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Opinion Article

Genetic Counselling for Families Affected by Patau Syndrome

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DESCRIPTION

Patau syndrome, also known as Trisomy 13, is a rare and serious genetic disorder caused by the presence of an extra copy of chromosome 13 in the cells of the body. This condition leads to severe developmental and physical abnormalities. The syndrome is named after the Swiss geneticist Klaus Patau, who first described it in 1960. Though the condition is extremely rare, with an incidence of approximately 1 in 10,000 live births, it remains one of the most well-known chromosomal disorders.

Patau syndrome is primarily caused by a chromosomal abnormality known as trisomy, where instead of the usual two copies of chromosome 13, an individual has three copies in each of their cells. This extra genetic material disrupts normal development, leading to a variety of health complications.

In most cases, the extra chromosome is present in all the body's cells, but there are instances where some cells have the normal two copies of chromosome 13 and others have three copies. This type of mosaic trisomy 13 occurs less frequently and typically results in a slightly less severe form of the disorder.

The cause of trisomy 13 is typically random and is not usually inherited. In some rare instances, a parent may carry a chromosomal rearrangement known as a balanced translocation, which can increase the risk of passing on trisomy 13. The condition is more commonly observed in pregnancies of older mothers, though it can affect women of all ages.

Symptoms and characteristics

The physical and intellectual effects of Patau syndrome are severe. Most infants born with the disorder experience extream developmental delays and multiple physical abnormalities. Common features of Patau syndrome include:

Craniofacial abnormalities: These may include a cleft lip and palate, low-set ears, small eyes (microphthalmia) or the absence of one or both eyes (anophthalmia).

Heart defects: Many children with Patau syndrome are born with congenital heart defects, including holes in the heart or other complex structural issues.

Neural tube defects: These may lead to abnormalities in the brain and spine, including an open spine (spina bifida) or other malformations of the central nervous system.

Polydactyly: Extra fingers or toes are often present.

Growth problems: Affected children often have a low birth weight and experience slow growth and development in the early stages of life.

Severe intellectual disability: Children with Patau syndrome typically have extream developmental and intellectual disabilities, with little to no cognitive function.

Other common complications include kidney problems, gastrointestinal issues and an increased susceptibility to infections. Many infants with Patau syndrome also experience seizures, respiratory difficulties and difficulty feeding.

Diagnosis

Patau syndrome is typically diagnosed before or shortly after birth. In many cases, ultrasound imaging during pregnancy can reveal physical abnormalities indicative of the syndrome. If suspected, a prenatal genetic test such as amniocentesis or Chorionic Villus Sampling (CVS) can be performed to confirm the presence of the extra chromosome 13.

After birth, further genetic testing, such as chromosomal analysis or karyotyping, can definitively diagnose the condition by identifying the additional chromosome 13.

Prognosis and life expectancy

The prognosis for individuals with Patau syndrome is typically very poor. Many infants with this condition die within the first days or weeks of life due to complications such as heart failure, infections, or brain abnormalities. Those who survive beyond infancy often face a variety of health challenges and require intensive care and support.

However, in rare cases, individuals with mosaic trisomy 13 may live longer and have a slightly better quality of life, although they will still experience significant developmental delays and health complications.

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CONCLUSION

Patau syndrome is a devastating genetic disorder that results from an extra chromosome 13. Although the condition is rare, its impact on affected individuals and their families is completely. Due to the severity of the developmental and physical challenges posed by the disorder, early diagnosis and comprehensive care are important. Families affected by Patau syndrome often benefit from genetic counseling to understand the condition and make informed decisions about care and management options. Despite the challenges, ongoing medical research continues to improve the understanding of Patau syndrome and the ways to support affected individuals and families.