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DOWN SYNDROME: ETIOLOGY, DIAGNOSIS, AND PREVENTION

J.Tashaliyeva
Central Asian Medical University
Clinical residiency

Abstract Down syndrome (DS) is the most common chromosomal disorder caused by trisomy 21. It is associated with intellectual disability, characteristic facial features, congenital anomalies, and increased risk of medical complications. This review presents an evidence-based overview of the etiology, diagnostic approaches, and preventive strategies for Down syndrome, in accordance with standards of American medical journals.
Keywords: Down syndrome; trisomy 21; prenatal screening; genetic diagnosis; prevention

Introduction

Down syndrome affects approximately 1 in 700 live births and represents a major cause of developmental disability worldwide. First described by John Langdon Down in 1866, the condition has since been extensively studied. Advances in molecular genetics and prenatal diagnostics have significantly improved early detection and management.

Etiology and Pathogenesis

In nearly 95% of cases, Down syndrome results from meiotic nondisjunction leading to complete trisomy 21. Robertsonian translocation accounts for approximately 3–4% of cases, while mosaicism occurs in 1–2%. Advanced maternal age remains the most significant risk factor.[1,2,3]

Clinical Features

Individuals with Down syndrome commonly present with hypotonia, brachycephaly, epicanthic folds, and a single transverse palmar crease. Frequent comorbidities include congenital heart disease, gastrointestinal anomalies, endocrine disorders, hearing and vision impairment, and increased susceptibility to leukemia and early-onset Alzheimer disease.[4]

Diagnostic Approaches

Prenatal screening includes first-trimester combined screening, second-trimester biochemical tests, and non-invasive prenatal testing (NIPT). Definitive diagnosis is achieved through invasive procedures such as amniocentesis or chorionic villus sampling, followed by karyotyping, FISH, or chromosomal microarray analysis. Postnatal diagnosis relies on cytogenetic analysis of peripheral blood.[5]

Prevention and Prophylaxis

While Down syndrome cannot be completely prevented, risk reduction strategies include genetic counseling, prenatal screening programs, and informed reproductive decision-making. Public health education and early intervention programs play a crucial role in improving outcomes.[6]

Management and Prognosis

Multidisciplinary care significantly improves life expectancy and quality of life. With modern medical support, individuals with Down syndrome can now live beyond 60 years.

Ethical Considerations

Prenatal diagnosis raises ethical issues regarding informed consent and reproductive autonomy. Physicians must provide balanced, non-directive counseling consistent with ethical guidelines.[7]

Conclusion

Down syndrome remains a significant genetic condition with important medical and social implications. Continued advances in diagnostic technologies and preventive strategies are essential for improving patient outcomes.

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