

Trisomy 13

➤ **What is Trisomy 13?**

- Trisomy 13, also called Patau Syndrome is a chromosomal condition associated with severe intellectual disability and physical abnormalities in many parts of the body. Trisomy 13 occurs in about 1 in 16,000 newborns. Although women of any age can have a child with Trisomy 13, the chance of having a child with this condition increases as women get older. Due to the presence of several life-threatening medical problems, many infants with Trisomy 13 die within the first days or weeks of life. Only five percent to 10 percent of children with this condition live past their first year of life.

➤ **What are the signs/symptoms?**

- Symptoms include:
 - Cleft lip or palate
 - Clenched hands (with outer fingers on top of the inner fingers)
 - Close-set eyes—eyes may actually fuse together into one
 - Decreased muscle tone
 - Extra fingers or toes (polydactyly)
 - Hernias: umbilical hernia, inguinal hernia
 - Hole, split, or cleft in the iris (coloboma)
 - Low-set ears
 - Intellectual disability, severe
 - Scalp defects (missing skin)
 - Seizures
 - Single palmar crease
 - Skeletal (limb) abnormalities
 - Small eyes
 - Small head (microcephaly)
 - Small lower jaw (micrognathia)
 - Undescended testicle (cryptorchidism)

➤ **What is Trisomy 13 a result of?**

- Trisomy 13 occurs when extra DNA from chromosome 13 appears in some or all of the body's cells. The extra material interferes with normal development. Trisomy 13 occurs in about 1 out of every 10,000 newborns. Most cases are not passed down through families. Instead, the events that lead to Trisomy 13 occur in either the sperm or the egg that forms the fetus.

➤ **What are the complications of Trisomy 13?**

- More than 90% of children with Trisomy 13 die within their first year of life. Complications begin almost immediately. Most infants with Trisomy 13 have congenital heart disease. Complications may include:
 - Breathing difficulty or lack of breathing (apnea)
 - Deafness
 - Feeding problems
 - Heart failure
 - Seizures
 - Vision problems

