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# Use of the bibliometric in rare diseases: taking Wilson disease personally

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## Abstract

**Background:** Bibliometric have been widely applied to the evaluation of academic productivity. However, those of individuals or institutions on a specific disease have not been explored. The aim of the present study is to conduct a bibliometric analysis of particular rare disease and investigate whether those doctors and hospitals with higher index screened by this method specialize in this disease.

**Methods:** A representative rare disease, Wilson disease (WD), was searched on Clarivate Analytics' Web of Science and Elsevier's Scopus, which was published in English between 1 January 2001 and 31 December 2020. Clinical authors and medical institutions with the most papers were screened, and their total number of publications and citations, h-index and g-index were computed and then ranked by h-index.

**Results:** A total of 6856 and 6193 papers and 200 and 160 authors were got from WoS and Scopus, respectively. Scopus provided 160 institutions. The above bibliometric indices were calculated in 100 researchers and 80 institutions, and top 30 authors (Top-30a) and top 20 institutions (Top-20i) of them based on the h-index were listed in the tables. Top-30a came from seven specialties and 13 countries whose median (interquartile range) h-index was 14 (12–19.5) (range 10–28) which was located between associate and full professors in some other disciplines. Top-20i was distributed in 13 countries whose mean  $\pm$  standard deviation of the h-index was  $15 \pm 4.9$  (range 10–27).

**Conclusions:** The related specialists and medical institutions of WD screened by specific disease bibliometric analysis are eminent and credible and benefit WD patients to obtain reliable medical treatment. This model may be suitable for other rare diseases.

**Keywords:** Academic productivity, Wilson disease, Bibliography, Bibliometric

## Background

In the field of academic medicine, scientific productivity is still a crucial and more objective factor for evaluating scholarly achievements and promotion [1, 2].

Bibliometric provides a quantitative method that is widely accepted thus far. The h-index [3] and g-index [4] are two of the most broadly known and used bibliometric parameters on the individual and departmental levels [5–9]. “The h-index is defined as an individual having h papers with at least h citations” [3]. It balances both the total publications and citations per publication, and has good stability since it is less affected by low-cited articles [10]. The g-index means that the most cited g articles earned at least  $g^2$  citations [4]. It could provide weight to a h-index that is too-low when a small number of very influential papers result in an uneven distribution of citations.

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At present, bibliometric studies focus on the global academic productivity of individuals or departments, rather than a single disease. It is impossible for an expert to have high scholarly achievement in varied diseases. A single disease's bibliometric analysis may be necessary when we need to assess the influence of a doctor on a particular disease, especially for the diagnosis and therapy of rare diseases.

To date, there is no exact and uniform definition and count of rare diseases. A disease that affects fewer than 200,000 people in the United States is rare [11]. Meanwhile, in the European Union in 2000, less than 5 in 10,000 people was considered a rare disease [12]. It is estimated the number of rare diseases is more than 10,000 [13]. Therefore, all rare diseases face a similar clinical dilemma, since most clinicians and departments do not have more opportunities to deal with rare disease patients which precisely prevents them from accumulating adequate experience to recognize and manage rare diseases. Then, diagnosis is often delayed or wrong followed by increased financial burden and physical torment. Therefore, the answers to the following questions may be particularly critical: Who are authentic clinical specialists of rare diseases? How can their clinical competence be evaluated? Would rare disease bibliometric research for individuals and clinical institutions be the perfect solution?

Wilson disease (WD) is a rare but potentially treatable and inherited disorder of copper metabolism with a prevalence of 2.9–5.87 per 100,000 [14–17]. As far as I know, there is abundant literature on WD available for analysis. In this study, we took WD as an example of a rare disease to conduct a bibliometric analysis based on literature published from 2001 to 2020, investigated the academic productivity of experts and institutions in this special field, and explored whether the experts and institutions singled out by particular disease bibliometrics were reliable in the field of that rare disease.

## Methods

### Search and inclusion criteria

All publications were searched using “Hepatolenticular Degeneration” as the medical subject heading (MeSH) term and related entry terms which came from the National Center for Biotechnology Information (NCBI). The above terms were retrieved in the “Article title, Abstract and Keywords” of Elsevier's Scopus and the “Topic” of Clarivate Analytics' Web of Science (WoS) to analyze the bibliometric information of relevant dissertations that were published between 1 January 2001 and 31 December 2020 in English and document types were limited to articles or review articles or case report or letters or clinical trial or report (Additional file 1). The

study data were collected in December 2021 over one week period. For the purpose of this study, the inclusion criteria of this literature were that (1) the details must be related to WD (known or called also hepatolenticular degeneration); and (2) their authors need to involve clinicians, not just fundamental science (nonclinical) researchers. We can only identify whether the author is a clinical researcher based on the institution of the latest published articles, and those obscure and uncertain results would not be involved in this study.

### Citation data sources

The list of authors with the most papers and their publication data were extracted from Scopus and WoS databases on the same day. One author may have various abbreviations of the first name, for instance, Michael L Schilsky, Michael Schilsky, MICHAEL Schilsky, ML Schilsky, M L Schilsky and M Schilsky are the same person. We sorted by author's last name to maximize the discovery of all his articles. Article title, authors, department, institution, abstract, PubMed unique identifier, publication year and cited times were extracted. Our colleagues checked and merged raw data from two databases to unify the author's name and ignore the nonclinical researcher and erase the duplicate items and unrelated themes. The top cited articles and the list of institutions were provided by Scopus. Their publication data were screened as above. Suspicious items were identified from the original literature. The above data were then saved as a spreadsheet for bibliometric calculations. We cannot identify the most-paper authors' sex, age, and work situation, for instance, in-service, retired or emeritus. Although we tried our best to retrieve the initials of those authors in various forms, the omission of a few papers may still be inevitable. Since there is no uniform named standard and some institutions have renamed, merged or ceased to exist, we did not consider the literature data of institutions sourced from Scopus to be accurate.

