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Perspective

Pathophysiology, Diagnosis and Treatment Involved in Patau Syndrome

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DESCRIPTION

Patau syndrome, also called trisomy 13, is a clinical syndrome that occurs when all or some cells of the body contain an extra copy of chromosome 13. It is characterized by cleft lip, cleft palate, cerebral defects, anophthalmia, simian creases, polydactyly, trigger thumbs, and capillary hemangiomata.

This can happen if each cell has an extra copy of chromosome 13 (a condition known as trisomy 13 or trisomy D or T13), if each cell has an extra partial copy of the chromosome, or if there are 2 distinct lines of cells, one with regular chromosome 13 and the other with an extra copy, is an chromosome-mosaic Patau syndrome.The non disjunction of chromosomes during meiosis results in full trisomy 13. (The mosaic form is caused by nondisjunction during mitosis).

Microcephaly and intrauterine growth restriction are common in infants with Patau syndrome. The most common midline facial deformities are cyclopia, cleft lip, and cleft palate. A sloping forehead, small, deformed ears, anophthalmia or microphthalmia, micrognathia, and pre-auricular tags are among the features of the face. Midline is also common for central nervous system defects, with alobar holoprosencephaly being the most prevalent problem. Postaxial polydactyly, congenital talipes equinovarus, and rocker-bottom feet are examples of common extremity abnormalities. A ventricular septal defect, an atrial septal defect, a tetralogy of Fallot, an atrioventricular septal defect, and a double outlet right ventricle are among the heart conditions seen in Patau syndrome.

Additional organ systems affected by abnormalities include the lungs, liver, kidneys, genitourinary tract, digestive tract, and pancreas. Defects in these organ systems that occur in greater than 50% of patients with Patau syndrome include cryptorchidism, hypospadias, labia minora hypoplasia, and bicornuate uterus. Abnormalities in these organ systems occurring in less than 50% of patients with Patau syndrome include omphalocele, incomplete rotation of the colon, Meckel diverticulum, polycystic kidney, hydronephrosis, and horseshoe kidney. Patients surviving past infancy have a severe psychomotor disorder, failure to thrive, intellectual disability, and seizures.

Pathophysiology

An extra copy of chromosome 13 causes the defects in Patau syndrome. Advanced maternal age is a risk factor for this pathology because of the increased frequency of nondisjunction in meiosis. This extra copy of chromosome 13 disrupts normal embryonic development and leads to multiple defects.

Diagnosis

Prenatal chorionic villi sampling, amniocentesis, or foetal free DNA testing can be used to diagnose Patau syndrome. These diagnostic techniques identify trisomy 13. The common Patau syndrome malformations such as holoprosencephaly or other central nervous system abnormalities, facial variances, skeletal abnormalities, renal or cardiac problems, and growth restriction, can also be found with the aid of prenatal ultrasound. The Patau syndrome abnormalities can be found *via* prenatal ultrasonography after 17 weeks of pregnancy. It is important to confirm abnormal findings using a cytogenetic analysis of foetal cells. Tissue microarray has improved the ability to determine genetic changes in foetal mortality.

Treatment

Newborns with Patau syndrome may require post-delivery oxygenation and breathing; due to facial deformities, this may necessitate intubation or tracheostomy. To treat common cardiac problems in patients with cardiac defects, cardiac surgery may be necessary. For common problems, further procedures such herniorrhaphy, cleft lip surgery, feeding tube insertion, or corrective orthopaedic surgeries can be necessary. Other therapies include the use of hearing aids, prophylactic antibiotics for urinary tract infections, specific nutritional feeds, and seizure prophylaxis. Despite rigorous management, the median survival time in the most recent patient cohorts is only 733 days.

CONCLUSION

Patau syndrome is a disease with variable expression and is characterized by a pattern of abnormal prenatal development characterized by facial dysmorphia, polydactyly and severe birthdefects (heart, brain) that generate an increased in utero and perinatal mortality.

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