european burden-eu of disease network

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European Burden of Disease Network

Status of rare diseases in burden of disease studies: A bibliometric analysis

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Background

Decision-makers have considerable challenges in properly allocating resources to **rare diseases** with **low frequency** and **high complexity.**

Approaching the issue from a broad viewpoint based on a bibliometric study of the literature will minimize uncertainties and provide a broad perspective on the subject.

Method

- The method focuses on monitoring a scientific field, delineating its **cognitive structure, and constraining research areas** to determine its evolution (Noyons et al., 1999).»
- Three steps were followed for the bibliometric mapping used in the study:
- **Study design,**
- **Data collection, and**
- **Data analysis**

- **The bibliometrix program based on R** was utilized for the analyses.

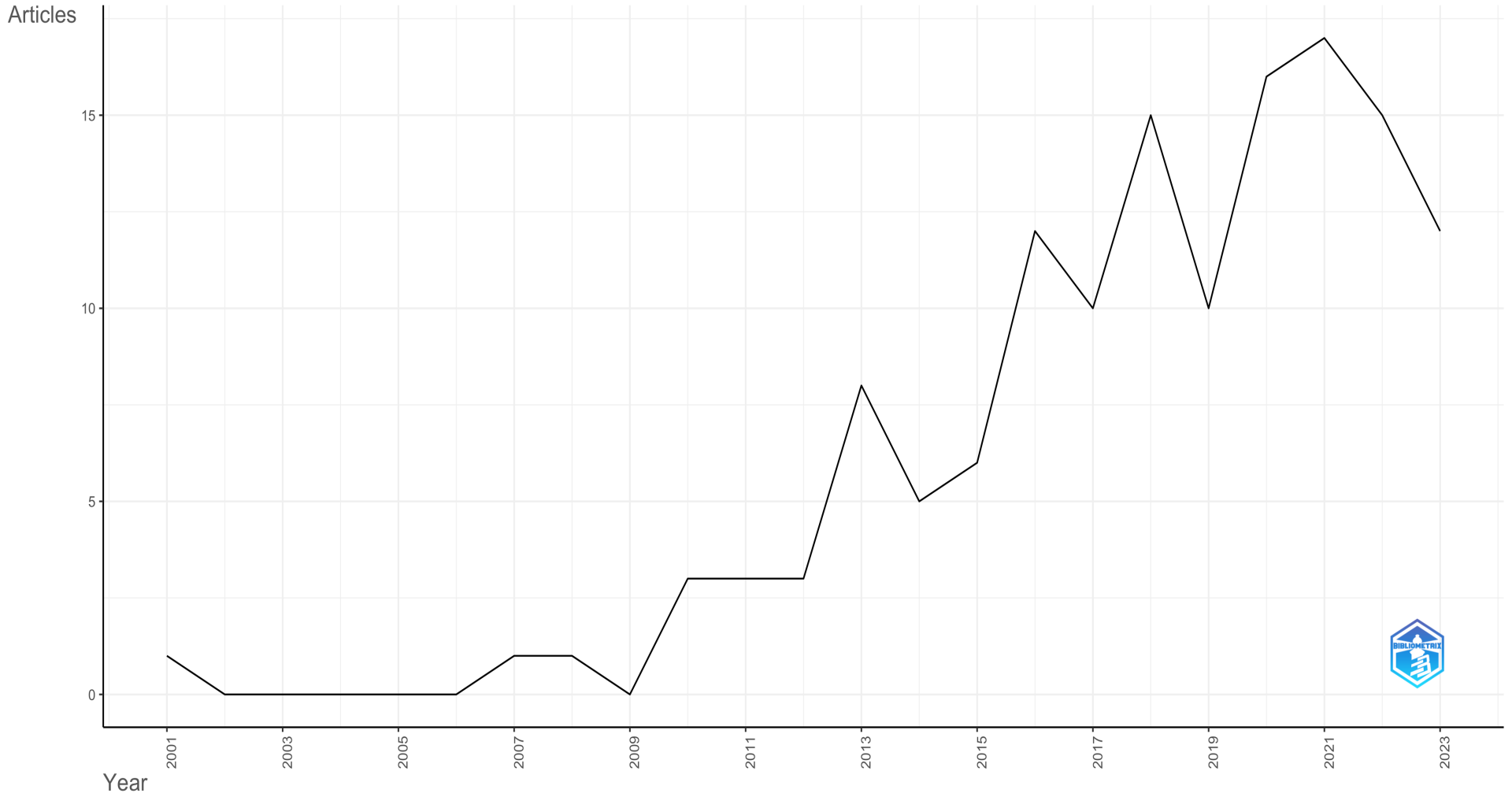
Data Collection

- Web of Science database was employed to collect data according to the specified search strategy.
- Authors conducted initial searches with keywords and combinations.
- Following exclusion criteria, analyses were performed with 138 studies.

Main Information

Description	Results
MAIN INFORMATION	
Timespan	2001:2023
Sources (Journals, Books, etc)	101
Documents	138
Annual Growth Rate %	**11,96**
Document Average Age	**5**
Average citations per doc	**16,74**
References	6641
DOCUMENT CONTENTS	
Keywords Plus (ID)	597
Author's Keywords (DE)	456
AUTHORS	
Authors	1178
Authors of single-authored docs	9
AUTHORS COLLABORATION	
Single-authored docs	9
Co-Authors per Doc	8,96
International co-authorships %	**40,58**
DOCUMENT TYPES	
Article (book chapter, early access, editorial material)	104
review	34