### Bibliometric indices

The most-paper authors' following indices were calculated: total number of publications; the total number of citations; h-index; g-index. The top 20 most-cited papers (Top-20p) and their authors were listed. We ranked the top 30 authors/doctors (Top-30a) worldwide according to their scientific research output and h-index. The network mapping of Top-30a and their coauthors was charted with the VOSviewer program [18], which reflects their influence power in this specialty field. The h-index, which is used to calculate individual academic output, [3] was borrowed here to calculate the academic performance of organizations and obtain the top 20 medical institutions (Top-20i).

## Results

### Characteristics of publications

A total of 6856 and 6193 papers related to WD were searched from WoS and Scopus respectively. The different names of article types in these databases resulted in discrepancies in the results, although the MeSH was the same. The Top-20p, which was taken part in by clinicians, is shown in Table 1. Seven of the top 10 most-cited papers are guidelines or reviews, and most of them were published 10 years ago [19–25]. Notably, compared with only one laboratory study on the mouse model of WD [26], the other 19 studies were all clinical investigations or reviews. The highest citation, 772, was produced by a practice guideline article [23], which belongs to Eve A. Roberts and Michael L. Schilsky, published in *Hepatology* in 2008 and represented the position of the American Association for Study of Live Disease (AASLD). Michael L. Schilsky and Peter Ferenci, two prestigious professors, were the principal coauthors in Top-20p. Michael L. Schilsky [27], professor of medicine and medical director in Adult Liver Transplant at Yale-New Haven Transplantation Center, was involved in seven publications, four of which ranked first to fourth [19, 22–24] on Top-20p. Moreover, the first and fourth papers are clinical practice guidelines of AASLD and the European Association for the Study of the Liver (EASL) respectively. Another eminent researcher, Peter Ferenci [28, 29], liver expert in the division of gastroenterology and hepatology, comes from the department of medicine III of Medical University Vienna. He was the chairman of EASL Clinical Practice Guidelines: Wilson's disease [24] and had 6 publications listed on Top-20p.

### Top authors' academic productivity

WoS and Scopus presented 200 and 160 authors whose median (interquartile range, IQR) paper numbers were 25(19, 35) (range 13–102) and 12(10, 17) (range 9–75) respectively (data not shown). The top 100 authors on WoS with the most articles were retrieved, whose article numbers ranged from 18 to 102. Then, their records on Scopus were downloaded and merged with those of WoS to calculate the h-index. According to the h-index, Top-30a who were productive in the WD field, coming from seven specialties and 13 countries (Fig. 1A), are listed in Table 2. The median (IQR) of the h-index is 14 (12–19.5), ranging from 28 to 10. Anna Czlonkowska tops the list with the highest h-index, 28, and the largest number of papers, 98, which generated 3039 citations. Peter Ferenci and Michael L. Schilsky followed behind closely with h-index values of 27 and 26, respectively. Nevertheless, their g-indices were higher than Anna Czlonkowska's and occupied the top two positions. Tomasz Litwin, similar to his colleague Anna Czlonkowska, had a number

of articles and a disproportionate h-index. In terms of total cites, Michael L. Schilsky, who was coauthor of both AASLD and EASL's guidelines about WD [23, 24], ranked first by virtue of 4953 citations of 77 articles. Furthermore, his g-index score, 70, was far higher than his peers, just like his total cites. Surprisingly, Dominik Huster and Eve A. Roberts, ranking 13th and 19th based on the h-index, merely relied on 20 papers to earn 1276 citations and 1504 citations, respectively. We made a heatmap based on the number of articles published by each author per year (Fig. 2). It showed that the academic output of top researchers, such as Anna Czlonkowska, Peter Ferenci, Michael L. Schilsky and Wolfgang Stremmel, was not only consistent but also productive. Impressively, Jean Marc Trocello and Aurelia Poujois, who both came from the French National Reference Centre for Wilson Disease (Paris), and Karolina Dziezyc (Warsaw) have had a good start to the last decade and may have a bright future (Fig. 2).

Broadly speaking, Asian researchers' academic productivity, especially Japan and China, at least in English publications, was inefficient. The higher h-index, g-index, total articles and total cites of authors, by contrast, all belonged to Europe and the United States (Table 2, Fig. 1A). In addition, approximately half of Top-30a came from neurology and one third of them were gastroenterologists and hepatologists (Fig. 1B). Most of Top-30a were employees of the hospital affiliated with the medical university (Table 2).

The network relationships of Top-30a and their coauthors are presented in Fig. 3. Obviously, in the lower left corner, Chinese researchers, RM. Yang, and ZY. Wu, and Japanese scholar, M. Harada, do not have academic cooperation with the right sophisticated and interlinked cluster, which was made up of European and American peers. In line with the performance of Top-30a in Table 2, generally, the higher up authors also had larger circles, more complex network relationships and a more central position in Fig. 2, for instance, A. Czlonkowska, P. Ferenci, K. Weiss and M. Schilsky. Top-30a in the same area and with the same color generally belong to the same cluster or even the same institution.

