THINK NEURO 2023 SUMMER RESEARCH

BIBLIOMETRIC ANALYSIS OF THE TOP 100 ARTICLES ON RETT SYNDROME:

RESEARCH IS INCREASINGLY FOCUSING ON THE EFFECTS OF THE MUTATED MECPG2 GENE ON RETT GROWTH AND DEVELOPMENT

Parinitha Balaji*, Sydney Munion*, Stephanie Hung, Alisha Joseph, Raelle Tiong, Joleen Wong, Jolena Phat, Sarah Lai

INTRODUCTION

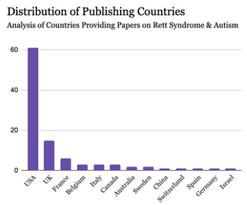
Cerebroatrophic hyperammonemia, also known as Rett Syndrome or RTT, is an intricate and extremely rare neurodevelopmental disorder caused by a mutation on the methyl CpG binding protein 2 (MeCP2) gene. The MeCP2 protein is vital to the regulation of gene activity and the depth of synaptic connections between neurons. Thus, a mutation in the gene that codes for the MeCP2 protein would result in a disorder that impairs nearly every aspect of life, predominantly in young girls.

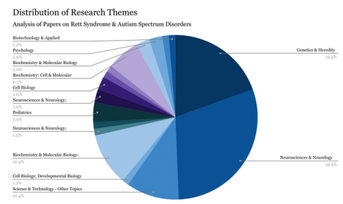
OBJECTIVE

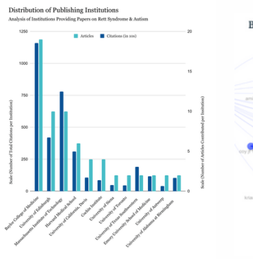
Current treatment for RTT can only improve patients' quality of life as there is no known cure for the disorder. Therefore, we conducted a comprehensive review of the existing studies and articles published on RTT to help the scientific community gain insight into important patterns and nuances in RTT research as well as aid in the development of novel treatments for RTT patients.

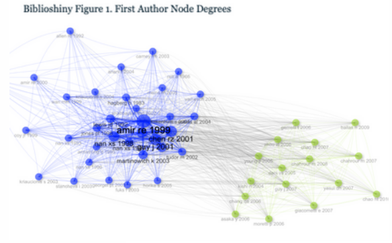
METHODS

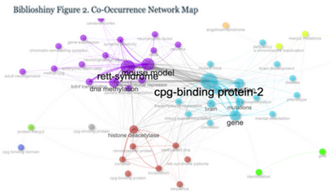
The database Web of Science was used to find the best academic articles on RTT. Once all desired articles were identified with the search term "Rett Syndrome", the top 200 most-cited articles were exported. These 200 articles were further manually reviewed to finalize a total of the top 100 articles on RTT. Inclusion criteria included: relevancy to RTT and no. of citations. The finalized 100 articles were slected for this bibliometric analysis study.

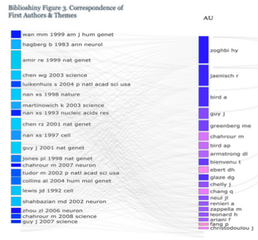












RESULTS & DISCUSSION

Publication years of the top RTT articles ranged from 1986 to 2016, with the United States being the largest contributing country. Baylor College of Medicine (TX) was the institution that contributed the largest amount of RTT articles. First authors Amir RE, Moretti P, and Hagberg B published the most-cited literature of this study. The 100 articles of this study covered a wide variety of themes from "Neuroscience & Neurology" to "Biochemistry & Molecular Biology", with "Genetics & Heredity" being the most published theme of interest. Frequencies of the keywords "mouse model", "DNA methylation", "histone deacetylase" and "mecp2" have increased dramatically over the decades, suggesting that these topics have received more attention in recent years. Within these top keywords, "mouse model" and "DNA methylation" are the largest common ideas (mouse models are specimens used to study patterns & mechanisms of human disease and to test for RTT treatments, while DNA methylation is an epigenetic mark pivotal for understanding the genetics behind RTT). These keywords signal a shift in the focus of research on RTT, indicating that researchers are now looking at the causes of the disorder, and using these findings to develop potential cures.

CONCLUSION

Based on the results of our network & bibliometric analyses, we conclude that research in RTT will continue to be of interest in the upcoming years, as researchers increasingly study the genetic basis and origins of disease (MeCPG-2). The latest RTT research heavily relies on mouse models, signifying a better understanding of RTT's mechanisms in patients. This new knowledge will be used to develop potential cures for the disorder and in the near future, will change the lives of young girls and boys struggling with RTT.

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