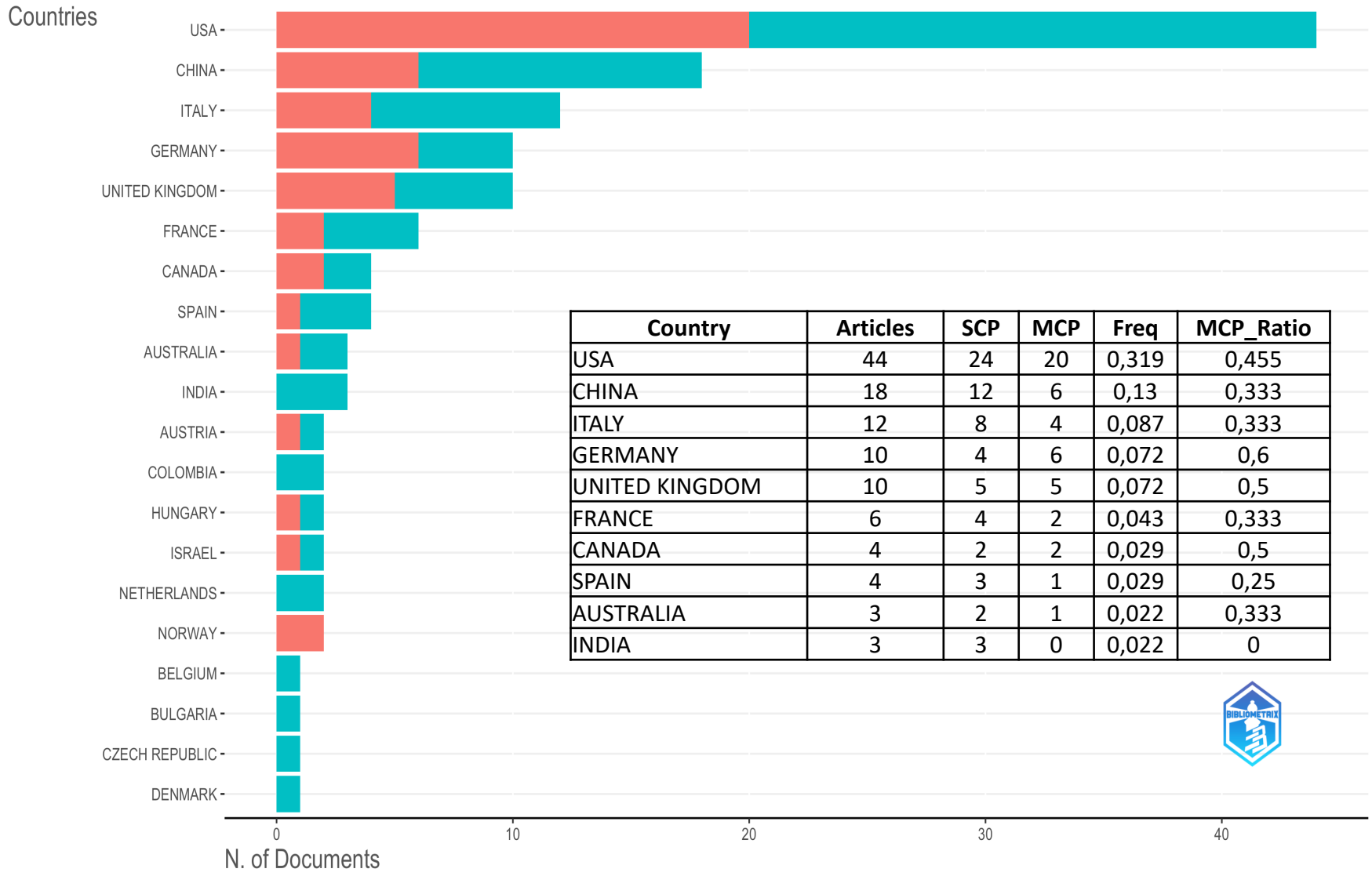
Annual Scientific Production



Most Global Cited Documents

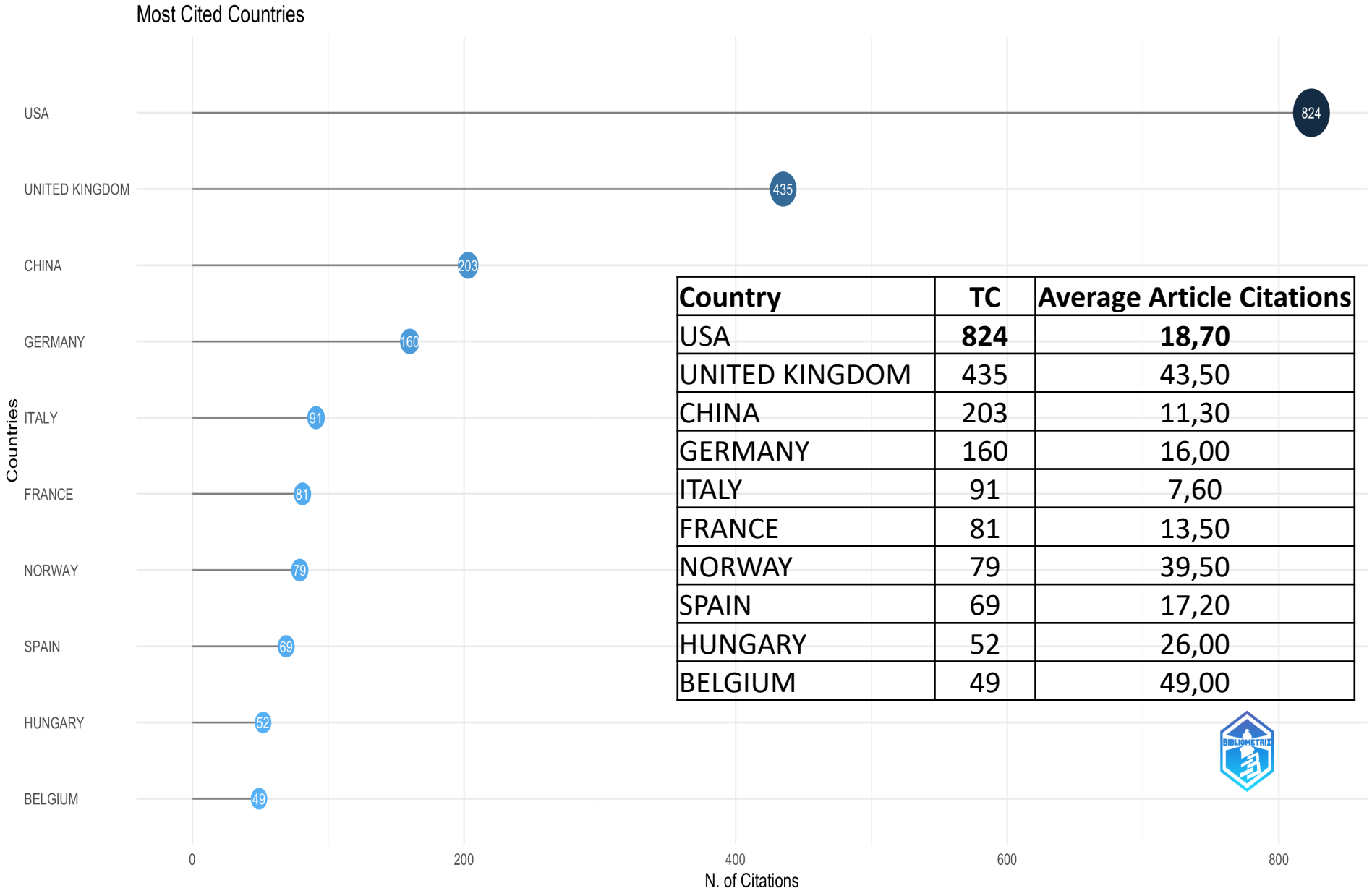
Paper	Title	Total Citations	TC per Year
ANGELIS A, 2015,	Socio-economic burden of rare diseases: A systematic review of cost of illness evidence	**153**	17,00
FERREIRA CR, 2019,	The burden of rare diseases	**133**	26,60
O'HARA J, 2017,	The cost of severe haemophilia in Europe: the CHESS study	**90**	12,86
DHARSSI S, 2017,	Review of 11 national policies for rare diseases in the context of key patient needs	88	12,57
ANKER MS, 2019,	Orphan disease status of cancer cachexia in the USA and in the European Union: a systematic review	78	15,60
SUSSEX J, 2013,	A pilot study of multicriteria decision analysis for valuing orphan medicines	76	6,91
VECCHIE A, 2021,	IL-18 and infections: Is there a role for targeted therapies?	55	18,33
MISTRY PK, 2017,	Outcomes after 18 months of eliglustat therapy in treatment-naïve adults with Gaucher disease type 1: The phase 3 ENGAGE trial	53	7,57
BOLLERSLEV J, 2019,	Management of Endocrine Disease: Unmet therapeutic, educational and scientific needs in parathyroid disorders	52	10,40
DUPONT AG, 2011,	Access to orphan drugs despite poor quality of clinical evidence	49	3,77