### Top institution' academic productivity

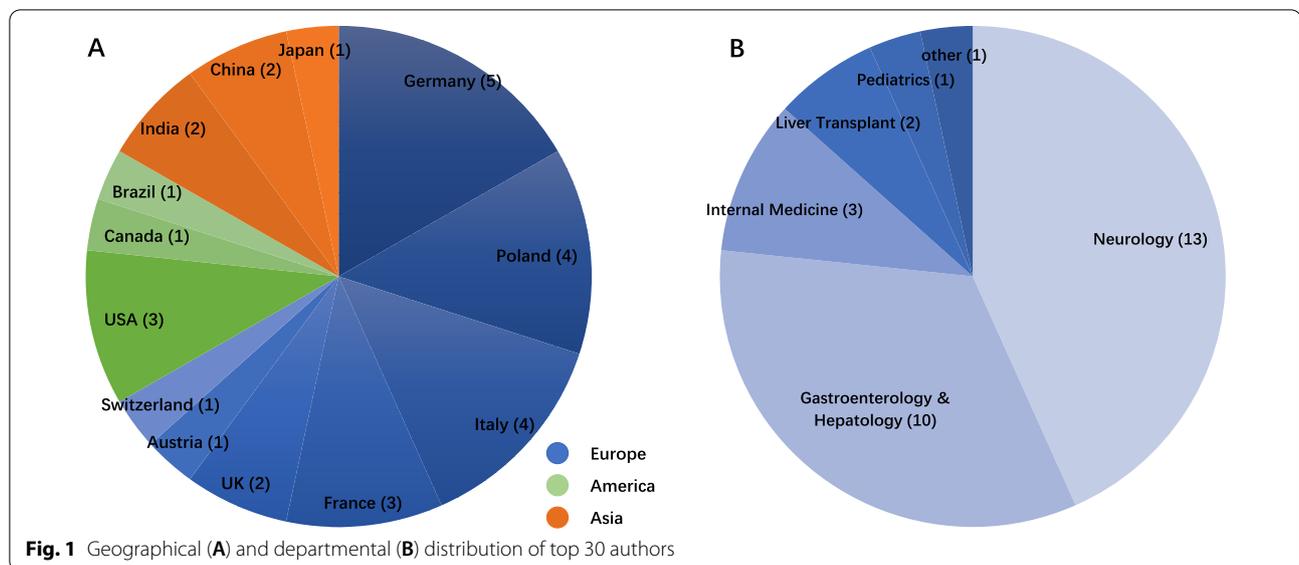
Scopus demonstrated 160 institutions based on article counts whose median (IQR) was 25 (19, 35) (range 16–88). The top 80 (50%) institutions' papers, which ranged from 25 to 88, were proven and their related indices were calculated. In light of the h-index again, Table 3 ranks the Top-20i located in 13 countries whose h-index was between 10 and 27, with a mean  $\pm$  standard deviation of  $15 \pm 4.9$ . In contrast with the affiliated institutions of Top-30a in Table 2, unexpectedly, all indices of Top-20i

**Table 1** Top 20 most-cited papers related to Wilson Disease from 2001 to 2020, whose authors involved clinicians

| Rank | Title   | Total cites | Authors  | Journal                                    | Year | Type      |
|------|---|-------------|--|--|------|-----------|
| 1    | Diagnosis and treatment of Wilson disease: An update  | 772         | Eve A. Roberts, Michael L. Schilsky  | Hepatology                                 | 2008 | Guideline |
| 2    | Wilson's disease  | 745         | Aftab Ala, Ann P Walker, Keyoumars Ashkan, James S. Dooley, Michael L. Schilsky  | Lancet                                     | 2007 | Review    |
| 3    | Diagnosis and phenotypic classification of Wilson disease   | 549         | Peter Ferenci, Karel Caca, Georgios Loudianos, Giorgina Mielì-Vergani, Stuart Tanner, Irmin Sternlieb, Michael L. Schilsky, et al                                | Liver International                        | 2003 | Review    |
| 4    | EASL Clinical Practice Guidelines: Wilson's disease   | 548         | Peter Ferenci, Anna Czlonkowska, Wolfgang Strommel, Roderick Houwen, William Rosenberg, Michael L. Schilsky, et al   | Journal of Hepatology                      | 2012 | Guideline |
| 5    | Wilson's disease and other neurological copper disorders  | 345         | Oliver Bandmann, Karl Heinz Weiss, Stephen Kaler   | The Lancet Neurology                       | 2015 | Review    |
| 6    | Clinical presentation, diagnosis and long-term outcome of Wilson's disease: A cohort study  | 337         | UTA Merle, Matthias R. Schaefer, Peter Ferenci, Wolfgang Strommel  | Gut  | 2007 | Article   |
| 7    | A practice guideline on Wilson disease  | 307         | Eve A. Roberts, Michael L. Schilsky  | Hepatology                                 | 2003 | Review    |
| 8    | Wilson Disease  | 266         | Jonathan D Gitlin  | Gastroenterology                           | 2003 | Review    |
| 9    | Wilson's disease in children: 37-year experience and revised King's for liver transplantation   | 230         | Anil Dhawan, Rachel M. Taylor, Paul Cheeseman, Pamela De Silva, Leah Katsiyannakis, Giorgina Mielì-Vergani   | Liver Transplantation                      | 2005 | Article   |
| 10   | Treatment of Wilson disease with ammonium tetrathiomolybdate—IV. Comparison of tetrathiomolybdate and trientine in a double-blind study of treatment of the neurologic presentation of Wilson disease | 199         | George J. Brewer, Fred Askari, Matthew T. Lorincz, Martha Carlson, Michael L. Schilsky, Karen J Kluin, et al   | Archives of Neurology                      | 2006 | Article   |
| 11   | Wilson's disease: An update   | 197         | Shyamal K. Das, Kunal Ray  | Nature Clinical Practice Neurology         | 2006 | Review    |
| 12   | A genetic study of Wilson's disease in the United Kingdom   | 186         | Alison J Coffey, Miranda Durkie, Stephen Hague, Kirsten McLaughlin, Jennifer Emmerson, Christine Lo, Stefanie Klaffke, Christopher J Joyce, Anil Dhawan, et al   | Brain                                      | 2013 | Article   |
| 13   | Regional distribution of mutations of the ATP7B gene in patients with Wilson disease: Impact on genetic testing   | 177         | Peter Ferenci  | Human Genetics                             | 2006 | Review    |
| 14   | High copper selectively alters lipid metabolism and cell cycle machinery in the mouse model of Wilson disease   | 159         | Dominik Huster, Tina D. Purnat, Jason L. Burkhead, Martina Ralle, Oliver Fiehn, Franziska Stuckert, N Erik Olson, Daniel Teupser, Svetlana Lutsenko              | Journal of Biological Chemistry            | 2007 | Article   |
| 15   | Wilson disease: Description of 282 patients evaluated over 3 decades  | 157         | Arun B. Taly, Meenakshi-Sundaram S., Sanjib Sinha, HS. Swamy, GR. Arunodaya  | Medicine                                   | 2007 | Article   |
| 16   | Late-Onset Wilson's Disease   | 155         | Peter Ferenci, Anna Czlonkowska, UTA Merle, Szalay Ferenc, Grazyna Gromadzka, Cihan Yurdaydin, Wolfgang Vogel, Radan Bruha, Hartmut T Schmidt, Wolfgang Strommel | Gastroenterology                           | 2007 | Article   |
| 17   | Neurologic Wilson's disease   | 148         | Matthew T. Lorincz   | Annals of the New York Academy of Sciences | 2010 | Review    |
| 18   | Wilson's disease: Cranial MRI observations and clinical correlation   | 144         | Sanjib Sinha, Arun B. Taly, Saiprasad Ravishankar, et al   | Neuroradiology                             | 2006 | Article   |

