Patau Syndrome

Patau syndrome, also called trisomy 13, is a genetic abnormality caused by an additional chromosome 13. The most common cause is nondisjunction of chromosome 13 during meiosis. The extra chromosome 13 disrupts normal development and as a result, many infants do not survive beyond the first few weeks of life. Those that survive are commonly severely mentally retarded and have clinical features of microcephaly, holoprosencephaly, microphthalmia, polydactyly, cleft lip and palate, and rocker bottom feet. Patients also have renal defects including cystic kidneys and cardiac defects like ventricular septal defects.



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Pathophysiology

Trisomy 13
Tricycle Friday-the-13th-guy
Patau syndrome is a genetic abnormality caused by an additional chromosome 13.

Signs and Symptoms

Severe Intellectual Disability
Severed Book Covered in Tar
Children with Patau syndrome display significantly impaired cognitive functioning and deficits in two or more adaptive behaviors.

Microcephaly
Small-head
A neurodevelopmental disorder in which the head circumference is more than two standard deviations smaller than the average circumference for the person's age and gender. It is a common clinical feature of Patau syndrome.

Holoprosencephaly
Halo-brain
Holoprosencephaly is caused by a failure of the forebrain to divide properly into two hemispheres and is commonly observed in Patau syndrome.

Microphthalmia
Small-pea-eyes
Microphthalmia literally means "small eye" and is a clinical feature of Patau's syndrome.

Polydactyly
Poly-with-extra-fingers
A congenital physical anomaly where people have extra fingers or toes.

Cleft Lip/Cleft Palate
Cleft-cracked Plate
A congenital deformity caused by abnormal facial development characterized by a fissure in lip and/or palate.

Rocker Bottom Feet
Rocking-chair with Rocker bottom feet
Characterized by a prominent calcaneus and a rounded bottom of the foot resembling the bottom of a rocking chair. Rocker bottom feet are seen in both Patau syndrome and Edwards syndrome.

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Omphalocele

O-full-of-eels

An omphalocele is a defect of the midline abdominal wall. The defect is covered by a three-layer membranous sac consisting of amnion, Wharton's jelly, and peritoneum.

Cystic Kidneys

Cystic Kidney

Renal defects including cystic kidneys are common in Patau syndrome.

Ventricular Septal Defect (VSD)

Vase-hole-heart

Ventricular septal defect is a congenital heart defect that enables blood to flow between the left and right ventricle. VSDs are common in children with Patau syndrome.

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