

# Mapping of India's Contribution on "Down Syndrome" During 40 Years From 1973-2012

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## ABSTRACT

Down syndrome, a genetic disease, is commonly diagnosed congenital malformation/mental retardation syndrome occurring in people of all races and economic levels. The present study is aimed to examine the contribution of Indian scientists on Down syndrome during the 40 years span from 1973-2012. The study analyses the Indian share in the research output, contribution and citation impact of top Indian institutions, most prolific Indian authors, top journals for publication, top collaborating countries, number of citations received and the highly cited papers in the Indian research on Down syndrome.

**Keywords:** Down syndrome; Trisomy 21; Bibliometrics; Indian research output

## 1. INTRODUCTION

Down syndrome (DS) or Down's syndrome, also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. It is the most commonly diagnosed congenital malformation/mental retardation syndrome (Jones, 2006). The additional genetic material alters the course of development and causes the characteristics associated with Down syndrome. A few of the common physical traits of Down syndrome are low muscle tone, small stature, an upward slant to the eyes, and a single deep crease across the center of the palm - although each person with Down syndrome is a unique individual and may possess these characteristics to different degrees, or not at all (NDDS, n.d.). Some of the complications due to Down syndrome, as given in Merck Manual Home Health Handbook for Patients and Caregivers, are shown in Table 1.

**Table 1.** Some complications of Down syndrome.

Body System	Complication*
Brain	Intellectual disability (mild to severe) Autistic behavior Delayed speech and motor skills Alzheimer disease
Digestive tract	Malformed intestines Hirschsprung disease

	Celiac disease
Endocrine system	Hypothyroidism Diabetes
Eyes and ears	Cataracts, glaucoma, crossed eyes (strabismus) Hearing loss Ear infections
Growth	Short stature Obesity
Heart	Abnormally formed heart chambers
Muscles and bones	Unstable connections between the first 2 bones in the neck Loose joints
*Not all complications are present in each person	

Down syndrome is estimated to affect 1 in 750 live births (Jones, 2006); however, several reports have indicated variability in the estimates of Down syndrome among different ethnic groups. According to WHO, the estimated incidence of Down syndrome is between 1 in 1,000 to 1 in 1,100 live births worldwide. In India, the reported incidence of Down syndrome is around 1 in 1250 live births (Verma, 2006). According to an article published in the leading newspaper Deccan Herald, every year between 23,000 and 29,000 children are born in India with Down syndrome.

Down syndrome occurs in people of all races and economic levels, though older women have an increased chance of having a child with Down syndrome. A 35 year old woman has about a one in 350 chance of conceiving a child with Down syndrome, and this chance increases gradually to 1 in 100 by age 40. At age 45 the incidence becomes approximately 1 in 30. The age of the mother does not seem to be linked to the risk of translocation. To study the effect of advanced maternal age on Down syndrome many hypotheses have been formulated. Initially Penrose identified that advanced maternal age as risk for Down syndrome birth (Penrose 1933, 1934) and postulated that the maternal age dependent increase in birth rate of Down syndrome is in some way associated with the non-disjunction mechanism. The biological ageing hypothesis was originally proposed by Brook et al (1984), the central idea of which was that the increasing rate of meiotic errors and subsequent aneuploid birth is related to 'biological aging' of ovary not to the chronological age of women. Ghosh et al. (2010), proposed 'genetic aging' hypothesis which states that some of the mothers who have Down Syndrome baby are genetically older than the mothers of same chronological age who have euploid baby.

## 2. OBJECTIVES OF THE STUDY

The main objective of the present study is to analyse India's research productivity on Down syndrome during 1973-2012. The specific objectives of the study include:

- To find out Indian share in the research output on Down syndrome.
- To study the contribution and citation impact of top Indian institutions conducting research on Down syndrome.
- To find out the most prolific Indian authors conducting research on Down syndrome.
- To study the various source journals and the top journals preferred for publication.

- To find out the top countries collaborating with India for research on Down syndrome.
- To analyse the citation profile of the papers and to find out highly cited papers.

