving various molecular biology related aspects on these disorders. In fact among these disorders von Willebrand disease was under-diagnosed up to 2005 but later on there was a quick improvement and 42.4% of the total articles were published on molecular biology based aspects.

3) However the articles published from all over the world did not show such difference in the case of six genetic disorders. Here the range of articles published on all the six disorders using molecular biology based tools various between 18.23-26.91%.

4) When the growth rate seen in publications on six genetic disorders from India during two time intervals (viz. 1991-2002 and 2003-2014) and compared with that from all over the world for the same time intervals, it is observed that it is very high (range 107.6-685.71%) for the five disorders. In the case of G6PD deficiency it is only 31.37%. Global growth rate ranges between 26.11-64.38% for all the disorders. This suggests that work in India on these disorders picked up considerably during second time interval.

5) Author collaboration in India is evaluated by network analysis. The data suggests that author collaboration in India is not satisfactory to develop advanced research work on these disorders. This has been also pointed out by Sharma et al (2019); they say that the average citation per publication received by India was less as compared

to other developed counties . Data retrieved from SCOPUS shows that only 10% of the articles on these disorders are single authored, while 90% of the articles multi authored indicating that they are written on collaborative work. However, number of such articles are far less as compared to those published from all over the world. This analysis also shows that in a vast country like India only 3-4 main research institutes like ICMR National Institute of Immunohematology, Mumbai, All India Institute of Medical Sciences, New Delhi, Christian Medical College, Vellore etc. are working on these disorders.

6) The information on top five journals publishing maximum number of articles on above mentioned six genetic disorders is helpful to the libraries of the institutes who are working on these disorders. This will help the librarians to order these most prominent journals which will give access to the scholars to understand the current status of these disorders and to design the projects in such a way to fill up the lacunae.

7) The output of this work will be useful to the librarians who are working in the institutes where the research work on these disorders is being carried out. They can provide this information to their scientists who can design their work accordingly.

5.5 Suggestions

Network analysis shows that authors collaboration is not satisfactory in India. This situation should be changed. More and more scientists from different institutes should collaborate in research work on these disorders to improve the research output.

This information will be useful to policy makers and various research institutes to design the future projects. . Similarly using this information funding agencies can precisely fund the projects which will be useful to fill up the gaps in the research on these disorders.

5.6 Current scenario

The articles published up to 2014 on six disorders have been analysed during the present study. To understand the current scenario, articles published on these disorders during 2015-2020 were retrieved from PUBMED database and the trend of publications was observed. The articles published during 2015-2020 on these disorders showed that the Indian share in the global publications is increased marginally and varies between 1.92 – 8.65%. However still lot of efforts are required by the Indian scientists to increase this share substantially.

5.7 Further research

Considering the findings and suggestions, researcher has noted some points which can be considered for further research for future scholars who would like to undertake the similar topic in other genetic disorders because this is a new area of research.

1. Studies can be conducted on other genetic disorders like Thalassemia, Cystic Fibrosis, Duchene Muscular Dystrophy, Huntington Disease etc.seen in India.
2. Such type of Scientometric analysis can be used to see the growth rate of publications.
3. It can also useful to know the trend of publications and to identify the gaps to understand the disease process.

APPENDIX

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