**Table 1** (continued)

| Rank | Title   | Total cites | Authors  | Journal                                  | Year | Type    |
|------|---|-------------|--|--|------|---------|
| 19   | Screening for Wilson disease in acute liver failure: A comparison of currently available diagnostic tests | 142         | Jessica D. Korman, Irene Yolenberg, Jody Balko, Joe Webster, Frank V Schiodt, Robert H. Squires Jr, Robert J Fontana, William M. Lee, Michael L. Schilsky, | Hepatology                               | 2008 | Article |
| 20   | Diagnostic value of quantitative hepatic copper determination in patients with Wilson's disease           | 141         | Peter Ferenci, Petra Steindl-Munda, Wolfgang Vogel, Wolfgang Jessner, Michael Gschwantler, Rudolf Stauber, et al   | Clinical Gastroenterology and Hepatology | 2005 | Article |



were much lower, which was why we only retrieved 20. Due to universal nomenclature being nonexistent and job-hopping, the above differences may be reasonable. The Institute of Psychiatry and Neurology in Warsaw, Poland, whose four authors are presented in the Top-30a list (Table 2), ranked first in the Top-20i list with 27 of h-index, 45 of g-index and 2440 total cites yielded by 88 papers, followed by the University Hospital of Heidelberg, from which three researchers in the Top-30a list came (Tables 2, 3). In addition, there were four institutions that acquired very higher cites that rely on relatively fewer articles: University (Hospital) of California Davis, First Faculty of Medicine of Charles University, King's College of Hospital and University of Toronto Hospital for Sick Children. Geographically, as with Top-30a, Germany, the US and Italy remained the main players. Furthermore, Switzerland, Japan and China disappeared from Table 3, albeit they had outstanding individual performance (Table 2).

## Discussion

Taking the WD as an example and using bibliometric methods, we have screened the Top-30a and Top-20i, which may be the most trustworthy in the diagnosis and treatment of WD. This model may provide a basis to help WD patients choose the appropriate doctor or medical institution and may even be beneficial for undiagnosed dubious patients.

Regarding bibliometric, the h-index is the most broadly used and accepted measure of scholarly productivity and even for hiring, promotion, award and funding decisions [1, 6, 30–32]. Some studies have been conducted in fields such as neurosurgery, pediatric, academic

otolaryngology, anesthesia, radiology and chronic pain medicine [2, 5–9]. This study may be the first attempt to calculate the h-index using papers from a specific disease rather than all literature. The h-index increases gradually with advancing academic ranks from lecturer to assistant professor, associate professor (AP), full professor (FP) and finally chairman [32]. Hirsch found that a general value for promotion to AP or FP would be ~12 and ~18 respectively [3]. The h-index is robust because of its insensitivity to lowly cited articles in a researcher's album [5, 10, 32]. The common view is that the h-index will never fall and with no doubt is influenced by the author's scholarly career period, which is seen as a drawback. However, this could be a benefit in the rare disease field, where senior physicians tend to have abundant experience.

Almost all physicians of Top-30a are affiliated with university hospitals or research institutes (Table 2). Top-20i better illustrates this point (Table 3). Based on data from 2001 to 2020 alone and limited in particular disease (WD), the median (IQR) h-index of Top-30a is 14 (12–19.5), nearly equal to that of FP of chronic pain physicians in the USA: 16.5 (6, 30) [2], lower than that of anesthesia FP in the UK: 21 (16–26), both in their whole period [7]. Compared to the general academic pediatrician, the median h-index (14) and g-index (22.5) of Top-30a were slightly lower than those of FP (16 and 29) and markedly higher than AP (6 and 11) [9]. Although we did not list researchers' titles, by comparison with the h- and g-index of AP and FP of other disciplines, we believe that these data obtained from our method can prove to us that these experts' and institutions' academic productivity in the field of WD is sufficiently convincing, they are also most likely to be good at WD. In fact, considering