### 3. MATERIALS AND METHODS

The present study is based on the publications data on Down syndrome by Indian research community retrieved from Scopus database during November 2013. Scopus is the world's largest abstract and citation database of peer-reviewed literature. It covers nearly 20,500 titles from over 5,000 international publishers, of which 19,500 are peer-reviewed journals in the scientific, technical, medical, and social sciences (including arts and humanities). For retrieving relevant information for the present study the phrases "Down's syndrome", "Down syndrome", "Downs syndrome", "Trisomy 21" were used in the TITLE-ABS-KEY search option and "India" in the AFFILCOUNTRY option. The data was selected for the 40 years time period from 1973-2012. The results were refined to obtain relevant data regarding authors, institutions, source journals, etc. A different search strategy was used for finding the international collaborations and the results obtained were analyzed manually to obtain relevant results. The citations received by the article were taken as the citations which any article received since its publication. However, Scopus does not provide complete citations before 1996. This may be considered the limitation of the present study.

### 4. ANALYSIS AND INTERPRETATION

#### 4.1. Global Publication Share

The global publication share of top 15 most productive countries on Down syndrome research varies from 1.20% to 28.34% during 1973-2012. United States tops the list with 8144 publications with a share of 28.34% of total publications. Second rank is held by United Kingdom with 3680 publications and a share of 12.81%. Italy ranks third (4.98%) and France fourth (4.57%). These are followed by Japan (3.83%, 5<sup>th</sup> rank), Germany (3.73%, 6<sup>th</sup> rank), Canada (3.53%, 7<sup>th</sup> rank), Spain (3.07%, 8<sup>th</sup> rank), Netherlands (2.66%, 9<sup>th</sup> rank) and Australia (2.47%, 10<sup>th</sup> rank). Israel, India, Switzerland, Brazil and Sweden ranks 11<sup>th</sup> to 15<sup>th</sup> respectively (with publication share from 1.20% to 1.73%). Thus, India holds the 12<sup>th</sup> rank with 405 (1.41%) publications.

The overall publication output of maximum countries shows an increasing trend from 1973-2012 except for some countries which showed a decline at some point of time. United Kingdom had a decline in its share from 16.14% in 1993-2002 to 12.62% in 2003-2012. Japan, Israel, Switzerland and Sweden also had a decline in their share from 1993-2002 to 2003-2012. Germany, Australia, India, Switzerland and Brazil showed a decline in their publication share from 1973-1982 to 1983-1992. Maximum countries had an increase in their publication share from 1983-1992 to 1993-2002 but the highest increase was in publication share of Spain from 0.88% to 3.26%. United States and United Kingdom have retained their 1<sup>st</sup> and 2<sup>nd</sup> rank respectively throughout from 1973-2012.

The average citation per paper (ACPP) is the highest for Switzerland (23.84), followed by United States (23.61), United Kingdom (21.46), Germany (20.5), Australia (20.26) and Sweden (20.19) as shown in Table 2. It was lowest for India (4.14). The h-index for Down syndrome publications was highest for United States (157), followed by United Kingdom (106) and Germany (68). It was low for Brazil (25) and India (19).

**Table 2.** Publication output of top 15 countries on Down syndrome research.

Rank	Country	TP					TC	ACPP	h-index
		1973-1982	1983-1992	1993-2002	2003-2012	1973-2012			
1	United States	936	1416	2297	3495	8144	192269	23.61	157
2	United Kingdom	290	577	1308	1505	3680	78962	21.46	106
3	Italy	126	198	415	693	1432	19125	13.36	60
4	France	141	191	376	606	1314	17295	13.16	59
5	Japan	127	189	379	407	1102	17604	15.97	61
6	Germany	174	150	259	484	1067	21871	20.50	68
7	Canada	104	157	268	486	1015	19930	19.64	61
8	Spain	33	43	264	541	881	9828	11.16	49
9	Netherlands	33	82	202	448	765	13025	17.03	56
10	Australia	79	94	180	357	710	14382	20.26	51
11	Israel	35	79	166	218	498	7159	14.38	42
12	India	27	31	59	288	405	1676	4.14	19
13	Switzerland	36	25	149	182	392	9344	23.84	52
14	Brazil	24	12	55	300	391	3032	7.75	25
15	Sweden	40	70	107	127	344	6945	20.19	40
	<b>World</b>	<b>3815</b>	<b>4895</b>	<b>8106</b>	<b>11922</b>	<b>28738</b>			

