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Abstract

The present study was conducted to explore various aspects of Cornelia de Lange Syndrome (CdLS) research publications including *annual scientific productivity, top contributing authors and their impact, top contributing countries and organizations, most relevant sources of publication, highly cited documents, and most frequently used words*. Bibliometric methods were used to investigate these aspects of CdLS research publications. Results of the study disclosed that the annual scientific productivity of CdLS literature is increasing gradually with the passage of time. A. Selicorni contributed the highest number of publications (45) to CdLS literature while I. D. Krantz had the highest impact in the CdLS research field. The United States was the highest contributing country with 962 occurrences. The University of Birmingham was the highest contributing organizations with 144 documents. *American Journal of Medical Genetics* was the most relevant source of publication for Cornelia de Lange Syndrome research literature which published 63 documents. The most cited paper on CdLS was contributed by Krantz et al. in *Nature Genetics*, volume 36, number 6, 2004, which received the highest number of citations (491) at a rate of 28.88 citations per year. "de Lange Syndrome" was the most frequently used word having

1,332 occurrences followed by a human (857 occurrences) and female (751 occurrences). It is recommended that researchers should consider using multiple publications crediting methods while working on author productivity rankings. It is further recommended to explore different aspects of CdLS research literature that have not been covered in this study with different data sets.

Key Words

Cornelia de Lange Syndrome, CdLS, Brachmann de Lange Syndrome, Genetic disorder, Bibliometrics, Productivity analysis.

Introduction

Cornelia de Lange Syndrome (CdLS) is an infrequent hereditary condition. Origin of its name is linked to a Dutch pediatrician Cornelia Catharina de Lange who defined it in a research paper published in 1933. Though Dutch anatomist Vrolik and German physician Brachmann have written about this syndrome in 1849 and 1916 respectively, it was de Lange who first described it in detail (Boyle et al., 2015). CdLS is also known as Brachmann de Lange Syndrome or Bushy Syndrome (Kline et al., 2007). Normally it is not a contagious ailment. It is believed that most cases are due to random genetic mutations. Cornelia de Lange Syndrome (CdLS) usually occurs owing to genetic mutation in one of five major progressive genes (Boyle et al., 2015; Grados et al., 2017; Kuzniacka et al., 2013; Sarogni et al., 2019). Approximately 80% of cases are due to the Nipped-B-like protein gene mutation (Boyle et al., 2015). The exact manifestation of CdLS is uncertain but it is estimated that there is one case of CdLS in 10,000 to 50,000 live births (Boyle et al., 2015; Dowsett et al., 2019; Kline et al., 2007; Opitz & Reynolds, 1985; Sarogni et al., 2019).

It is a rare disorder that affects multiple organs of newly born babies. Children affected with this syndrome have physical, cognitive, and medical challenges, from trivial to severe, throughout their life. CdLS patients have typical distinctive facial features such as curved eyebrows meeting at the midline, long eyelashes, small ears, thick hair, short and widely spaced teeth, narrow nose with anteverted nostrils, thin lips with downward mouth, low voice, weak eyesight, hernia and seizures. This syndrome is related to development and mental disorders with low IQ level, low birth weight, developmental delay or growth retardation, skeletal defect, upper and lower limb abnormalities, Gastroesophageal Reflux Disease (GERD), and hearing loss. Complications like respiratory illness, cardiovascular or heart defects, endocrine and gastrointestinal abnormalities, sleeping disorder, and feeding problem may occur (Kline et al., 2007; Mulder et al., 2017).

The family is shocked if their child is born with CdLS without prenatal diagnosis. Kinjo et al. (2019) suggested that early prenatal diagnosis of CdLS might be improved significantly thorough ultrasound examination of the limb and fetal growth restriction (FGR). However, Kanellopoulos et al. (2011) informed that prenatal diagnosis of the syndrome is rare as 68 percent of cases of significant abnormalities could not be detected through a regular prenatal ultrasound. The individuals affected with CdLS face social anxiety and are normally quiet (Crawford et al., 2020). They expect and require more love, affection, care, and attention. Mood disorder and behavioral changes are often noticed in the patient. Regular advice and follow up is required with pediatrician and different specialists.

Bibliometrics is the field of research in which written communications are studied and examined to evaluate the scientific output. The term bibliometry was coined by Dr. S. R. Ranganathan in 1948 (Raja & Murugan, 2015). However, the word bibliometrics was introduced

by Pritchard in 1969 (Naseer & Mahmood, 2009; Raja & Murugan, 2015). Bibliometric analysis provides reliable knowledge about the quantity and nature of scientific work (Wang et al., 2016). Bibliometrics is a valued instrument to describe and promote scientific productivity (Tallolli & Mulla, 2016). Bibliometric studies are used to examine various types of information i.e. productivity in the area of study, most prolific authors and highly cited papers, authorship collaboration (global or local), top contributing organizations, top contributing countries, single and multi-authored communications for articles, books or any other format etc.

