

Trisomy 13 (Patau Syndrome)

Condition Description

Trisomy 13, also known as Patau Syndrome, is a rare genetic disorder that causes multiple abnormalities due to the presence of an extra copy of chromosome 13 in the cells of the body. The cause is unknown; however there is a significant association with increased maternal age. This condition is often fatal.

Trisomy 13 is usually not inherited and is a random occurrence during the formation of eggs and sperm in healthy parents.⁵ Trisomy 13 may be diagnosed prenatally or shortly after birth through genetic testing.

There are two variations of trisomy 13 that may occur in very rare situations. In mosaic trisomy 13, the extra copy of chromosome 13 is present in only some of the cells. The symptoms related to this variation will depend on the number of cells that have the extra chromosome and may be milder than those of full trisomy 13. There is also partial trisomy where only a part of the extra chromosome 13 is in each of the cells. The symptoms related to this variation may also be different than those found in full trisomy 13.³

Prevalence

Trisomy 13 is seen in approximately 1 in 10,000-16,000 births.^{4,5} There are no significant racial, ethnic or socioeconomic differences in frequency. The recurrence rate is typically very low (1%) and increases with maternal age.¹

Ninety-five percent of fetuses affected by trisomy 13 are spontaneously aborted.² The median survival rate for infants born with trisomy 13 is under three days with eighty-two percent dying within the first month and ninety-five percent

within the first six months.^{1,2,5} Only one in twenty children with trisomy 13 will survive to six months of age. Although, some children survive into their teens and seem to do better than expected.¹

Associated Conditions

Severe neurologic impairment is consistent in all children born with trisomy 13.¹ Other associated conditions may include cleft lip and/or palate, neural tube defects, congenital heart disease, microcephaly (small head size), scalp defects, hernias, and polydactyly (extra fingers and toes).

Parents and family members will most likely need to make many difficult decisions after their child with trisomy 13 is born, such as identifying their preferences for extraordinary life-prolonging measures.

Children who survive the neonatal period may experience additional complications such as deafness, heart failure, seizures, breathing difficulty and vision problems.⁵ Surgical interventions are usually withheld for the first few months due to the high mortality rate.

Developmental Concerns and Outcomes

Children with trisomy 13 will express significant developmental delays when compared to typically-developing peers. Even though the majority of these children do not survive the first six months, it is still critically important for early intervention services to be provided in order to maximize the child's potential through increased stimulation and interaction.⁵ Those that do survive will have severe developmental delays, seizures and growth problems.



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Trisomy 13 (Patau Syndrome) – page 2

For More Information

There are several support organizations for families and physicians available on-line. Genetic testing and counseling can help families understand the condition and the likelihood of having another child with the condition.

This fact sheet is for use in determining eligibility for early intervention services in Minnesota. For more information contact:

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5. Genetic and Rare Diseases Information Center (GARD). National Institutes of Health, Office of Rare Diseases.
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