*TP= Total papers; TC= Total citations; ACPP= Average citation per paper*

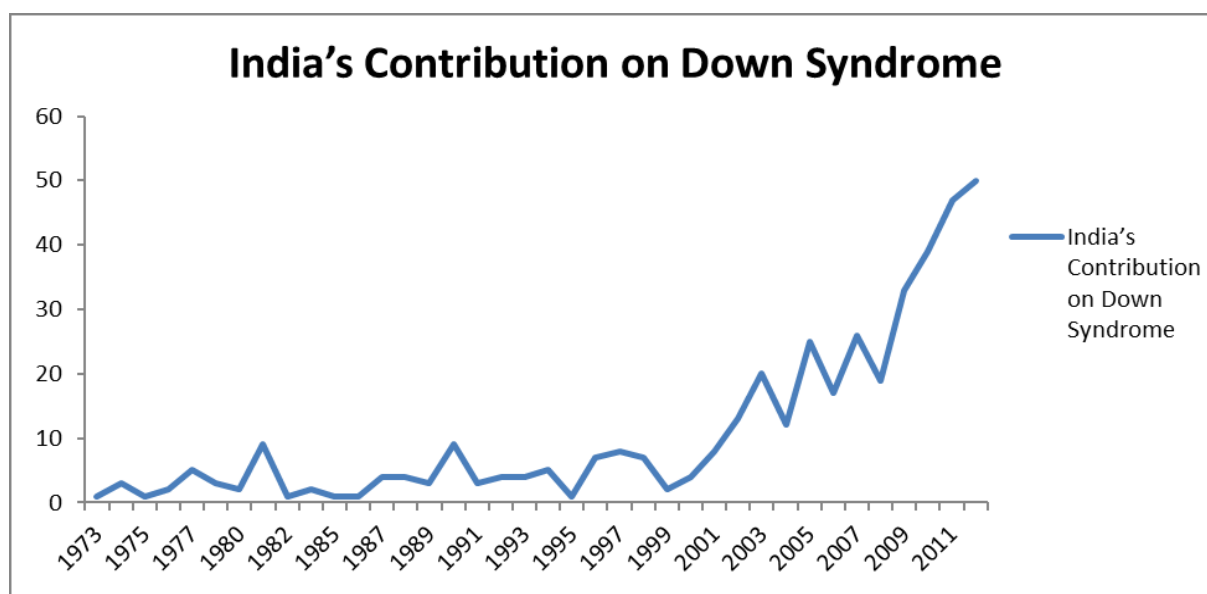
#### 4.2. India's Contribution on Down Syndrome

India's cumulative publication output in Down syndrome research is 405 papers from 1973-2012 and it holds the 12<sup>th</sup> rank in total world output. These overall publications appears in different document types consisting of 296 articles (73.09%), 37 letters (9.14%), 35 reviews (8.64%), 13 conference papers (3.21%), 5 editorials (1.23%), 5 notes (1.23%), 4 short surveys (0.99%), 2 articles in press (0.49%), 1 book chapter (0.25%) and 7 undefined papers (1.73%). The Indian output increased from 1 paper in 1973 to 50 papers in 2012. The cumulative papers increased from 27 in 1973-1982 to 31 in 1983-1992 and from 59 in 1993-2002 to 288 in 2003-2012. Thus, the maximum Indian papers on Down syndrome (71.11%) were published during last 10 years from 2003-2012. As far as citations are concerned, these 405 papers receive a total of 1676 citations with an average of 4.14 citations per paper. The ACPP is highest for the papers published during 1998-2012 (11.06).