Bibliometrics plays an important role in the assessment of research community for promotion and other incentives. "Bibliometrics helps to explore, organize and analyze large amounts of historical data helping researchers to identify "hidden patterns" that may help researchers in the decision-making process" (Daim et al., 2006), p. 983). Web of Science (WOS) and Scopus are the main sources for the bibliometric studies. Web of Science is a subscription-based bibliographic and citation database maintained by Clarivate Analytics. Scopus is Elsevier's bibliographic and citation database. It also provides subscription-based access. Scholars can obtain data from these databases and conduct the research study. Data for bibliometric research studies can also be obtained from other free sources like PubMed, Google Scholar, and SCImago Journal Rank (SJR).

As mentioned above, children affected with CdLS have physical, cognitive, and medical challenges and their families need support and encouragement (Doreen & Annette, 2016). Counseling is suggested for the affected individuals and their families (Kline et al., 2007). In such cases, information about CdLS is very important for all stakeholders (doctors, families, and patients). Information produced by a bibliometric study in this area will be useful addition to body of knowledge because comprehensive literature searching shows that no bibliometric study has

been conducted on CdLS so far. Hence, this study was envisioned and conducted to achieve the following objectives.

Objectives

1. To explore annual scientific productivity of CdLS literature

2. To find the top contributing authors in CdLS research and their impact in the field

3. To examine the top contributing countries and organizations in CdLS research

4. To discover the most relevant sources of publication for CdLS research

5. To explore the most cited documents in CdLS research

6. To find the most frequently used words in CdLS research

Literature Review

As mentioned above, no bibliometric study has been conducted on CdLS so far. Therefore, bibliometric studies on related areas of medical sciences have been reviewed here. A recent study on Carpal Tunnel Syndrome (CTS) found that United States was the most productive country and Luca Podua was the most productive author in CTS research (Ram, 2019). The results of the study revealed that papers published in 1998 received highest number of citations during the period under study (1983-2017). Annual increase in documents published on CTS was 9.86%. The article written by Lawrence et al. received the highest number of citations (1,989). Highly productive journal on CTS was *Journal of Hand Surgery - American Volume* which published 895 articles followed by *Muscle and Nerve* (302 articles) and *Journal of Hand Surgery - European Volume* (166 articles).

Bibliometric analysis of 5,128 documents about Coronavirus by Danesh and Ghavidel (2020) revealed that the year 2005 was the most productive year with highest scientific output on

Coronavirus (6.8%) followed by 2004 (6.78%) and 2006 (5.92%). These publications received 165,451 citations in the last 50 years. Documents published in 2019 received the highest number of citations (11,385). L. Enjuanes was most prolific author, United States was highest contributing country, and University of Hong Kong was top contributing institution in the Coronavirus research in the past 50 years. Investigation of the research output on regulatory T-cell (Treg) from 2000 to 2015 found that the productivity of scientific literature on Treg increased with the time (Zongyi et al., 2016). The *Journal of Immunology* was the most preferred source of publication for Treg research followed by *PLOS One* and *Blood*. The United States contributed the highest number of publications on Treg research, followed by China, Germany, United Kingdom, and Japan. The highest contributing author was Y. Wang with 186 publications. The research paper by S. Sakaguchi (2008) attracted highest number of citations. The Harvard University was highly contributing organization followed by University of Pittsburgh and Oxford University.

A latest study on trends of Coronavirus (CoV) research showed that the United States was the highest contributing country for coronavirus research with 31.67% publications and highly contributing organization was the University of Hong Kong (Ram, 2020). The findings of study disclosed that *Journal of Virology* published the highest number of documents on coronavirus research during the period under study (1970-2019). "A New Coronavirus Associated with Severe Acute Respiratory Syndrome" by Ksiazek et al. published in 2003, accumulated the highest number of citations. A Spanish researcher L. Enjuanes emerged as the most productive author on the coronavirus research. An analysis of 66 research papers from Arab countries related to health communication published since 2004 noted that first research paper originated from Lebanon (Mheidly & Fares, 2020). Contribution of Arab countries in health communication research was very low as it was only 0.03% of the total biomedical research from Arab world and only 1% of

the global health communication research. Contribution of different Arab countries to health communication literature ranged from zero to 12. Lebanon contributed the highest number of publications while ten Arab countries did not contribute any publication.

