OPEN ACCESS Freely available online

Journal of Down Syndrome & Chromosome Abnormalities

Short Communication

The Genetic Mechanisms and Therapeutic Interventions of Patau Syndrome a Chromosomal Syndrome

Niveditha Thomas*

Department of Genetics, University of Osmania, Hyderabad, Telangana, India

DESCRIPTION

Patau syndrome, also known as trisomy 13, is a chromosomal disorder characterized by the presence of an extra copy of chromosome 13. This additional genetic material disrupts normal development and leads to a wide range of physical and intellectual disabilities. Named after the physician Klaus Patau who first described the syndrome in 1960, Patau syndrome is a rare condition, occurring in approximately 1 in 5,000 to 1 in 20,000 live births.

Chromosomal aberration

The human body typically has 23 pairs of chromosomes, and any deviation from this normal chromosomal count can result in genetic disorders. In the case of Patau syndrome, individuals have three copies of chromosome 13 instead of the usual two. This extra genetic material can lead to a myriad of abnormalities affecting various organ systems.

Clinical manifestations

Patau syndrome is associated with a range of physical and intellectual disabilities, and the severity of symptoms can vary widely among affected individuals. Common physical features include facial abnormalities such as cleft lip and palate, small or malformed eyes, and extra fingers or toes. Additionally, individuals with Patau syndrome may experience heart defects, kidney malformations, and problems with other internal organs [1].

Intellectual impairments

Cognitive development is significantly affected in individuals with Patau syndrome. Intellectual disabilities are common, and most affected individuals experience developmental delays. Severe learning difficulties and challenges in acquiring motor skills are typical, requiring specialized care and support throughout their lives [2].

Survival rates and prognosis

The prognosis for individuals with Patau syndrome is often poor, and many affected pregnancies end in miscarriage. Those who survive birth face a shortened life expectancy, with the majority not living beyond the first year of life. The severity of associated health issues contributes to the high mortality rate, although some individuals may live into their teens or twenties with intensive medical care [3].

Diagnosis

Patau syndrome can be diagnosed through prenatal screening or diagnostic testing. Non-invasive methods, such as maternal serum screening and ultrasound, can indicate an increased risk of chromosomal abnormalities, prompting further testing like amniocentesis or chorionic villus sampling for a definitive diagnosis.

Impact on families

Receiving a diagnosis of Patau syndrome can be emotionally challenging for families. The realization that their child will face significant health challenges and may have a limited life expectancy can be overwhelming. Families often require extensive support, both emotionally and practically, as they navigate the complex medical and caregiving needs of their child [4].

Management and treatment

There is no cure for Patau syndrome, and treatment is focused on managing the individual symptoms and providing supportive care. This may involve surgeries to correct physical abnormalities, interventions to address cardiac or renal issues, and early intervention programs to support developmental delays. A multidisciplinary approach involving medical professionals, therapists, and educators is essential to provide comprehensive care.

Correspondence to: Niveditha Thomas, Department of Genetics, University of Osmania, Hyderabad, Telangana, India, Email: niveditha_thomas@usedu.com

Received: 27-Nov-2023, Manuscript No. JDSCA-24-29293; **Editor assigned:** 30-Nov-2023, PreQC No. JDSCA-24-29293 (PQ); **Reviewed:** 14-Dec-2023, QC No. JDSCA-24-29293; **Revised:** 21-Dec-2023, Manuscript No. JDSCA-24-29293 (R); **Published:** 28-Dec-2023, DOI: 10.35248/2472-1115.23.9.245

Citation: Thomas N (2023) The Genetic Mechanisms and Therapeutic Interventions of Patau Syndrome a Chromosomal Syndrome. J Down Syndr Chr Abnorm. 9:245.

CONCLUSION

Patau syndrome remains a rare and challenging genetic disorder that significantly impacts affected individuals and their families. While advances in medical care have improved the management of symptoms and increased survival rates, the prognosis for individuals with Patau syndrome remains guarded. Research into understanding the underlying genetic mechanisms and potential therapeutic interventions continues, offering hope for improved outcomes in the future. In the meantime, support networks, early intervention, and compassionate care play crucial roles in enhancing the quality of life for individuals with Patau syndrome and their families.

REFERENCES

1. Tal R, Schwartz Y, Zolotushko J, Lorber A. Trisomy 13 (Patau syndrome) with tetralogy of Fallot—To treat or not to treat?. Int J Cardiol. 2014;172(1):175-176.
2. Fogu G, Maserati E, Cambosu F, Moro MA, Poddie F, Soro G, et al. Patau syndrome with long survival in a case of unusual mosaic trisomy 13. Eur J Med Genet. 2008;51(4):303-314.
3. Khan U, Hussain A, Usman M, ul Abiddin Z. An infant with patau syndrome associated with congenital heart defects. Ann Med Surg (Lond). 2022;80:104100.
4. Kamal M, Varghese D, Bhagde J, Singariya G, Simon AM, Singh A. Anesthesia in a child operated for cleft lip associated with Patau's syndrome. Braz J Anesthesiol. 2018;68:197-199.