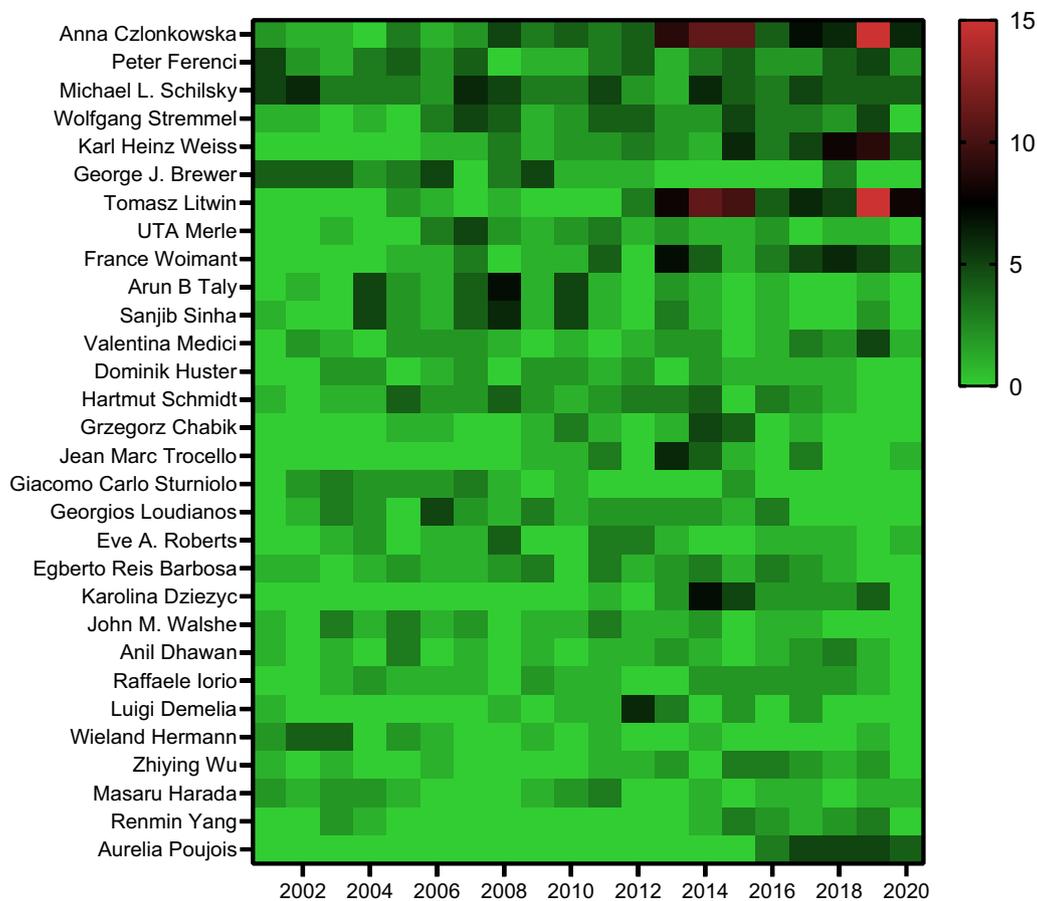
**Table 2** Top 30 authors who are clinicians and had the highest h-index according to their articles related to Wilson Disease from 2001 to 2020

| Rank | Name                    | h-index | g-index | Total articles | Total cites | Specialty                       | Department & Institution   |
|------|-------------------------|---------|---------|----------------|-------------|---------------------------------|--|
| 1    | Anna Czlonkowska        | 28      | 51      | 98             | 3039        | Neurology                       | Second Department of Neurology, Institute of Psychiatry and Neurology, Warsaw, Poland  |
| 2    | Peter Ferenci           | 27      | 53      | 53             | 3726        | Gastroenterology and Hepatology | Division of Gastroenterology and Hepatology, Medical University of Vienna, Vienna, Austria   |
| 3    | Michael L. Schilsky     | 26      | 70      | 77             | 4953        | Liver Transplant                | Departments of Medicine and Surgery, Yale University Medical Center, New Haven, CT, USA  |
| 4    | Wolfgang Stremmel       | 25      | 48      | 48             | 2556        | Gastroenterology and Hepatology | Department of Gastroenterology and Hepatology, University Hospital of Heidelberg, Heidelberg, Germany                              |
| 5    | Karl Heinz Weiss        | 23      | 40      | 51             | 1674        | Gastroenterology and Hepatology | Department of Gastroenterology and Hepatology, University Hospital of Heidelberg, Heidelberg, Germany                              |
| 6    | George J. Brewer        | 23      | 36      | 36             | 1801        | Internal Medicine               | Department of Human Genetics and Department of Internal Medicine, University of Michigan Medical School, Ann Arbor, USA            |
| 7    | Tomasz Litwin           | 21      | 38      | 74             | 1695        | Neurology                       | Second Department of Neurology, Institute of Psychiatry and Neurology, Warsaw, Poland  |
| 8    | France Woimant          | 19      | 27      | 45             | 846         | Neurology                       | Neurology Department and National Reference Centre for Wilson's Disease, AP-HP, Lariboisière University Hospital, Paris, France    |
| 9    | UTA Merle               | 19      | 26      | 26             | 1318        | Gastroenterology and Hepatology | Department of Gastroenterology, University Hospital Heidelberg, Heidelberg, Germany  |
| 10   | Arun B Taly             | 18      | 32      | 33             | 1049        | Neurology                       | Department of Neurology, National Institute of Mental Health and Neuro Sciences, Bangalore, India                                  |
| 11   | Sanjib Sinha            | 17      | 31      | 33             | 1004        | Neurology                       | Department of Neurology, National Institute of Mental Health and Neuro Sciences, Bangalore, India                                  |
| 12   | Valentina Medici        | 17      | 29      | 29             | 918         | Gastroenterology and Hepatology | Division of Gastroenterology and Hepatology, Department of Internal Medicine, University of California Davis, Sacramento, USA      |
| 13   | Dominik Huster          | 17      | 20      | 20             | 1276        | Gastroenterology and Oncology   | Department of Gastroenterology and Oncology, Deaconess Hospital Leipzig, Academic Teaching Hospital University of Leipzig, Germany |
| 14   | Hartmut Schmidt         | 17      | 31      | 36             | 1013        | Transplantation                 | University Hospital of Muenster, Muenster, Germany   |
| 15   | Grzegorz Chabik         | 14      | 18      | 18             | 602         | Neurology                       | Second Department of Neurology, Institute of Psychiatry and Neurology, Warsaw, Poland  |
| 16   | Jean Marc Trocello      | 14      | 20      | 20             | 566         | Neurology                       | Neurology Department and National Reference Centre for Wilson's Disease, AP-HP, Lariboisière University Hospital, Paris, France    |
| 17   | Giacomo Carlo Sturniolo | 14      | 18      | 18             | 631         | Gastroenterology                | Department Surgery & Gastroenterol, University of Padova, Padova, Italy  |
| 18   | Georgios Loudianos      | 13      | 30      | 30             | 1032        | NA                              | Ospedale Regionale Microcitemie, Cagliari, Italy   |