Sweileh (2017) conducted the bibliometric analysis of publications on Stevens Johnson Syndrome (SJS) and Toxic Epidermal Necrolysis (TEN). The h-index for 3,856 articles analyzed was 95. Publication grew at the highest rates during 1966-75 and 2006-15. The United States was leading in SJS and TEN research with 640 (16.57%) publications. Comparatively low level of international collaboration was seen between active countries. The highest collaboration was found for Swiss researchers (32.5%) and lowest collaboration was for Spanish researchers (1.47%). Majority of SJS and TEN research was published in journals related to dermatology, especially in *Archives of Dermatology*. Liao et al. (2016) revealed that Chinese authors contributed most of the research related to Blood Stasis Syndrome (BSS). They found that on average each article received 4.65 citations. The h-index of 27.74% of the papers was higher than two while h-index of only 3 articles was higher than 10 during the study period (1989-2015).

An analysis of research on Severe Acute Respiratory Syndrome Coronavirus (SARS-CoV), Middle East Respiratory Syndrome Coronavirus (MERS-CoV) and 2019-novel CoV, published from January 1951 to January 2020 Bonilla-Aldana et al. (2020), found that United States contributed highest number of publications to Coronavirus research and contributed nearly one third of the total research output followed by China which contributed 22% publications indexed in Science Citation Index (SCI). The contributions of Hong Kong and South Korea, affected by SARS-CoV and MERS-CoV, were notable in this area as both were among top ten countries in SCI. However, Middle East countries affected by MERS-CoV, like Saudi Arabia, had low contribution in Coronavirus research during the period under study.

Wang et al. (2016) reported that Middle East Respiratory Syndrome (MERS), a communicable disease, first appeared in April 2012 in Saudi Arabia. Later it also appeared in other Asian countries, including South Korea and China. According to the results of their study the United States was top contributing country in MERS research followed by China and Saudi Arabia. United States was providing most of the funding for MERS research. China was paying special attention to emerging infectious diseases. Most of the MERS literature was contributed by universities. *Journal of Virology* published highest number of research articles on MERS (44, 9.9%). Z. A. Memish was highly contributing author on MERS with 21 publications.

Zyoud (2016) in their study on trends of MERS Coronavirus (MERS-CoV) analyzed 883 publication and found that the United States was the leading contributing country in MERS-CoV research with 319 papers published during four years of the study (2012-2015) followed by Kingdom of Saudi Arabia (113 papers). The publications analyzed received 8,015 citations with an average of 9.01 citations per publication. The h-index of publications related to MERS-CoV was 48. The United States had the maximum h-index of 32 followed by Kingdom of Saudi Arabia (26) and United Kingdom (22). The Netherlands produced the largest proportion of foreign research collaborative publications (72.7% of its output) followed by the United Kingdom (71% of its output) and Germany (69.1% of its output). *Journal of Virology* published the highest number of MERS-CoV items (46) followed by *Emerging Infectious Diseases* (43) and *Euro surveillance* (36). The papers published in the *New England Journal of Medicine,* Zaki et al. (2012) and Assiri et al. (2013), scored highest number of citations (645 and 256 respectively).

Analysis of Severe Acute Respiratory Syndrome (SARS) research publications revealed that the United States was highest contributing country with 30% of total publications followed by Hong Kong with 24% publications (Chiu et al., 2004). However, based on reprint address Hong

Kong was ranked on the top. Although Belgium, Mexico, Netherlands, Norway, Saudi Arabia, and Senegal contributed to the research on SARS, but they did not report any SARS case. Forty seven percent of the countries affected by SARS did not contribute any publication in Science Citation Index. The study disclosed that 48% of research publications were without reprint addresses. An analysis of 5,465 publications on mobile health (m-health) published during 2006-2016 showed that majority of the publications (4,638, 84.9%) were contributed by more than one authors (Sweileh et al., 2017). The United States emerged as the most productive country with 1,926 (35.2%) publications. VA Medical Center contributed the highest number of research publications while *Journal of Medical Internet Research* was most preferred source of publication on m-health.

Zyoud et al. (2015) analyzed 591 papers on Integrative and Complementary Medicine (ICM) research from 22 Arab countries, published in 22 ICM journals during 1980-2013. Results revealed that Morocco achieved an h-index of 27 while Jordan got an h-index of 21. Comoros, Qatar, and Syrian Arab Republic acquired the least h-index of one for each of them. Djibouti and Mauritania did not publish any research related to ICM. The publications analyzed received 9,466 citations with an average citation of 16 per paper. The first article from Arab countries was published in 1980 in Egypt by M. D. Sayed in *Journal of Ethnophramacology*. The study reported that 262 documents (44.3%) had collaborations with Arab and other foreign countries. Arab authors had most of the collaboration with European Countries (24.2%) followed by Asia-Pacific countries (9.8%). The results of the investigation discovered that research productivity of Arab countries in the area of ICM was very low in earliest two decades, but it increased significantly after 2011 and approximately 34.2% research publications were published in just three years (2011-2013).