**Table 3.** India's publication output and citation impact on Down syndrome.

Time Period	TP	%age	TC	ACPP
1973-1977	12	2.96	44	3.67
1978-1982	15	3.70	46	3.07
1983-1987	8	1.98	44	5.50
1988-1992	23	5.68	87	3.78

1993-1997	25	6.17	149	5.96
1998-2002	34	8.40	376	11.06
2003-2007	100	24.69	636	6.36
2008-2012	188	46.42	294	1.56
<b>Total (1973-2012)</b>	<b>405</b>	<b>100.00</b>	<b>1676</b>	<b>4.14</b>
<i>TP= Total papers; TC= Total citations; ACP= Average citation per paper</i>				



**Figure 1.** Year-wise distribution of India's contributions on Down syndrome.

#### 4.3. Down Syndrome Research Output in Context of Different Subjects

As reflected in Scopus database classification (based on journal subject), India's publication output on Down syndrome research during 1973-2012 has been published in context of 6 subjects with highest publications output coming from medicine (342 papers, 84.44% share), followed by Biochemistry, Genetics and Molecular Biology (109 papers, 26.91% share), Neuroscience (15 papers, 3.7% share), Pharmacology, Toxicology and Pharmaceutics (14 papers, 3.46% share), Dentistry (10 papers, 2.47% share) and Health Professions (5 papers, 1.23% share) (see Table 4).

The citation impact of each subject category was also analyzed and it was found that Neuroscience have highest citation impact (7.80 citations per paper), followed by Pharmacology, Toxicology and Pharmaceutics (7.50 citations per paper), Biochemistry, Genetics and Molecular Biology (4.99 citations per paper), Dentistry (4.7 citations per paper), Health Professions (4.6 citations per paper) and Medicine (3.86 citations per paper). The subject category Medicine has the highest number of papers as well as citations but the average citation per paper is low.

**Table 4.** Subject-wise breakup of Indian publications on Down syndrome.

Subject Area	TP	TC	ACPP
Medicine	342	1321	3.86
Biochemistry, Genetics and Molecular Biology	109	544	4.99
Neuroscience	15	117	7.80
Pharmacology, Toxicology and Pharmaceutics	14	105	7.50
Dentistry	10	47	4.70
Health Professions	5	23	4.60
<b>Total</b>	<b>405</b>	<b>1676</b>	<b>4.14</b>
<i>TP= Total papers; TC= Total citations; ACPP= Average citation per paper</i>			
<i>Note: The total of these five subjects exceeds the actual total due to overlapping of literature under different subject fields</i>			

#### 4.4. Top Contributing Institutions of India on Down Syndrome

The top 16 most productive Indian institutions in Down syndrome research publishing 5 or more papers are given in Table 5 along with their citation impact and h-index value. These 16 institutions accounts for 44.44% share (180 papers) of the total output of India with an average of 11.25 papers per institution. Only four institutions have registered higher productivity than this average. These four institutions are All India Institute of Medical Sciences (46 papers), Postgraduate Institute of Medical Education and Research (14 papers), Jawaharlal Institute of Postgraduate Medical Education and Research (12 papers) and University of Madras (12 papers). Apart from these, other major contributing institutions are St. John's Medical College, Institute of Immunohaematology (11 papers each), Manovikas Biomedical Research and Diagnostic Centre (10 papers), Sanjay Gandhi Postgraduate Institute of Medical Sciences (9 papers), King Edward Memorial Hospital, Manipal University Karnataka and Gujarat University (8 papers each). The average citation per paper for these top 16 institutions is 4.51. Seven institutions have registered more ACPP than this average. These includes Sir Ganga Ram Hospital (14.43 citations per paper), Osmania University (9.57 citations per paper), West Bengal University of Technology (7.4 citations per paper), King Edward Memorial Hospital (7.38 citations per paper), Postgraduate Institute of Medical Education and Research (6.5 citations per paper), Banaras Hindu University Institute of Medical Sciences (6.17 citations per paper) and Gujarat University (4.88 citations per paper). The highest h-index is registered by All India Institute of Medical Sciences (7), followed by Osmania University (5), Postgraduate Institute of Medical Education and Research, University of Madras, Institute of Immunohaematology, Gujarat University and West Bengal University of Technology (4 each).