**Table 2** (continued)

| Rank                 | Name                 | h-index      | g-index      | Total articles | Total cites | Specialty             | Department & Institution  |
|----------------------|----------------------|--------------|--------------|----------------|-------------|-----------------------|---|
| 19                   | Eve A. Roberts       | 13           | 20           | 20             | 1504        | Gastroenterology      | Division of Gastroenterology, Hepatology and Nutrition, The Hospital for Sick Children, University of Toronto, Canada,                          |
| 20                   | Egberto Reis Barbosa | 13           | 23           | 28             | 570         | Neurology             | Department of Neurology, University of São Paulo Medical School, São Paulo, Brazil  |
| 21                   | Karolina Dziezyc     | 13           | 21           | 25             | 470         | Neurology             | Second Department of Neurology, Institute of Psychiatry and Neurology, Warsaw, Poland   |
| 22                   | John M. Walshe       | 13           | 22           | 22             | 541         | Internal Medicine     | Formerly of Addenbrookes Hospital, Cambridge and the Middlesex Hospital, London, UK   |
| 23                   | Anil Dhawan          | 12           | 19           | 19             | 858         | Paediatric Hepatology | Paediatric Liver, GI and Nutrition Centre and Mowat Labs, King's College Hospital, Denmark Hill, London, UK                                     |
| 24                   | Raffaele Iorio       | 12           | 21           | 21             | 576         | Pediatrics            | Department of Pediatrics, University of Naples Federico II, Naples, Italy   |
| 25                   | Luigi Demelia        | 12           | 16           | 17             | 287         | Gastroenterology      | Department of Gastroenterology, Hospital of University of Cagliari, Cagliari, Italy   |
| 26                   | Wieland Hermann      | 11           | 17           | 17             | 435         | Neurology             | Department of Neurology, SRO AG Spital Langenthal, Langenthal, Switzerland  |
| 27                   | Zhiying Wu           | 11           | 18           | 18             | 449         | Neurology             | Department of Neurology and Research Center of Neurology in Second Affiliated Hospital, Zhejiang University School of Medicine, Hangzhou, China |
| 28                   | Masaru Harada        | 10           | 18           | 19             | 327         | Internal Medicine     | Third Department of Internal Medicine, School of Medicine, University of Occupational and Environmental Health, Fukuoka, Japan                  |
| 29                   | Renmin Yang          | 10           | 15           | 15             | 247         | Neurology             | Department of Neurology, The Affiliated Hospital of the Neurology Institute, Anhui University of Chinese Medicine, Hefei, China                 |
| 30                   | Aurelia Poujois      | 10           | 15           | 22             | 245         | Neurology             | Neurology Department and National Reference Centre for Wilson's Disease, AP-HP, Lariboisière University Hospital, Paris, France                 |
| Median (range)       | 14 (10–28)           | 22.5 (15–70) | 25.5 (15–98) | 888 (245–4953) |             |                       |   |
| Inter-quartile range | 12–19.5              | 18–33        | 19–38.3      | 523.3–1546.5   |             |                       |   |

USA United States of American, AP-HP Public Assistance-Paris Hospitals, UK United Kingdom