A study based on 6,896 Scopus indexed Obstructive Sleep Apnea (OSA) research articles, published from 1991-2012, revealed that the highest number of articles (685) was published in 2012 (Huamani et al., 2015). It identified that 6,896 articles were published in 1,422 journals but 34.8% articles were published in only 15 journals while more than 100 articles each were published by 11 journals. Highest number of OSA articles were published in the *Chest journal* (311, 4.51%) and 94.5% of these articles were cited. Overall, five thousand three hundred and thirty-eight (5,338) articles were cited. The United States was the highest contributing country with 1,758 (23.8%) articles.

Research Methodology

The analysis was conducted using bibliometric methods with the help of Biblioshiny open source software and MS Excel to explore the annual scientific productivity, top contributing authors and their impact, top contributing countries and organizations, most relevant sources of publication, most cited documents, and most frequently used words in CdLS research. Multidisciplinary database SCOPUS was used to download the bibliographic records of research output on Cornelia de Lange Syndrome to ensure that maximum number of records are analyzed. Scopus database was selected for bibliographic data because it has wider coverage as compared to Web of Science and other databases (Mongeon & Paul-Hus, 2016; Powell & Peterson, 2017; Sweileh et al., 2017; Zyoud, 2016). Data were downloaded on June 4, 2020 using TITLE-ABS-KEY search option of SCOPUS with the following search query:

"Cornelia de Lange Syndrome" OR "CdLS" OR "Cornelia de Lange" OR "Brachmann

de Lange Syndrome" OR "Brachmann de Lange" OR "BdLS" OR "Bushy Syndrome"

As compared to other search queries, we found the highest number of records for download with "TITLE-ABS-KEY" search option using the Boolean operator "OR" without applying any filter for time frame, country, or language. Total number of records downloaded were 1,229. Then data were manually scrutinized to ensure that only relevant records are used for analysis. Out of 1,229 records, 415 records not relevant to Cornelia de Lange Syndrome and three duplicate records were excluded from the analysis. Furthermore, editorial (n = 1), erratum (n = 12) and note (n = 10) were also not included in the analysis. Thus, total number of records analyzed were 788. Bushy Syndrome is another name of Cornelia de Lange Syndrome, but no record was found for the keywords Bushy Syndrome. It was noted that the pioneering research items on Cornelia de Lange Syndrome by Vrolik, Brachmann, and de Lange mentioned in Boyle, Jespersgaard, Brondum-Nielsen, Bisgaard and Tumer (2015) were not indexed in Scopus. The present study was limited to the publications on "Cornelia de Lange Syndrome" indexed in Scopus database only while other databases were not used for bibliographic data due to their less coverage of publications. Therefore, results of this study should be considered keeping in view the limitations of the study.

Results and discussion

The analysis of 788 documents on Cornelia de Lange Syndrome revealed that these documents were published in 403 sources (Journals, books, etc.) over the period of 67 years from 1953 to 2020 (see Table 1). Majority of these documents were research articles (82.49%, 650) followed by letters (6.85%, 54) and reviews (5.46%, 43). These documents were contributed by 3,117 authors with a total of 4,300 author appearances. The analysis disclosed that authors writing on CdLS had collaboration index of 4.36 with 3.96 authors per document and they contributed only 11.55% (91) single authored documents.

Table 1: Main information about data

Description	Results
Time span	1953:2020
Sources (Journals, Books, etc.)	403
Documents	788
Average years from publication	20.5
Average citations per documents	14.13
Average citations per year per doc	1.085
References	10144
DOCUMENT TYPES	
Article	650
Book chapter	7
Conference paper	31
Letter	54
Review	43
Short survey	3
DOCUMENT CONTENTS	
Keywords Plus	3915
Author's Keywords	954
AUTHORS	
Authors	3117
Author Appearances	4300
Authors of single-authored documents	81
Authors of multi-authored documents	3036
AUTHORS COLLABORATION	
Single-authored documents	91
Documents per Author	0.253
Authors per Document	3.96
Co-Authors per Documents	5.46
Collaboration Index	4.36

Annual productivity analysis (Table 2) indicated that growth of research publications on Cornelia de Lange Syndrome was increasing progressively. Research productivity was very low in the beginning as Scopus indexed only four documents (0.51%) in first decade (1953-1962). However, they contributed 229 documents during 2013-2020 (29.06%). The research scholars contributed only one third of the literature during first 40 years but, due to increased publication

activity, they contributed more than half of the literature in last 18 years. The most productive period was 2013-2020 as 229 documents (29.06%) appeared during this period. Citation analysis disclosed that 202 documents published during 2003-2012 received highest citations per article (30.54). Results of this study indicated that research productivity of Cornelia de Lange Syndrome increased with the passage of time. These results are like the investigation carried out by Zongyi et al. (2016) who found that the productivity of scientific literature on regulatory T-cell increased with the time.