**Table 5.** Productivity and citation impact of top Indian institutions.

S. No.	Name of the Institution	TP	TC	ACPP	h-index
1	All India Institute of Medical Sciences	46	186	4.04	7
2	Postgraduate Institute of Medical Education and Research	14	91	6.50	4
3	Jawaharlal Institute of Postgraduate Medical Education and Research	12	20	1.67	3

4	University of Madras	12	37	3.08	4
5	St. John's Medical College	11	14	1.27	3
6	Institute of Immunohaematology, Mumbai	11	45	4.09	4
7	Manovikas Biomedical Research and Diagnostic Centre	10	15	1.50	2
8	Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow	9	40	4.44	3
9	King Edward Memorial Hospital, Mumbai	8	59	7.38	3
10	Manipal University Karnataka	8	16	2.00	2
11	Gujarat University	8	39	4.88	4
12	Osmania University	7	67	9.57	5
13	Sir Ganga Ram Hospital	7	101	14.43	3
14	Manovikas Kendra Rehabilitation and Research Institute for the Handicapped	6	7	1.17	2
15	Banaras Hindu University Institute of Medical Sciences	6	37	6.17	2
16	West Bengal University of Technology	5	37	7.40	4
	<b>Total</b>	<b>180</b>	<b>811</b>	<b>4.51</b>	
	<b>Share of top 16 institutions in India's total output</b>	<b>44.44%</b>			
<i>TP= Total papers; TC= Total citations; ACP= Average citation per paper</i>					

#### 4.5. Most Productive Indian Authors on Down Syndrome

There are 14 authors who have published 6 or more papers on Down syndrome during 1973-2012. The publication profile of these 14 authors along with their research output, citations and h-index value are given in Table 6. Among these top authors 4 are affiliated to Manovikas Biomedical Research and Diagnostic Centre, Kolkata; 3 are affiliated to JIPMER, Pondicherry; and one each are affiliated to AIIMS, New Delhi; Sir Ganga Ram Hospital, New Delhi; International Medical School, Bangalore; St. John's Medical College, Bangalore; King Edward Memorial Hospital, Mumbai; Manipal Academy of Higher Education, Manipal, Karnataka; and University of Madras, Chennai.

These 14 authors have contributed a total of 120 papers with an average of 8.57 papers per author and accounts for 29.63% share of the total publication output of India during 1973-2012. Six authors have published higher number of papers than this group average (8.57). These authors are I.C. Verma of Sir Ganga Ram Hospital, New Delhi (15 papers), M. Kabra of AIIMS, New Delhi (11 papers), B. Vishnu Bhat and K. Ramachandra Rao of JIPMER, Pondicherry, S. Sinha of Manovikas Biomedical Research and Diagnostic Centre, Kolkata and S. Rajangam of International Medical School, Bangalore (10 papers each). Considering the citation impact of these top authors, it was revealed that these top 14 authors have received a total of 441 citations with an average of 3.68 citations per paper. Only three authors have registered higher impact than this average. These are: M.S. Tullu (12.67 citations per paper), I.C. Verma (11.13 citations per paper) and M. Kabra (6.64 citations per paper). When the performance of these authors was measured on the basis of h-index it was found that four authors have achieved higher h-index than the group average of 2.57. These authors are I.C. Verma (h-index 6), M. Kabra (h-index 5), N. Chandra and M.S. Tullu (h-index 3 each).