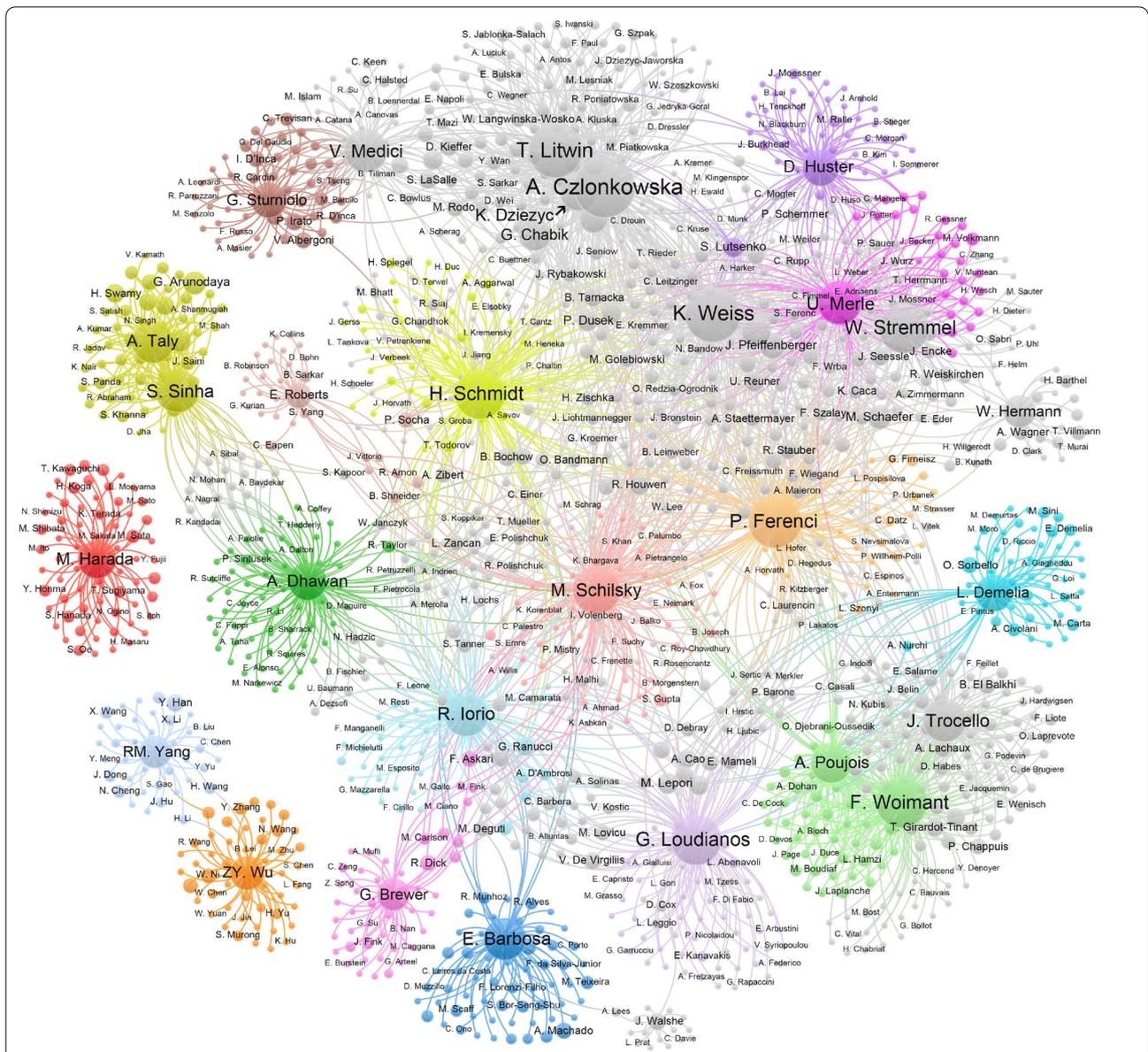
**Fig. 2** Heatmap of the top 30 authors' number of active years of publication

the enormous workload, we cannot retrieve all researchers and institutions listed by WoS and Scopus. We believe that the researchers/institutions who appear on this list are credible. Furthermore, there are four departments, coming from Switzerland, China and Japan, which appeared on the Top-30a table (Table 2) but disappeared on the Top-20i table (Table 3). We suspect that job-hopping had spread their work across different organizations, which may be the cause of the above results.

The European, American and Indian outputs were distinctly better than those of the rest of Asia, such as Japan and China (Tables 2 and 3, Fig. 1A). Language family [33] and collaboration may be the inescapable reasons (Fig. 3). Figure 3 demonstrates that Japanese and Chinese researchers had no academic collaboration with other clusters. Since bibliometric indices do not consider author rank in the manuscripts, multiple coauthors receiving equal credit, the citation and h-index can be strongly influenced by the size of his circle of collaborators [32]. The present study did not involve non-English papers; if these nonnative English speakers' academic

publications in their home countries were included, especially Japan and China, the rankings might change dramatically. We must always clearly realize that these lists only display academic productivity on WD field published in English between 2001 and 2020, which does not mean that the physicians on the list are definitely better at other periods than other scholars in other disease fields. If this method is applied to a specific country, such as Brazil, Japan, China and other non-English speaking countries, it may be more practical to use their native language papers as the object of analysis.

Although, academic influence about some disease does not completely equate to clinical diagnosis and treatment competence. In the domain of rare diseases with the background of rare patients, we believe that those who can sustain consistent and productive scholarly output can be relied upon by these patients. Therefore, the above bibliometric results are dependable. For rare disease patients, it could be used as a clue to help them discover the most suitable doctors and specialist institutions, which are bound to help reduce misdiagnosis and



**Fig. 3** Network visualization of the top 30 authors and their coauthors using VOSviewer. The line indicates that two people have cooperation, and the size of the circle demonstrates relative times they collaborate with others

misthery and reduce the disease burden on individuals and society. Rare disease researchers will have access to more rare clinical cases and experience, which will be more beneficial to their scientific study. An ideal format, we conceive, might be an application that can be installed and operated on smartphones and computers. When the user enters doubtful or definitive diagnosis keywords in the search box, he will obtain relevant helpful experts and medical institutions.

