Mapping of India’s Contribution on “Down Syndrome” During 40 Years From 1973-2012

Anil Kumar Siwach*
Department of Library and Information Science, Maharshi Dayanand University, Rohtak, Haryana (India)
*E-mail address: aks.mdu@gmail.com

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                                   |

This paper is an open access paper published under the terms and conditions of the Creative Commons Attribution license (CC BY) (https://creativecommons.org/licenses/by/4.0)
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%, 5th rank), Germany (3.73%, 6th rank), Canada (3.53%, 7th rank), Spain (3.07%, 8th rank), Netherlands (2.66%, 9th rank) and Australia (2.47%, 10th rank). Israel, India, Switzerland, Brazil and Sweden ranks 11th to 15th respectively (with publication share from 1.20% to 1.73%). Thus, India holds the 12th 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 1st and 2nd 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         | 1973-1982 | 1983-1992 | 1993-2002 | 2003-2012 | 1973-2012 | TC     | ACPP   | h-index |
|------|----------------|-----------|-----------|-----------|-----------|-----------|--------|--------|---------|
| 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      |
|      | World          | 3815      | 4895      | 8106      | 11922     | 28738     |        |        |         |

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 12th 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. A 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 |
## 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 |
| **Total**                                              | 405 | 1676  | 4.14 |

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

Note: The total of these five subjects exceeds the actual total due to overlapping of literature under different subject fields.

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       |
4.5. 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 proceeds 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                                   |
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  |
| Total  |                       | 38                         | 100.00|

Note: More than one country have collaborated in many papers. So, the sum of all countries exceeds the total international collaborative papers (38).

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      |
| Total            | 405           | 100.00     |

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 et al                | FEBS Letters, 2005, 579 (6)       | 63               |
| 3      | Selenium - Its biological perspectives            | R.S. Bedwal et al                 | 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 et al                  | 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 et al                   | 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 et al | Nature, 1976, 262 (5563)          | 32               |
### 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 12th 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.

References

[1] Brook J.D., Gosden R.G., Chandley A.C. Maternal ageing and aneuploid embryos—evidence from the mouse that biological and not chronological age is the important influence. Human Genetics 66 (1) (1984) 41-45.

[2] Deccan Herald News Service. Down syndrome still fatal in India. Deccan Herald, April 19, 2013.

[3] Ghosh S., Feingold E., Chakraborty S., Dey S.K. Telomere length is associated with types of chromosome 21 nondisjunction: a new insight into the maternal age effect on Down Syndrome birth. Human Genetics, 127(4) (2010) 403-409.

[4] Jones, K. L. Recognizable patterns of human malformation. Philadelphia: Elsevier (2006).

[5] NDSS - National Down Syndrome Society. What Is Down Syndrome? Retrieved from www.ndss.org/Down-Syndrome/What-Is-Down-Syndrome/

[6] Penrose, L.S. The relative effect of paternal and maternal age in Mongolism. Journal of Genetics, 27(2) (1933) 219-224.

[7] Penrose, L.S. The relative etiological importance of birth order and maternal age in Mongolism. Proceedings of the Royal Society B Biological Sciences, 115 (1934) 431-450.

[8] Powell-Hamilton, Nina N. Down Syndrome (Trisomy 21; Trisomy G). In Merck Manual Home Health Handbook for Patients and Caregivers. Retrieved from www.merckmanuals.com/home/childrens_health_issues/chronosomal_and_genetic_abnormalities/down_syndrome_trisomy_21_trisomy_g.html?qt=&sc=&alt=

[9] Verma, I.C. Personal communication. As cited in Jayalakshamma et al. Cytogenetic Analysis in Down Syndrome. International Journal of Human Genetics, 10(1-3) (2010) 95-99.

[10] WHO Genomic Resource Centre. Gene and human disease. Retrieved from http://www.who.int/genomics/public/geneticdiseases/en/index1.html#

(Received 01 February 2015; accepted 10 February 2015)