**Table 6.** Productivity and citation impact of most prolific authors.

S. No.	Name of Author	Affiliating Institution	TP	TC	ACPP	h-index
1	I.C. Verma	Sir Ganga Ram Hospital, New Delhi	15	167	11.13	6
2	M. Kabra	AIIMS, New Delhi	11	73	6.64	5
3	B. Vishnu Bhat	JIPMER, Pondicherry	10	8	0.80	2
4	S. Sinha	Manovikas Biomedical Research and Diagnostic Centre, Kolkata	10	13	1.30	2
5	K. Ramachandra Rao	JIPMER, Pondicherry	10	8	0.80	2
6	S. Rajangam	International Medical School, Bangalore	10	11	1.10	2
7	K. Mukhopadhyay	Manovikas Biomedical Research and Diagnostic Centre, Kolkata	8	14	1.75	2
8	S. Dutta	Manovikas Biomedical Research and Diagnostic Centre, Kolkata	8	15	1.88	2
9	P. Chand	JIPMER, Pondicherry	7	3	0.43	1
10	I.M. Thomas	St. John's Medical College, Bangalore	7	9	1.29	2
11	P.M. Gopinath	Manipal Academy of Higher Education, Manipal, Karnataka	6	14	2.33	2
12	N. Chandra	University of Madras, Chennai	6	18	3.00	3
13	K. Nandagopal	Manovikas Biomedical Research and Diagnostic Centre, Kolkata	6	12	2.00	2
14	M.S. Tullu	King Edward Memorial Hospital, Mumbai	6	76	12.67	3

*TP= Total papers; TC= Total citations; ACPP= Average citation per paper*

#### 4.6. Media of Research Communication

Out of the total 405 Indian publications on Down syndrome 398 appeared in journals, 6 in conference proceedings and 1 in book. The frequency distribution of publications in various source journals/ conferences is shown in Table 7. It was found that there are 118 journals in which only one paper is published and 28 journals in which two papers each were published. The Indian authors published 42 papers in just one journal titled *Indian Journal of Pediatrics*.

**Table 7.** Frequency distribution of papers in various source journals/ conferences.

Number of Papers	No. of Source Journals/ Conferences
1	118
2	28
3	13
4	8
5	3
6	1
7	1
9	1



11	1
12	1
15	1
17	1
26	1
42	1

#### 4.7. Top Source Journals for Publishing Indian Research on Down Syndrome

The 20 most productive journals publishing Indian research on Down syndrome are shown in Table 8. These 20 journals accounts for 47.41% (192 papers) of the total Indian output during 1973-2012. The journal most preferred by Indian authors for publishing research on Down syndrome is *Indian Journal of Pediatrics* in which 42 papers were published during 1973-2012. This is followed by *Indian Pediatrics* (26 papers), *Indian Journal of Human Genetics* (17 papers), *International Journal of Human Genetics* (15 papers), *Indian Journal of Medical Research* (12 papers) and *Current Pediatric Research* (11 papers).

**Table 8.** Top journals publishing Indian papers on Down syndrome.

S. No.	Name of Journal	No. of Papers
1	Indian Journal of Pediatrics	42
2	Indian Pediatrics	26
3	Indian Journal of Human Genetics	17
4	International Journal of Human Genetics	15
5	Indian Journal of Medical Research	12
6	Current Pediatric Research	11
7	Perinatology	9
8	Genetic Testing and Molecular Biomarkers	7
9	Indian Journal of Hematology and Blood Transfusion	6
10	Annales De Genetique	5
11	Journal of the Indian Medical Association	5
12	Pediatric Hematology and Oncology	5
13	Indian Journal of Radiology and Imaging	4
14	Indian Journal of Practical Pediatrics	4
15	Indian Journal of Pathology and Microbiology	4
16	Biosciences Biotechnology Research Asia	4
17	Indian Journal of Medical Sciences	4
18	Journal of Association of Physicians of India	4
19	Journal of Medical Genetics	4
20	Journal of Pediatric Endocrinology and Metabolism	4