Table 2: Scientific productivity of CdLS research

Years	Articles (n=788)	Percentage	Cumulative Percentage	Citations	Citations per Article
1953-1962	4	0.51	0.51	5	1.25
1963-1972	78	9.90	10.41	348	4.46
1973-1982	78	9.90	20.30	321	4.12
1983-1992	75	9.52	29.82	728	9.71
1993-2002	122	15.48	45.30	1951	15.99
2003-2012	202	25.63	70.94	6169	30.54
2013-2020*	229	29.06	100.00	1613	7.04
Total	**788**	**100.00**	**100.00**	**11135**	**14.13**

* Data up to June 4, 2020

The author productivity analysis exhibited that 3,117 authors contributed to Cornelia de Lange Syndrome literature. Top 20 authors, who contributed to CdLS, have been presented in Table 3 in their rank order along with their productivity. Author productivity by full credit method has been presented in columns 1-3 while author productivity by fractional credit method has been presented in columns 4-6. The analysis revealed that A. Selicorni contributed highest number of publications (45) to CdLS literature followed by I. D. Krantz (44 documents) and C. Oliver (36 documents). However, C. Oliver emerged as the highest contributing author using fractionalized crediting method with 7.57 publications. In this case, I. D. Krantz remained on the second position

(5.52 documents) while A. Selicorni secured third position (5.16 documents). It demonstrated that author productivity varies with the change of crediting method. Data disclosed that author names for some publications included in the analysis were missing as "No author name available" ranked 16 in fractionalized crediting with two documents.

Table 3: Top 20 authors contributing to CdLS research

Rank	Authors (n = 3,117)	Articles	Rank-Fraction	Authors-Fraction (n = 3,117)	Articles Fractionalized
1	Selicorni A	45	1	Oliver C	7.57
2	Krantz I D	44	2	Krantz I D	5.52
3	Oliver C	36	3	Selicorni A	5.16
4	Deardorff M A	25	4	Moss J	4.16
4	Kline Ad	25	5	Sarimski K	4.00
6	Gervasini C	24	6	Kline A D	3.79
6	Moss J	24	7	Dorsett D	3.77
7	Cereda A	23	8	Beck B	3.64
8	Jackson L G	22	9	Opitz J M	3.62
9	Russo S	21	10	Ireland M	3.02
10	Larizza L	20	11	Deardorff M A	2.98
11	Kaur M	18	12	Jackson L G	2.95
12	Masciadri M	16	13	Cereda A	2.62
13	Kaiser F J	15	14	Gervasini C	2.11
14	Mariani M	14	15	Russo S	2.01
14	Wierzba J	14	16	No author name available	2.00
16	Musio A	13	16	Tsukahara M	2.00
16	Parenti I	13	17	Wierzba J	1.90
16	Pie J	13	18	Larizza L	1.86
16	Ramos F J	13	19	Arron K	1.78

Author impact analysis discovered that I. D. Krantz had highest impact in the CdLS research field (see Table 4). Krantz started publishing in the year 2001 and contributed 44 publications, which received 2,735 citations with an h-index of 24. According to author impact, L. G. Jackson, who started publishing in 1985, occupied the second position. Jackson contributed 22 publications but received 1,909 citations with an h-index of 21. Although A. Selicorni was the highest contributing author with 45 publications, but secured 6th position according to author impact. Selicorni started publishing in 1993 and received 1,156 citations with an h-index of 18. It

demonstrated that author impact depends on a mix of several variables including but not limited to the length of publishing career and number of publications.

Table 4: Authors with highest impact in CdLS research

Rank	Author Name	Number of Publications	Publication Starting Year	Total Citations	h-Index	g-Index	m-Index
1	Krantz I D	44	2001	2735	24	44	1.20
2	Jackson L G	22	1985	1909	21	22	0.58
3	Kaur M	18	2004	1739	13	18	0.77
4	Kline A D	25	1993	1294	14	25	0.50
5	Deardorff M A	25	2007	1221	17	25	1.21
6	Selicorni A	45	1993	1156	18	33	0.64
7	Musio A	13	2006	970	7	13	0.47
8	Oliver C	36	2001	797	16	28	0.80
9	Gervasini C	24	2006	742	14	24	0.93
10	Larizza L	20	2006	728	14	20	0.93
11	Russo S	21	2006	643	11	21	0.73
12	Ramos F J	13	2007	620	8	13	0.57
13	Kaiser F J	15	2012	542	8	15	0.89
14	Moss J	24	2005	435	11	20	0.69
15	Masciadri M	16	2007	277	10	16	0.71
16	Cereda A	23	2009	275	10	16	0.83
17	Pie J	13	2010	231	7	13	0.64
18	Wierzba J	14	2006	219	8	14	0.53
19	Parenti I	13	2013	168	7	12	0.88
20	Mariani M	14	2012	101	5	10	0.56

Country wise analysis revealed that 57 countries contributed 788 documents on CdLS (Table 5). The United States was highest contributing country with 962 occurrences. Publications having United States' affiliation received 4,333 citations at an average of 35.52 citations per article. Italy was on second position with 571 occurrences having 1,175 citations at an average of 23.98 citations per article. The United Kingdom was on third position having 389 occurrences with 1,806 citations at an average of 37.62 citations per article. Though occurrences for the United Kingdom were less in numbers than the United States and Italy but publications received higher number of citations than Italy and higher average article citations than both the United States and

Italy. Japan, Spain, Germany, China, France, Netherlands, and Canada were ranked 4 to 10 having more than 100 occurrences. China gained 35 citations with 127 occurrences at an average of 3.89 citations per article.