Some limitations might influence the reliability of our results. First, we cannot include all authors and

institutions listed by WoS and Scopus, which may omit some outstanding targets. However, from the perspective of our research purposes, we must ensure that the doctors on the list are excellent, and comprehensiveness is not compulsory. Their median h-index is 14 which is already higher than the AP of many disciplines and nearly reaches the level of FP. Therefore, their academic power is believable. Comparing the paper number of 200 authors (range 13–102) listed on WoS and the top 100 writers of them with the most publications (range 18–102) that were retrieved by us, we hold the

**Table 3** Top 20 institutions contributing manuscripts of Wilson Disease between 2001–2020

| Rank                                | Institution  | Location               | h-index            | g-index            | Total articles      | Total Cites               |
|-------------------------------------|--|------------------------|--------------------|--------------------|---------------------|---------------------------|
| 1                                   | Institute of Psychiatry and Neurology                                | Warsaw, Poland         | 27                 | 45                 | 88                  | 2440                      |
| 2                                   | University Hospital of Heidelberg                                    | Heidelberg, Germany    | 25                 | 43                 | 54                  | 2274                      |
| 3                                   | National Institute of Mental Health and Neuro Sciences               | Bangalore, India       | 20                 | 35                 | 39                  | 1276                      |
| 4                                   | University (Hospital) of Leipzig                                     | Leipzig, Germany       | 19                 | 27                 | 27                  | 1172                      |
| 5                                   | University of Michigan Medical School (Hospital)                     | Ann Arbor, USA         | 19                 | 25                 | 25                  | 1776                      |
| 6                                   | Medical University (Hospital) of Vienna                              | Vienna, Austria        | 18                 | 31                 | 31                  | 1717                      |
| 7                                   | AP-HP, Lariboisière University Hospital                              | Paris, France          | 18                 | 29                 | 40                  | 856                       |
| 8                                   | University (Hospital) of Naples Federico II                          | Naples, Italy          | 15                 | 26                 | 26                  | 856                       |
| 9                                   | University (Hospital) of California Davis                            | California, USA        | 15                 | 22                 | 22                  | 1669                      |
| 10                                  | University (Hospital) of Padova                                      | Padova, Italy          | 15                 | 18                 | 18                  | 640                       |
| 11                                  | Münster University Hospital  | Münster, Germany       | 14                 | 26                 | 26                  | 739                       |
| 12                                  | University of São Paulo School of Medicine (Hospital)                | São Paulo, Brazil      | 13                 | 23                 | 29                  | 581                       |
| 13                                  | First Faculty of Medicine, Charles University                        | Prague, Czech Republic | 13                 | 19                 | 19                  | 1181                      |
| 14                                  | Sanjay Gandhi Post Graduate Institute (Hospital) of Medical Sciences | Lucknow, India         | 12                 | 18                 | 20                  | 337                       |
| 15                                  | University Medical Center Utrecht                                    | Utrecht, Netherlands   | 12                 | 15                 | 15                  | 855                       |
| 16                                  | University (Hospital) of Cagliari                                    | Cagliari, Italy        | 11                 | 22                 | 24                  | 494                       |
| 17                                  | King's College of Hospital   | London, UK             | 10                 | 19                 | 19                  | 1348                      |
| 18                                  | University of Toronto Hospital for Sick Children                     | Toronto, Canada        | 10                 | 16                 | 16                  | 1232                      |
| 19                                  | Yale University School of Medicine (Hospital)                        | New Haven, USA         | 10                 | 15                 | 15                  | 379                       |
| 20                                  | Asan Medical Center, University of Ulsan College of Medicine         | Seoul, South Korea     | 10                 | 15                 | 15                  | 294                       |
| Median ± Standard deviation (range) |  |                        | 15.3 ± 4.9 (10–27) | 24.5 ± 8.7 (15–45) | 28.4 ± 17.2 (15–88) | 1105.8 ± 622.6 (294–2440) |

USA United States of American, AP-HP Public Assistance-Paris Hospitals, UK United Kingdom

opinion that the comprehensiveness is acceptable. The Top-20i also had the same situation. Second, the confidence level of our results only is proven by comparing their bibliometric indices with other subjects and specialties, whereas there is no dependable external validation. Peer review is probably a common practice; however, we doubt that they cannot ignore the impact of academic productivity. Third, WD is a sufficiently researched rare disease, and its related articles

are abundant. When we handle other rare diseases, for instance, Dubin-Johnson Syndrome, which only has 3690 publications searched as above on WoS (data not shown) and may also include many unrelated topics, the situation could be entirely different, and the results could be obscure. Searching in larger categories may be a solution, such as inherited jaundice or inherited liver disease. Further research is still needed to verify these findings.

## Conclusion

As the research has demonstrated, through specific disease bibliometric analysis, we calculated a number of academic productivity indices of researchers and medical institutions in the field of WD. According to the h-index, we ranked and screened out the relevant credible specialists and specialized medical institutions that benefit WD patients to obtain most appropriate medical treatment. This model may be applied to other rare diseases and perhaps to some intractable diseases.

## Abbreviations

AASLD: American Association for Study of Live Disease; AP: Associate professor; EASL: European Association for the Study of the Liver; FP: Full professor; IQR: Interquartile range; MeSH: Medical subject headings; NCBI: National Center for Biotechnology Information; Top-30a: Top 30 authors; Top-20i: Top 20 medical institutions; Top-20p: Top 20 most-cited papers; WD: Wilson disease; WoS: Web of Science.

## Supplementary Information

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**Additional file 1.** The search strategy of database.

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## Author contributions

LC and ZL prepared the material, collected and analyzed the data, and wrote and reviewed the manuscript. YF, LP and JZ collected and analyzed the data and visualization. YZ, YW and NW checked the data and reviewed the manuscript. BR designed and supervised the research and interpreted the results. All authors read and approved the final manuscript.

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## Availability of data and materials

The data that support the findings of this study are available from the corresponding author upon reasonable request.

## Declarations

### Ethics approval and consent to participate

Not applicable. This article does not contain any studies with human or animal subjects. This manuscript does not involve a research protocol requiring approval by the relevant institutional review board or ethics committee.

### Consent for publication

Not applicable.

### Competing interests

The authors declare that they have no competing interests.

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