#### 4.8. International Collaboration

The Indian authors have collaborated with authors of other countries in 38 papers which accounts for 9.38% of the total papers on Down syndrome. The maximum international collaborations have been in the year 2011 (7 international collaborative papers), which is

followed by 6 collaborative papers in 2010 and 5 in 2012. Thus, almost half of the international collaborative papers appeared in the last three years. S. Ghosh and S.K. Dey have the highest number of international collaborative papers (4 papers each), followed by N. Chandra (3 papers) and A. Chatterjee, P.M. Gopinath, S. Dutta, P. Ghosh, P. Bhaumik, S. Sinha, I.C. Verma and R. Yashwant (2 papers each). In Down syndrome research India has collaborated with 16 different countries (see table 9). The maximum international collaborative papers are with United States (13 papers) and United Kingdom (10 papers). The top international institutions collaborating with India includes University of Pittsburgh Graduate School of Public Health (4 papers), The University of North Carolina at Chapel Hill (2 papers) and Dongguk University, Gyeongju (2 papers).

**Table 9.** Major countries collaborating with India in Down syndrome research.

S. No.	Name of the Country	Total Collaborative Papers	%age
1	United States	13	34.21
2	United Kingdom	10	26.32
3	Italy	3	7.89
4	South Korea	3	7.89
5	Canada	2	5.26
6	China	2	5.26
7	Germany	2	5.26
8	Nepal	2	5.26
9	Spain	2	5.26
10	Switzerland	2	5.26
11	Czech Republic	1	2.63
12	Japan	1	2.63
13	Kuwait	1	2.63
14	Netherlands	1	2.63
15	New Zealand	1	2.63
16	Singapore	1	2.63
<b>Total</b>		<b>38</b>	<b>100.00</b>
<i>Note: More than one country have collaborated in many papers. So, the sum of all countries exceeds the total international collaborative papers (38).</i>			

#### 4.9. Citation Profile and Highly Cited Papers

The citation profile of Indian publications on Down syndrome during 1973-2012 is shown in Table 10. Out of the total 405 papers 249 papers (61.48%) have received atleast one citation during 1973-2012 with a cumulative total of 1676 citations. 156 papers (38.52%) did not receive any citation at all. Out of the 249 papers receiving citations, 1 paper received more than 100 citations, 3 papers received citations between 51 to 100, 3 papers received citations between 41 to 50, 4 papers received 31 to 40 citations, 7 papers received 21 to 30 citations, 19 papers received 11 to 20 citations and 212 papers received upto 10 citations.

The top 18 highly cited papers which have been cited more than 20 times are shown in Table 11. Out of these, only 2 are single authored papers and the rest have two or more

authors. These 18 papers have been published in 17 different national and international journals. Out of these 18 papers, 1 paper each was published in 1976, 1987, 1993, 1998, 2003, 2005, 2006, 2010 and 2 papers each in 2000, 2001, 2002, 2004 and 2007. The highly cited 18 papers received a total of 727 citations (43.37% of total citations) with an average of 40.38 citations per paper. Seven papers have registered higher citations than this average. The top cited paper (122 citations) is “*Superoxide dismutase - Applications and relevance to human diseases*” by R. Noor, S. Mittal & J. Iqbal which appeared *Medical Science Monitor* in 2002.

**Table 10.** Citation profile of papers.

No. of Citations	No. of Papers	Percentage
>100	1	0.25
51-100	3	0.74
41-50	3	0.74
31-40	4	0.99
21-30	7	1.73
11-20	19	4.69
1-10	212	52.35
Zero citations	156	38.52
<b>Total</b>	<b>405</b>	<b>100.00</b>

