

Trisomy 13

Description

Trisomy 13, also called Patau syndrome, is a chromosomal condition associated with severe intellectual disability and physical abnormalities in many parts of the body. Individuals with trisomy 13 often have heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), extra fingers or toes, an opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (a cleft palate), and weak muscle tone (hypotonia). Due to the presence of several life-threatening medical problems, many infants with trisomy 13 die within their first days or weeks of life. Only five percent to 10 percent of children with this condition live past their first year.

Frequency

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 a woman gets older.

Causes

Most cases of trisomy 13 result from having three copies of chromosome 13 in each cell in the body instead of the usual two copies. The extra genetic material disrupts the normal course of development, causing the characteristic features of trisomy 13.

Trisomy 13 can also occur when chromosome 13 becomes attached (translocated) to another chromosome during the formation of reproductive cells (eggs and sperm) or very early in fetal development. Affected people have two normal copies of chromosome 13, plus an extra copy of chromosome 13 attached to another chromosome. In rare cases, only part of chromosome 13 is present in three copies. The physical signs and symptoms in these cases may be different than those found in full trisomy 13.

A small percentage of people with trisomy 13 have an extra copy of chromosome 13 in only some of the body's cells. In these people, the condition is called mosaic trisomy 13. The severity of mosaic trisomy 13 depends on the type and number of cells that have the extra chromosome. The physical features of mosaic trisomy 13 are often milder than those of full trisomy 13.

Learn more about the chromosome associated with Trisomy 13

- chromosome 13

Inheritance

Most cases of trisomy 13 are not inherited and result from random events during the formation of eggs and sperm in healthy parents. An error in cell division called nondisjunction results in a reproductive cell with an abnormal number of chromosomes. For example, an egg or sperm cell may gain an extra copy of chromosome 13. If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra chromosome 13 in each cell of the body.

Translocation trisomy 13 can be inherited. An unaffected person can carry a rearrangement of genetic material between chromosome 13 and another chromosome. These rearrangements are called balanced translocations because there is no extra material from chromosome 13. A person with a balanced translocation involving chromosome 13 has an increased chance of passing extra material from chromosome 13 to their children.

Other Names for This Condition

- Bartholin-Patau syndrome
- Complete trisomy 13 syndrome
- Patau syndrome
- Patau's syndrome
- Trisomy 13 syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Complete trisomy 13 syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0152095/>)

Genetic and Rare Diseases Information Center

- Trisomy 13 (<https://rarediseases.info.nih.gov/diseases/7341/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Trisomy 13%22](https://clinicaltrials.gov/search?cond=%22Trisomy+13%22))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28trisomy+13%5BTIAB%5D%29+OR+%28Patau+syndrome%5BTIAB%5D%29%29+AND+%28trisomy%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>)

References

- Chen M, Yeh GP, Shih JC, Wang BT. Trisomy 13 mosaicism: study of serialcytogenetic changes in a case from early pregnancy to infancy. *Prenat Diagn.* 2004Feb;24(2):137-43. doi: 10.1002/pd.814. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14974123>)
- Crider KS, Olney RS, Cragan JD. Trisomies 13 and 18: population prevalences, characteristics, and prenatal diagnosis, metropolitan Atlanta, 1994-2003. *Am J Med Genet A.* 2008 Apr 1;146A(7):820-6. doi: 10.1002/ajmg.a.32200. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18348276>)
- Di Giacomo MC, Susca FC, Resta N, Bukvic N, Vimercati A, Guanti G. Trisomy 13mosaicism in a phenotypically normal child: description of cytogenetic andclinical findings from early pregnancy beyond 2 years of age. *Am J Med Genet A.* 2007 Mar 1;143A(5):518-20. doi: 10.1002/ajmg.a.31515. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17163529>)
- FitzPatrick DR, Ramsay J, McGill NI, Shade M, Carothers AD, Hastie ND. Transcriptome analysis of human autosomal trisomy. *Hum Mol Genet.* 2002 Dec15; 11(26):3249-56. doi: 10.1093/hmg/11.26.3249. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12471051>)
- Graham EM, Bradley SM, Shirali GS, Hills CB, Atz AM; Pediatric Cardiac Care Consortium. Effectiveness of cardiac surgery in trisomies 13 and 18 (from the Pediatric Cardiac Care Consortium). *Am J Cardiol.* 2004 Mar 15;93(6):801-3. doi: 10.1016/j.amjcard.2003.12.012. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15019900>)
- Hall HE, Chan ER, Collins A, Judis L, Shirley S, Surti U, Hoffner L, Cockwell AE, Jacobs PA, Hassold TJ. The origin of trisomy 13. *Am J Med Genet A.* 2007 Oct1; 143A(19):2242-8. doi: 10.1002/ajmg.a.31913. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/17853475>)
- Iliopoulos D, Sekerli E, Vassiliou G, Sidiropoulou V, Topalidis A, Dimopoulou D, Voyiatzis N. Patau syndrome with a long survival (146 months): a clinical report and review of literature. *Am J Med Genet A.* 2006 Jan 1;140(1):92-3. doi:10.1002/ajmg.a.31056. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16333832>)

- Parker MJ, Budd JL, Draper ES, Young ID. Trisomy 13 and trisomy 18 in a defined population: epidemiological, genetic and prenatal observations. *PrenatDiagn.* 2003 Oct;23(10):856-60. doi: 10.1002/pd.707. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14558033>)
- Pont SJ, Robbins JM, Bird TM, Gibson JB, Cleves MA, Tilford JM, Aitken ME. Congenital malformations among liveborn infants with trisomies 18 and 13. *Am J Med Genet A.* 2006 Aug 15;140(16):1749-56. doi: 10.1002/ajmg.a.31382. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16835915>)
- Rasmussen SA, Wong LY, Yang Q, May KM, Friedman JM. Population-based analyses of mortality in trisomy 13 and trisomy 18. *Pediatrics.* 2003 Apr;111(4 Pt1):777-84. doi: 10.1542/peds.111.4.777. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12671111>)

Last updated September 9, 2021