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Structural Variations Identified in Patients with Autism Spectrum Disorder (ASD) in the Chinese Population: A Systematic Review of Case-Control Studies

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ADMINISTRATIVE INFORMATION

Support - The Nethersole School of Nursing, CUHK.

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 15 August 2024 and was last updated on 15 August 2024.

INTRODUCTION

Review question / Objective The current review aims to summarise the types of SVs reported in ASD among the Chinese population. It also discusses the genes potentially implicated in ASD. By examining the intersection of genetics and ethnicity, this review seeks to enhance the under-standing of ASD's complex etiology and pave the way for more effective interventions tailored to different populations.

Rationale The research of genetic variations in ASD was extensive. However, much of this was conducted in Caucasian populations, leaving a gap in the understanding of how ASD manifests in non-Caucasian populations, including the Chinese population, which contributes to 17% of the world's population.

Condition being studied Autism spectrum disorder (ASD) is a complex neurodevelopmental condition charac-terised by diverse symptoms that affect communication, behaviour, and social interactions. For a period, the prevalence of the disorder in the US was believed to be around 1 in 110 children, but it was found that it raised substantially to 1 out of 36 in a re-cent report by the Centers for Disease Control and Prevention (CDC). In China, according to an investigation in 2019, the prevalence of ASD children was 1%, which is comparable to the worldwide data, but the number was believed to be underestimated due to lack of awareness of the disorder and therefore under diagnosis.

METHODS

Search strategy Literature searches were completed in five Western databases, including PubMed, EMBASE, Ovid Medline, Ovid Nursing, and CINAHL, and four Chinese databases, including CNKI, Wanfang, Sinomed, and VIP. Searches in electronic databases identified studies published from inception to March 2024.

Participant or population Chinese ASD cohort recognised using any standard diagnostic criteria

(Diagnostic and Statistical Manual of Mental Disorders or other clinical diagnosis).

Intervention Not applicable.

Comparator Not applicable.

Study designs to be included Case-control study.

Eligibility criteria Inclusion criteria included (i) human case-control cohort studies; (ii) ASD cohort recognised using any standard diagnostic criteria (Diagnostic and Statistical Manual of Mental Disorders or other clinical diagnosis).
Exclusion criteria included: (i) studies not having a control group; (ii) non-English/Chinese publication; (iii) in vitro and/or animal studies; and (iv) abstracts, reviews, and study protocol.

Information sources PubMed, EMBASE, Ovid Medline, Ovid Nursing, and CINAHL, and four Chinese data-bases, including CNKI, Wanfang, Sinomed, and VIP.

Main outcome(s) The types of SVs reported in ASD among the Chinese population.

Quality assessment / Risk of bias analysis The assessment of study quality was carried out using 8 specific items derived from the Strengthening the Reporting of Genetic Association Studies (STREGA) checklist for cohort studies.

Strategy of data synthesis Articles that satisfied the inclusion criteria were subsequently evaluated in full text and were assessed for their suitability for data extraction and analysis. The screening and data extraction of the articles were conducted by two authors independently. Any discrepancies regarding inclusion were resolved through discussion and consensus among the researchers involved.The screening and data extraction of the articles were conducted by two authors independently.

Subgroup analysis Not Applicable.

Sensitivity analysis Not Applicable.

Language restriction Studies with non-English or Chinese publication will not be included.

Country(ies) involved Hong Kong SAR.

Keywords Keywords used for searching included “Autism OR Autistic” AND “Structural varia* OR Transposition* OR Transposon* OR Retrotransposition* OR Retrotransposon* OR.

Contributions of each author

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