**Table 11.** Highly cited papers in Down syndrome research during 1973-2012.

S. No.	Document Title	Author/s	Source Title	No. of Citations
1	Superoxide dismutase - Applications and relevance to human diseases	R. Noor, S. Mittal & J. Iqbal	Medical Science Monitor, 2002, 8 (9)	122
2	Ribosome inactivating proteins and apoptosis	S. Narayanan <i>et al</i>	FEBS Letters, 2005, 579 (6)	63
3	Selenium - Its biological perspectives	R.S. Bedwal <i>et al</i>	Medical Hypotheses, 1993, 41 (2)	58
4	Mechanisms of disease: DNA repair defects and neurological disease	K.S. Rao	Nature Clinical Practice Neurology, 2007, 3 (3)	52
5	Epidemiology of childhood psoriasis: A study of 419 patients from northern India	B. Kumar <i>et al</i>	International Journal of Dermatology, 2004, 43 (9)	46
6	Burden of genetic disorders in India	I.C. Verma	Indian Journal of Pediatrics, 2000, 67 (12)	45
7	Down syndrome: Clinical profile from India	M.P. Kava <i>et al</i>	Archives of Medical Research, 2004, 35 (1)	44
8	Premature ovarian failure	D. Goswami & G.S. Conway	Hormone Research, 2007, 68 (4)	36
9	Inbreeding and the incidence of childhood genetic disorders in Karnataka, South India	A.R.R. Devi, N.A. Rao & A.H. Bittles	Journal of Medical Genetics, 1987, 24 (6)	33
10	Down's syndrome and related abnormalities in an area of high	N. Kochupillai <i>et al</i>	Nature, 1976, 262 (5563)	32

	background radiation in coastal Kerala			
11	MTHFR C677T and A1298C polymorphisms are risk factors for Down's syndrome in Indian mothers	A.K. Rai <i>et al</i>	Journal of Human Genetics, 2006, 51 (4)	31
12	The burden of genetic disorders in India and a framework for community control	I.C. Verma & S. Bijarnia	Community Genetics, 2002, 5 (3)	30
13	Safety of oseltamivir in pregnancy: A review of preclinical and clinical data	B. Donner, V. Niranjana & G. Hoffmann	Drug Safety, 2010, 33 (8)	26
14	Gene polymorphism and folate metabolism: A maternal risk factor for Down syndrome	J.J. Sheth & F.J. Sheth	Indian Pediatrics, 2003, 40 (2)	24
15	Parental age and the origin of extra chromosome 21 in Down syndrome	A. Jyothy <i>et al</i>	Journal of Human Genetics, 2001, 46 (6)	22
16	Morgagni hernia with down syndrome: A rare association - Case report and review of literature	R.C. Parmar <i>et al</i>	Journal of Postgraduate Medicine, 2001, 47 (3)	21
17	Cytogenetic studies of 1001 Down syndrome cases from Andhra Pradesh, India	A. Jyothy <i>et al</i> P.P.	Indian Journal of Medical Research, 2000, 111	21
18	Trisomy 21 and accelerated aging: DNA-repair parameters in peripheral lymphocytes of Down's syndrome patients	N.S. Raji & K.S. Rao	Mechanisms of Ageing and Development, 1998, 100 (1)	21

## 5. SUMMARY AND CONCLUSIONS

The analysis of literature on Down syndrome (during 1973-2012) obtained through Scopus database reveals that the United States account for maximum literature (28.34%) on Down syndrome followed by United Kingdom (with a share of 12.81%). India, with a global share of 1.41% (405 publications), stands at 12<sup>th</sup> place in publication output on Down syndrome indicating that the contributions by Indian authors in this area are low. The maximum Indian publications (71.11%) appeared during last 10 years from 2003-2012. The overall citation impact is also not very high with 4.14 citations per paper from 1973-2012.

All India Institute of Medical Sciences, New Delhi is the most productive institution with 46 publications during 1973-2012. The most prolific Indian author is I.C. Verma of Sir Ganga Ram Hospital, New Delhi who published 15 papers and received 167 citations (11.13 citations per paper). The *Indian Journal of Pediatrics* was most preferred by Indian authors for publishing their research. International collaboration was seen in 38 papers (9.38%) and the maximum collaboration (13 papers) was with United States. About 62% of the total Indian papers have been cited by others and there are 18 papers which have been cited more than 20 times.

The bibliometric analysis of Down syndrome indicates that India's publication output is very small as compared to other countries. The Indian scientists need to focus more on this

area of research and also increase international collaborations for research. Efforts can also be increased towards obtaining more research funding and creating awareness about the disease.

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