Table 5: Top 20 countries contributing to CdLS research

Rank	Country (n = 57)	Occurrences	Total Citations	Average Article Citations
1	USA	962	4333	35.52
2	Italy	571	1175	23.98
3	UK	389	1806	37.62
4	Japan	232	305	10.52
5	Spain	227	165	8.68
6	Germany	131	253	14.06
7	China	127	35	3.89
8	France	120	117	23.40
9	Netherlands	118	410	27.33
10	Canada	111	259	14.39
11	Poland	96	95	7.31
11	Turkey	96	37	2.64
13	India	81	41	2.56
14	South Korea	42	31	3.88
15	Belgium	41	44	6.29
16	Denmark	38	199	28.43
17	Sweden	36	38	19.00
18	Brazil	32	5	2.50
19	Israel	29	50	10.00
20	Australia	22	52	10.40
20	Mexico	22	11	2.75

The results of this investigation are like earlier researches, which found that the United States was most productive country for the research on Carpal Tunnel Syndrome Ram (2019), Coronavirus Bonilla-Aldana et al. (2020); (Danesh & Ghavidel, 2020; Ram, 2020), Middle East Respiratory Syndrome (Wang et al., 2016), MERS-CoV (Zyoud, 2016), m-health (Sweileh et al., 2017), regulatory T-cell (Zongyi et al., 2016), Severe Acute Respiratory Syndrome (Chiu et al., 2004), Stevens Johnson Syndrome and Toxic Epidermal Necrolysis (Sweileh, 2017), and Obstructive Sleep Apnea (Huamani et al., 2015).

Analysis for contributing organizations discovered that 839 organizations contributed to the CdLS literature. Ten top contributing organizations have been presented in the Figure 1 along with their input. The University of Birmingham from the United Kingdom was the highest contributing organization with 144 documents. The University of Milan from Italy was on second position for contributing 138 documents. Organizational affiliations of 133 documents were not recorded. Thus, organizational affiliation "Not Reported" was on the third place. Children's Hospital of Philadelphia and University of Pennsylvania from United States ranked fourth and fifth
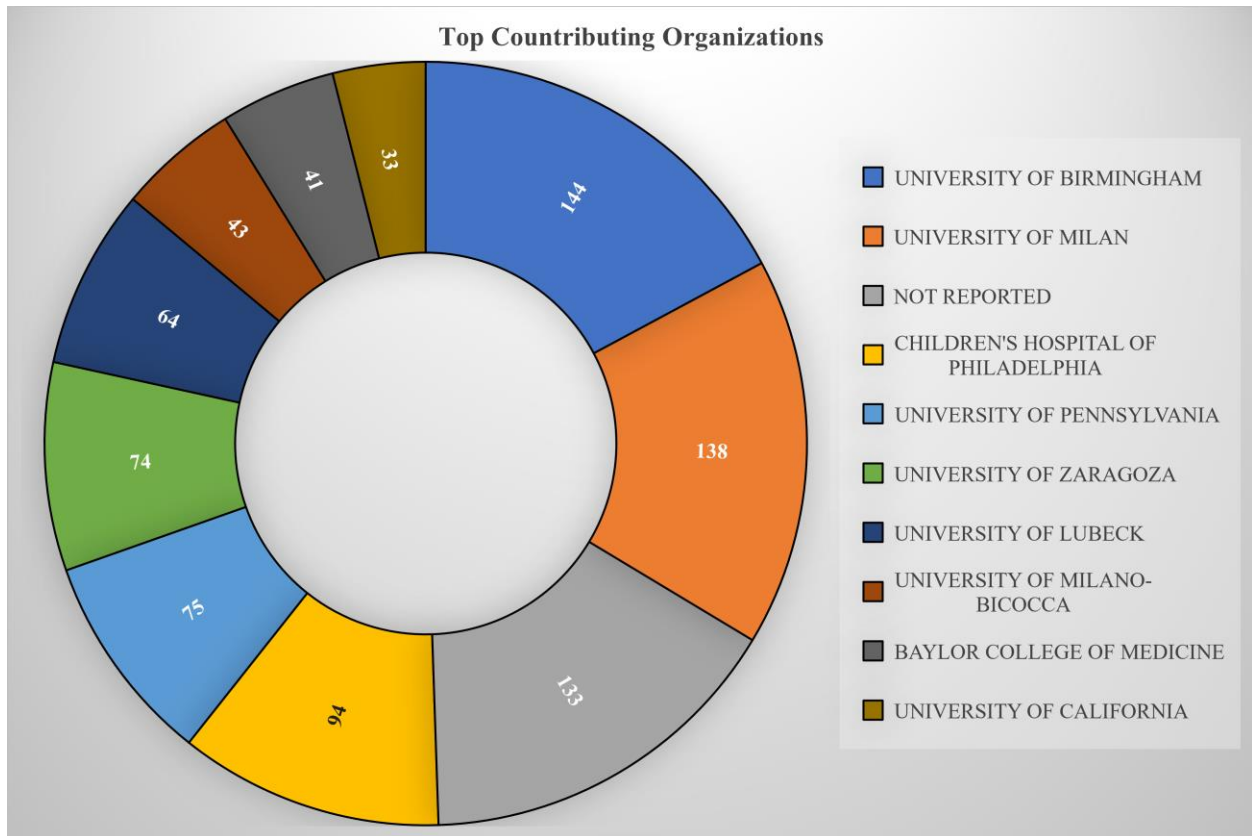