Corresponding Author's Countries



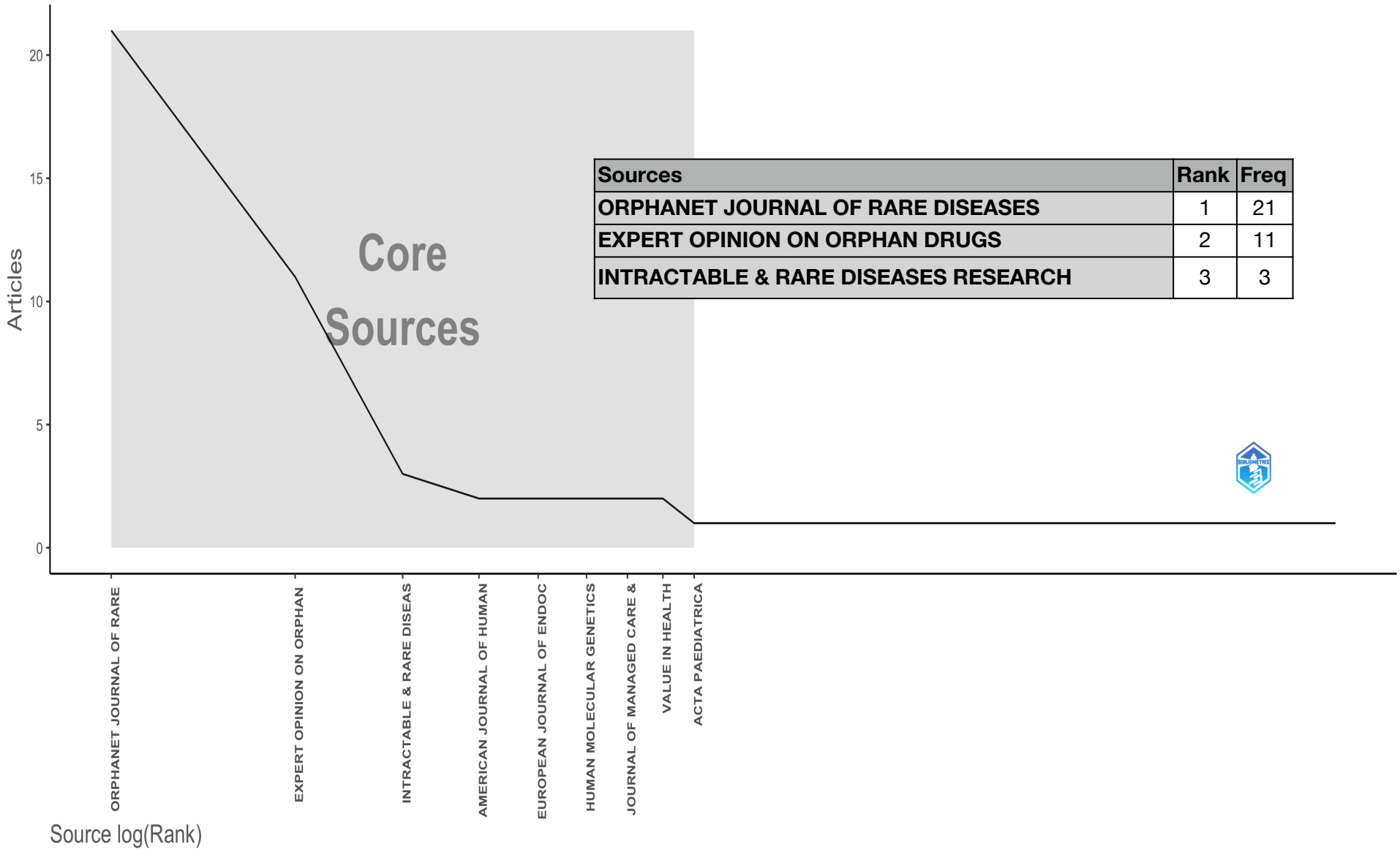
Country	Articles	SCP	MCP	Freq	MCP_Ratio
USA	44	24	20	0,319	0,455
CHINA	18	12	6	0,13	0,333
ITALY	12	8	4	0,087	0,333
GERMANY	10	4	6	0,072	0,6
UNITED KINGDOM	10	5	5	0,072	0,5
FRANCE	6	4	2	0,043	0,333
CANADA	4	2	2	0,029	0,5
SPAIN	4	3	1	0,029	0,25
AUSTRALIA	3	2	1	0,022	0,333
INDIA	3	3	0	0,022	0

SCP: Single Country Publications, MCP: Multiple Country Publications



Country	TC	Average Article Citations
USA	**824**	**18,70**
UNITED KINGDOM	435	43,50
CHINA	203	11,30
GERMANY	160	16,00
ITALY	91	7,60
FRANCE	81	13,50
NORWAY	79	39,50
SPAIN	69	17,20
HUNGARY	52	26,00
BELGIUM	49	49,00

