INPLASY

DDX3X SYNDROME BEHAVIORAL MANIFESTATIONS WITH PARTICULAR EMPHASIS ON PSYCHO-PATHOLOGICAL SYMPTOMS- A SYSTEMATIC REVIEW

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ADMINISTRATIVE INFORMATION

Support - Department of Clinical Psychology, Poznan University of Medical Sciences and Department and Clinic of Rheumatology, Rehabilitation and Internal Medicine, Poznań University of Medical Sciences.

Review Stage at time of this submission - Completed but not published.

Conflicts of interest - None declared.

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Amendments - This protocol was registered with the International Platform of Registered Systematic Review and Meta-Analysis Protocols (INPLASY) on 26 September 2023 and was last updated on 26 September 2023.

INTRODUCTION

Review question / Objective What behavioral psychopathological symptoms are present in individuals with DDX3X syndrome?

Condition being studied DDX3X syndrome is caused by a spontaneous mutation within a DDX3X gene at conception. Also this rare disease can be inherited; however, it does not happen on a frequent basis. Mutations in DDX3X are one of the most common causes of intellectual disability (ID), accounting for 1%-3% of unexplained ID in women. It has also been shown that this rare genetic disease is not only associated with ID but also with neurological symptoms, motor delays, behavioral problems, cardiac dysfunction, ophthalmic and gastrointestinal abnormalities. Notably, DDX3X variants have been linked with peculiar brain MRI abnormalities and brain tumor. Nowadays, the population of patients with DDX3X accounts for 848 known cases across 54 countries. 809 cases are females and 39 cases are males.

METHODS

Participant or population Cases of individuals with DDX3X syndrome reported in the literature.

Intervention No applicable.

Comparator No applicable.

Study designs to be included Case study, cohort study, research and review.

Eligibility criteria The following inclusion criteria were defined in this review: (1) the article type:

case study, cohort study, research and review; (2) date of publication: from 2015, justified by the fact that the first paper on DDX3X syndrome was published in 2015; (3) population: studies conducted on human participants with diagnosed DDX3X syndrome; (4) indication: manuscripts selected for this review were required to investigate the psycho-pathological behaviors; (5) language restrictions: only originally English language papers were taken into consideration. Exclusion criteria were as follows: (1) population: animal models; (2) indication: lack of identification of the psycho-pathological symptoms; (3) language restrictions: language other than English.

Information sources We conducted a systematic review by computerized searching of the following medical and public databases: PubMed, Medline Com-plete, Science Direct, Scopus, and Web of Science by using the following combinations of keywords and MeSH terms: "DDX3X syndrome"," intellectual disability", "neurodevelopmental disor-der*"," autism spectrum disorder*", "DDX3X ".

Main outcome(s) Two hundred seventy-two papers were found through the literature search. 172 of those were identified on PubMed, 32 on Medline Complete, 23 on Science Direct, 23 on Scopus, and 22 on Web of Science. A total of 105 papers were excluded because they were duplicates. From 167 screened records reviewers extracted 17 potentially relevant articles. Eventually, only 9 full-texts met the standards and were included in the review. One was a case study; Stefaniak et al, one was a review; Levy et al, one was a research; Tang et al, and six were cohort studies; Snijders et al, Wang et al, Beal et al, Lennox et al, Ng-Cordell et al, and Dai et al . Most of the studies were retrospective. The most common psychopathological behavior manifestations were demonstrated as ID or DD, communication/speech delay, autism spectrum disorder (ASD) or autistic-like behaviors, attention deficit hyperactivity disorder (ADHD), general anxiety disorder (GAD), self-injury behaviors (SIBs), sensory symptoms, and sleep dis-turbance. Moreover all of the 9 papers that evaluated the DDX3X concluded ID and/or DD and speech delay or communication problems as the most common features.

Quality assessment / Risk of bias analysis Two authors independently extracted pertinent information from each paper using a predefined and standardized data form in Microsoft Excel 365. In the event of any discrepancies between the researchers, discussions were conducted to achieve consensus. Detailed data regarding author(s) names, publication year, study design, sample size, as well as the type and prevalence of psycho-pathological symptoms of DDX3X syndrome and important findings were meticulously documented.

Strategy of data synthesis Due to the substantial methodological variations observed, particularly in measuring and reporting mental health and psy-cho-pathological outcomes, a meta-analysis of the gathered studies was not conducted. Instead, a narrative approach was employed to synthesize the findings, which were subsequently summarized and presented in tables. By adopting this approach, a comprehensive overview of the research outcomes was obtained, despite the methodological heterogeneity among the included studies. The narrative synthesis and tabular summaries provided a concise and in-formative representation of the findings, enabling a nuanced understanding of the collected data.

Subgroup analysis No applicable.

Sensitivity analysis No applicable.

Language restriction English.

Country(ies) involved Poland.

Keywords DDX3X Syndrome. Intellectual Disability. Developmental Disabilities. Autism Spectrum Disorder. Self-Injurious Behavior. Anxiety Disorders.

Contributions of each author

Author 1 - Urszula Stefaniak - A - Review Design; B - Data Collection; C - Data Analysis; D - Data Interpretation; E - Manuscript Preparation; F - Literature Search.
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Author 2 - Roksana Malak - A - Review Design; B - Data Interpretation; C - Manuscript Preparation; D- Content-related supervision.
Author 3 - Ada Kaczmarek - A - Review Design; B - Data Collection; C - Data Analysis; D - Data Interpretation; E - Manuscript Preparation; F - Literature Search.
Author 4 - Włodzimierz Samborski - A - Review Design; B- Review and editing; C- Content-related supervision.
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