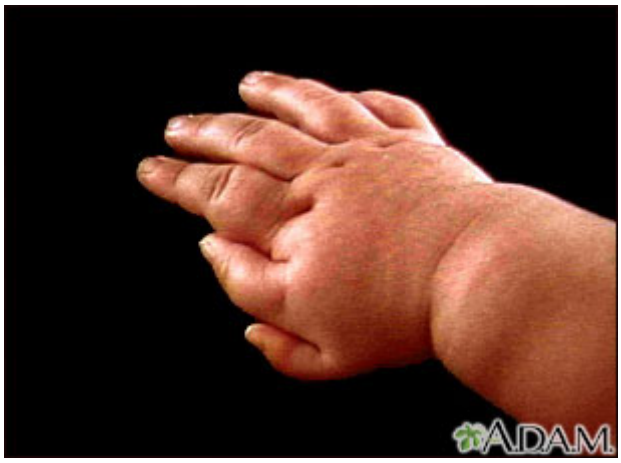
Trisomy 13

Definition

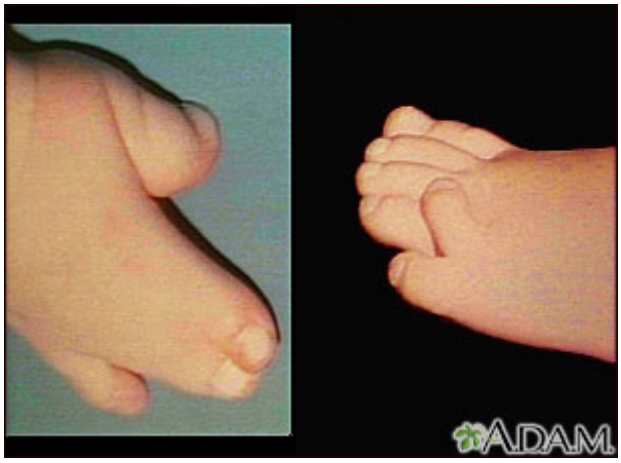


Trisomy 13 (also called Patau syndrome) is a genetic disorder in which a person has 3 copies of genetic material from chromosome 13, instead of the usual 2 copies. Rarely, the extra material may be attached to another chromosome (translocation).

Alternative Names

Patau syndrome

Causes



Trisomy 13 occurs when extra DNA from chromosome 13 appears in some or all of the body's cells.

- Trisomy 13: the presence of an extra (third) chromosome 13 in all of the cells.
- Mosaic trisomy: the presence of an extra chromosome 13 in some of the cells.
- Partial trisomy: the presence of a part of an extra chromosome 13 in the 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 (inherited). Instead, the events that lead to trisomy 13 occur in either the sperm or the egg that forms the fetus.

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)

Exams and Tests

The infant may have a single umbilical artery at birth. There are often signs of congenital heart disease, such as:

- Abnormal placement of the heart toward the right side of the chest instead of the left
- Atrial septal defect
- Patent ductus arteriosus
- Ventricular septal defect

Gastrointestinal x-rays or ultrasound may show rotation of the internal organs.

MRI or CT scans of the head may reveal a problem with the structure of the brain. The problem is called holoprosencephaly. It is the joining together of the 2 sides of the brain.

Chromosome studies show trisomy 13, trisomy 13 mosaicism, or partial trisomy.

Treatment

There is no specific treatment for trisomy 13. Treatment varies from child to child and depends on the specific symptoms.

Support Groups

Support groups for trisomy 13 include:

- Support Organization for Trisomy 18, 13 and Related Disorders (SOFT): trisomy.org
- Hope for Trisomy 13 and 18: www.hopefortrisomy13and18.org

Outlook (Prognosis)

More than 90% of children with trisomy 13 die in the first year.

Possible Complications

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

When to Contact a Medical Professional

Call your health care provider if you have had a child with trisomy 13 and you plan to have another child. Genetic counseling can help families understand the condition, the risk of inheriting it, and how to care for the person.

Prevention

Trisomy 13 can be diagnosed before birth by amniocentesis with chromosome studies of the amniotic cells.

Parents of infants with trisomy 13 that is caused by a translocation should have genetic testing and counseling. This may help them avoid having another child with the condition.

References

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Review Date: 8/6/2017
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