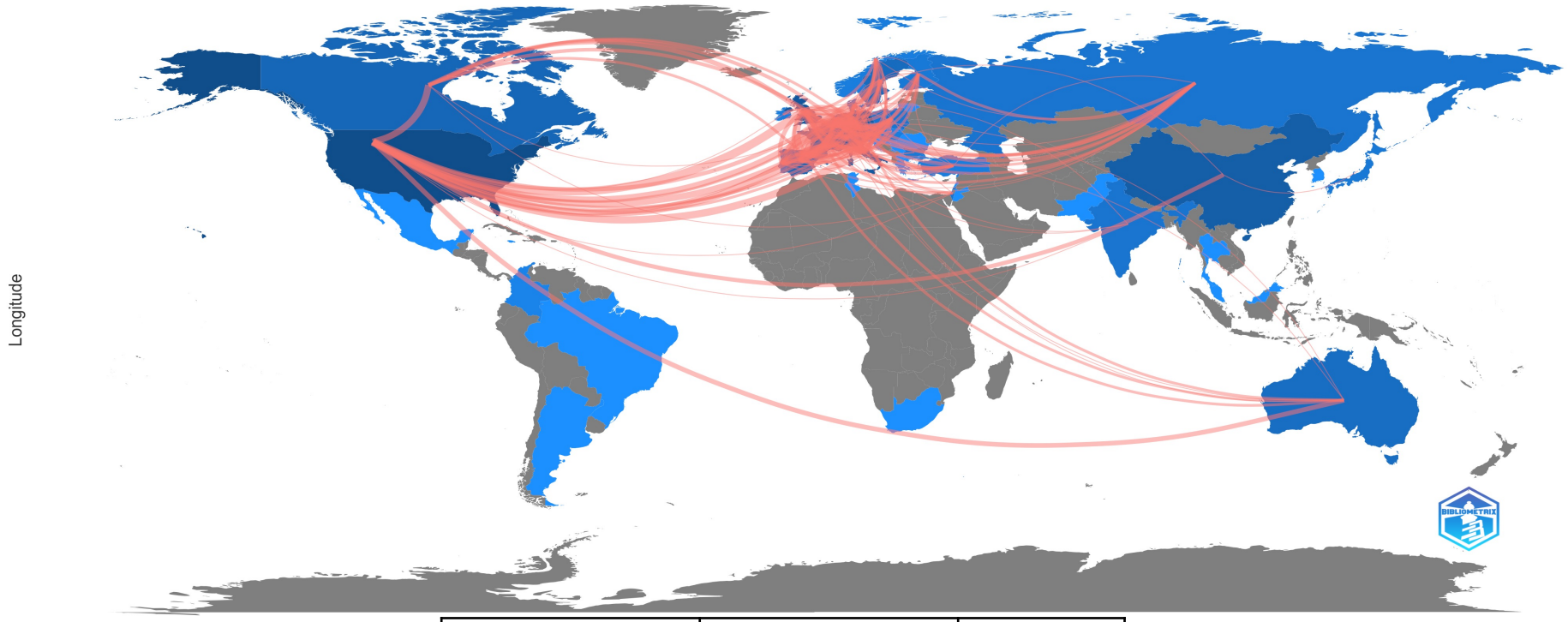
Core Sources by Bradford's Law



Sources	Rank	Freq
ORPHANET JOURNAL OF RARE DISEASES	1	21
EXPERT OPINION ON ORPHAN DRUGS	2	11
INTRACTABLE & RARE DISEASES RESEARCH	3	3

Co-occurrence network (Subject categories WoS)

Country Collaboration Map



From	To	Frequency
USA	**UNITED KINGDOM**	**16**
USA	**ITALY**	**13**
GERMANY	**ITALY**	**12**
ITALY	NETHERLANDS	12
UNITED KINGDOM	FRANCE	12
GERMANY	NETHERLANDS	10
GERMANY	UNITED KINGDOM	10
UNITED KINGDOM	ITALY	10
UNITED KINGDOM	NETHERLANDS	10
USA	CANADA	10

Terms	Frequency
orphan drugs	16
quality-of-life	14
rare diseases	14
management	12
mutations	9
access	7
challenges	7

Conclusion

The study reveals the status of the existing literature on the subject, information on the citations of the studies, the keywords used, the authors of the studies and their relationship networks, and analyzes such as cross-country cooperation.

The results of the analysis show that the concepts of burden of disease and rare diseases have started to be used together in the last ten years and the importance of the subject is gradually increasing.

Key Messages

- Rare diseases present significant challenges for policy decision makers in allocating resources.

- In this respect, it is important to present a picture of the current developments in the field to provide evidence to decision makers.

Thank You!

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