

The Significant Challenges of Patau Syndrome, its Complexities and Clinical Features on Individuals

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DESCRIPTION

Patau syndrome, also known as trisomy 13, stands as one of the less common yet profoundly impactful genetic disorders affecting human beings. Named after the renowned scientist Klaus Patau, who initially described it in 1960, this syndrome stems from a chromosomal abnormality leading to a myriad of physical and intellectual disabilities. While medical advancements have improved our understanding and management of genetic disorders, Patau syndrome poses significant challenges due to its complexity and the profound effects it has on individuals and families.

Genetic basis and occurrence

Patau syndrome is primarily caused by the presence of an additional copy of chromosome 13 in each cell of the body, instead of the usual pair. This additional chromosome disrupts normal development, resulting in the wide array of physical and intellectual disabilities associated with the syndrome. The condition typically arises from errors in cell division during the formation of reproductive cells (eggs and sperm) or in early fetal development.

The occurrence of Patau syndrome is relatively rare compared to other chromosomal abnormalities, with an estimated incidence of about 1 in 5,000 to 1 in 16,000 live births worldwide. However, the syndrome is more common among pregnancies that end in miscarriage with some studies suggesting that up to 1 in 100 miscarriages may involve Patau syndrome.

Clinical features

Individuals with Patau syndrome often exhibit a distinctive set of physical features that may include:

Cleft lip and palate: A significant number of affected individuals are born with a cleft lip and/or palate, which can contribute to feeding difficulties and require surgical intervention.

Microcephaly: Many infants with Patau syndrome have smaller than average head size due to abnormal brain development.

Polydactyly: Extra fingers or toes, known as polydactyly are common in individuals with Patau syndrome.

Heart defects: The majority of affected individuals have congenital heart defects, which can range from mild to severe and may require surgical correction.

Eye abnormalities: Eye problems such as small eyes (microphthalmia) or defects in the iris (coloboma) are frequently observed.

Neurological abnormalities: Individuals with Patau syndrome often experience severe intellectual disability, seizures and other neurological issues.

Renal abnormalities: Kidney abnormalities are common and can contribute to complications such as urinary tract infections and kidney failure.

It is essential to note that the severity and combination of symptoms can vary widely among individuals with Patau syndrome. While some may exhibit only a few of the characteristic features, others may experience more extensive medical and developmental challenges.

Diagnosis and management

Prenatal screening and diagnostic tests are crucial for identifying Patau syndrome early in pregnancy, allowing parents to make informed decisions about their pregnancy and access appropriate medical care. Screening tests such as ultrasound and maternal serum screening can indicate an increased risk of chromosomal abnormalities, prompting further diagnostic testing such as Chorionic Villus Sampling (CVS) or amniocentesis to confirm the diagnosis.

Once diagnosed, the management of Patau syndrome focuses on providing supportive care to address the specific needs of the individual. This may involve a multidisciplinary approach involving pediatricians, geneticists, cardiologists, neurologists and other specialists to address the diverse medical and developmental challenges associated with the syndrome.

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Prognosis and outlook

The prognosis for individuals with Patau syndrome varies depending on the severity of their symptoms and the presence of associated medical complications. Unfortunately, the majority of affected individuals do not survive beyond the first year of life, with many succumbing to complications related to congenital heart defects or other severe medical issues.

For those who do survive infancy, life expectancy remains significantly reduced, and ongoing medical care and support are often necessary to manage the complex health needs associated with the syndrome. Despite these challenges, many families of individuals with Patau syndrome find meaning and fulfillment in caring for their loved ones and advocating for greater awareness and support for individuals with rare genetic disorders.

Patau syndrome represents a complex and challenging genetic disorder with significant implications for affected individuals and their families. While advances in medical technology and understanding have improved our ability to diagnose and manage the syndrome, much remains to be learned about its underlying mechanisms and optimal treatment approaches.

Efforts to enhance prenatal screening, expand access to genetic counseling and supportive services and promote research into the underlying causes of Patau syndrome are essential for improving outcomes and quality of life for affected individuals and their families. Through increased awareness, education and support, we can strive to create a more inclusive and compassionate society that embraces the diversity of human genetic variation and ensures that individuals with Patau syndrome and other genetic disorders receive the care and support they need to thrive to the best of their abilities.

Patau syndrome serves as a poignant reminder of the complexities of human genetics and the profound impact that chromosomal abnormalities can have on individuals and families. By fostering collaboration among healthcare professionals, policymakers and advocacy groups we can work towards improving the lives of those affected by Patau syndrome and other rare genetic disorders, offering hope, dignity and support in the face of adversity.