Figure 1: Top 10 organizations contributing to CdLS research

for contributing 94 and 75 documents respectively. These findings are like the results of Wang et al. (2016) who discovered that majority of MERS literature was contributed by the universities. Likewise, the University of Hong Kong and Harvard University were highly contributing

organizations for Coronavirus research (Danesh & Ghavidel, 2020; Ram, 2020) and regulatory T-cell research (Zongyi et al., 2016), respectively. However, VA Medical Center contributed the highest number of research publications on m-health (Sweileh et al., 2017).

Table 6: Top 20 sources of publication for CdLS research

Rank	Sources (n = 403)	Articles	Percentage	Cumulative percentage
1	American Journal of Medical Genetics	63	7.99	7.99
2	American Journal of Medical Genetics Part A	43	5.46	13.45
3	Clinical Genetics	16	2.03	15.48
3	Journal of Medical Genetics	16	2.03	17.51
5	American Journal of Medical Genetics Part C: Seminars in Medical Genetics	15	1.90	19.42
6	Journal of Intellectual Disability Research	13	1.65	21.07
7	Minerva Pediatrica	10	1.27	22.34
8	International Journal of Pediatric Otorhinolaryngology	9	1.14	23.48
8	Prenatal Diagnosis	9	1.14	24.62
10	Clinical Dysmorphology	8	1.02	25.63
10	Japanese Journal of Anesthesiology	8	1.02	26.65
12	Genetic Counseling	7	0.89	27.54
12	Human Molecular Genetics	7	0.89	28.43
12	Human Mutation	7	0.89	29.31
15	European Journal of Medical Genetics	6	0.76	30.08
15	Journal of Autism and Developmental Disorders	6	0.76	30.84
17	European Journal of Human Genetics	5	0.63	31.47
17	Human Genetics	5	0.63	32.11
17	Journal of Neurodevelopmental Disorders	5	0.63	32.74
17	Paediatric Anaesthesia	5	0.63	33.38

The analysis for sources of publication disclosed that 788 publications on Cornelia de Lange Syndrome were published in 403 sources. Majority of these documents were published in journals while less than one percent were published as book chapters. Top 20 sources of publication have been presented in Table 6, which published one third of total documents. Top seven sources published CdLS literature in double digit while remaining sources published less than ten documents. The *American Journal of Medical Genetics* was the most relevant source of publication for Cornelia de Lange Syndrome research literature, which published 63 documents

(7.99%), followed by *American Journal of Medical Genetics Part A,* with 43 documents (5.46%) and *Clinical Genetics,* with 16 documents (2.03%).

Results of this study exhibited that *American Journal of Medical Genetics* was the most relevant source of publication for Cornelia de Lange Syndrome research. However, some earlier studies found that *Journal of Hand Surgery - American Volume, Journal of Virology, Archives of Dermatology, Journal of Medical Internet Research, Journal of Immunology, and Chest journal* were most preferred journals for research on Carpal Tunnel Syndrome (Ram, 2019), Coronavirus (Ram, 2020), Middle East Respiratory Syndrome (Wang et al., 2016), MERS-CoV (Zyoud, 2016), Stevens Johnson Syndrome and Toxic Epidermal Necrolysis (Sweileh, 2017), m-health (Sweileh et al., 2017), regulatory T-cell (Zongyi et al., 2016), and Obstructive Sleep Apnea (Huamani et al., 2015), respectively.

Citations are vital indicator for comparative analysis of publications to evaluate importance of research publications and performance of researchers in any field of research. The importance of any publication in its field is indicated by the number of citations it has received (Ram, 2020). Twenty most cited research publications on CdLS have been presented in Table 7 showing the name of first author, year of publication, and source of publication. Data analysis discovered that the research paper titled "Cornelia de Lange Syndrome is caused by mutations in NIPBL, the human homolog of Drosophila melanogaster Nipped-B" by Krantz et al. published in *Nature Genetics*, volume 36, number 6, 2004 was the most important research article on CdLS as it received highest number of citations (491) at a rate of 28.88 citations per year. Another article titled "NIPBL, encoding a homolog of fungal Scc2-type sister chromatid cohesion proteins and fly Nipped-B, is mutated in Cornelia de Lange Syndrome" by Tonkin et al., from same issue of *Nature Genetics,* received second highest number of citations (434) at a rate of 25.53 citations per year.

However, an article titled "HDAC8 mutations in Cornelia de Lange Syndrome affect the cohesin acetylation cycle" by Deardorff et al., published in *Nature*, volume 489, number 7415, 2012, received 319 citations at the highest rate of citations (35.44) per year.

Table 7: Most cited documents in CdLS research

Rank	Documents (n = 788)	Total Citations	Total Citations per Year
1	Krantz I D, 2004, Nat Genet	491	28.88
2	Tonkin Et, 2004, Nat Genet	434	25.53
3	Deardorff M A, 2007, Am J Hum Genet	343	24.50
4	Musio A, 2006, Nat Genet	330	22.00
5	Deardorff M A, 2012, Nature	319	35.44
6	Gillis L A, 2004, Am J Hum Genet	225	13.24
7	Kline A D, 2007, Am J Med Genet Part A	176	12.57
8	Kawauchi S, 2009, PLOS Genet	150	12.50
9	Liu J, 2009, Clin Genet	139	11.58
10	Moss J, 2009, J Autism Dev Disord	133	11.08
11	Dorsett D, 2007, Chromosoma	125	8.93
12	Bhuiyan Z A, 2006, J Med Genet	110	7.33
13	Berney T P, 1999, Arch Dis Child	108	4.91
14	Mannini L, 2013, Hum Mutat	93	11.63
15	Kline A D, 2007, Am J Med Genet Part C Semin Med Genet	92	6.57
15	Ireland M, 1993, Am J Med Genet	92	3.29
17	Hyman P, 2002, Am J Ment Retard	86	4.53
18	Dorsett D, 2009, Ann New York Acad Sci	85	7.08
18	Zhang B, 2007, Development (Cambridge)	85	6.07
20	Miller J L, 2003, Early Hum Dev	84	4.67

The word analysis of CdLS publications based on "Keywords Plus" has been presented in Figure 2 showing ten most frequently used words and number of their occurrences. Words have been listed in the legend from left to right in rows. The analysis discovered that "de Lange Syndrome" was the most frequently used word having 1,332 occurrences followed by human (857 occurrences) and female (751 occurrences).
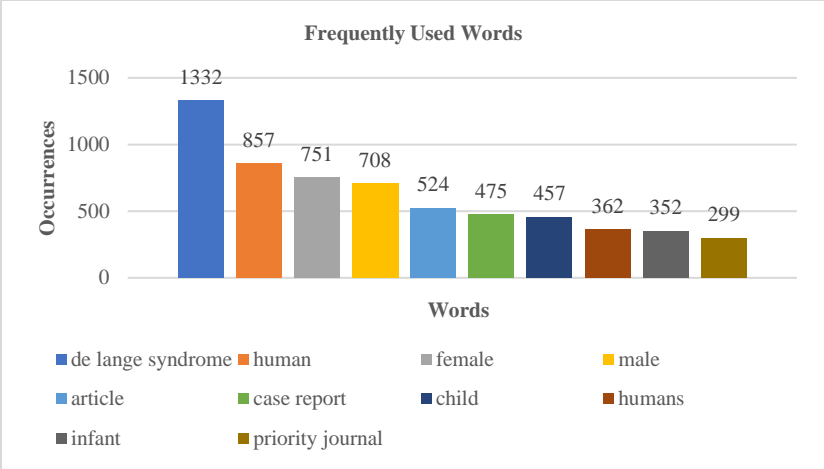
Figure 2: Most frequently used words in CdLS research

Conclusion

Based on the results of this study, it can be concluded that research on Cornelia de Lange Syndrome is published in diverse publication sources in a variety of output formats. CdLS research originated from a large community of researchers who enjoy a high level of collaboration. Interest in CdLS research is increasing with the passage of time which is evident from the increased publication activity. It can also be concluded that results may vary for the author productivity rankings depending upon the crediting method used for publications and author impact depends upon a combination of several variables including but not limited to the length of publishing career and number of publications. CdLS research is largely concentrated in American and European institutions, which is mostly published by the journals originating from these countries. It is recommended that researchers should consider using multiple publication crediting methods while working on author productivity rankings. It is further recommended to explore different aspects of CdLS research literature which have not been covered in this study with different data sets.

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