

What Do We Know *Now* About Trisomy 13?

Trisomy 13 syndrome (Patau syndrome) is a disorder of human chromosomes which occurs in approximately 1 in 10,000-25,000 live-born infants. Trisomy refers to three copies of a chromosome instead of the normal two and in Trisomy 13 there is the presence of an extra #13 chromosome. Approximately 80% of infants with Trisomy 13 syndrome will have a full trisomy (affecting all cells) while the remainder will have a trisomy due to a rearrangement of cells called a translocation (an attachment of all or part of one chromosome to another chromosome) or have mosaicism (two different cell lines in an individual).

Most often a diagnosis of Trisomy 13 is **suspected** by findings seen on fetal ultrasound, or screening by maternal blood tests. **Optional invasive testing**, amniocentesis or chorionic villus sampling, **is needed to confirm a diagnosis** but carry a small risk to the fetus. A prenatal diagnosis of Trisomy 13, before 24 weeks, is often followed with an option to terminate; a decision made by 75% in the USA and 90% in Europe. A diagnosis can affect the care provided to those continuing pregnancy, their birthing options, and the care of a liveborn infant.

The new non-invasive prenatal test (NIPT) is increasing in use. A Positive Predictive Value (PPV) calculator is a tool used to determine accuracy of a NIPT *positive* result.

Infants born with Trisomy 13 have a recognizable pattern of physical features that often allows the health professional to make the diagnosis of the syndrome. Notable physical birth defects and, sometimes, anatomic changes of internal organs are present. Findings of significance include small head size (microcephaly); small eyes (microphthalmia) or sometimes an absent eye or faulty development of the retina; and failure of the forebrain to divide properly (holoprosencephaly). Cleft lip or cleft palate or both occur in about 60% of children and heart defects as described in the next paragraph. In addition, there are a number of less medically significant physical findings that are helpful in diagnosis. These include variations of ear shape, changes on the palm of the hand, and extra fingers and toes. Changes in foot development, including changes to the heel, the so-called rocker bottom foot, can occur.

Heart Defects

About 80% of children with Trisomy 13 will have a congenital heart defect. This can include: ventricular septal defect (VSD), an opening between the lower chambers of the heart which prevents the heart from pumping blood correctly (a heart murmur is generally heard from this finding); atrial septal defect (ASD), an opening between the two upper chambers of the heart making it difficult for the heart to pump sufficient oxygen-rich blood to body tissues (a heart murmur is often heard); patent ductus arteriosus (PDA), a defect involving the lack of closure of the channel that usually closes near the time of birth and thus remains open; and dextrocardia, which is a location of the heart on the right side of the chest. The majority of heart lesions are usually not those that cause death in the neonatal period but on occasion more medically serious heart defects can occur in Trisomy 13.

Medical Problems

The major implications of Trisomy 13 involve a predisposition to the congenital malformations (birth defects) mentioned above, an increased mortality in infancy, and developmental and motor disability in older children. In addition, older infants can have visual difficulties because of the findings mentioned above and a hearing loss. The increased mortality is related to difficulties with breathing due either to interrupted breathing (*apnea*) or problems of lung development. In addition, gastroesophageal reflux (backward flow of a small amount of stomach contents upward to the throat) and feeding problems can occur and predispose to aspiration (small amount of liquid inhaled or trickled into the lungs) which can precipitate aspirational pneumonia, a risk for survival.

Common Birth Defects seen in Trisomy 13

Omphalocele 10%
Holoprosencephaly 60% (an anatomic defect of the brain involving failure of the forebrain to divide properly)
Kidney defects 30%
Skin defects of the scalp 20%
Cleft lip and or palate or both 60%
Ocular (eye) abnormalities >50% such as small or absent eyes

Common Disorders and illness in Trisomy 13

Feeding difficulties
Gastroesophageal reflux
Slow postnatal growth
Apnea (central, obstructive or both)
Seizures
Hypertension
Developmental disabilities
Scoliosis
Frequent pneumonia
Urinary Tract infection
Chronic constipation

Routine follow-up care of infants with Trisomy 13

- Routine child care/anticipatory guidance
- Cardiac evaluation
- Eye evaluation
- Hearing test
- Scoliosis check through childhood
- Routine immunization
- Referral for Infant pre-school/early intervention program
- Ongoing Support

Survey of >350 parents of children with Trisomy 13 or 18

Most chose comfort care for their infant; 30% lived >1 year.
Of infants who received interventions; 50% lived >1 year.
Half said care of a disabled child is/was harder than expected.
89% reported a positive enriching experience regardless of lifespan.
[Janvier et al., 2012]

Survival in Trisomy 13

High risk of fetal loss early in pregnancy decreases as pregnancy progresses but during labor risk of loss is higher than for an unaffected fetus

5-8% survive past 1 year **without** extraordinary measures. Recent studies report increased survival in Trisomy 13 **with** medical interventions; *respiratory, nutritional, cardiac, and surgery when appropriate.*

Largest Trisomy 18/13 survival study in the USA reported Trisomy 13 survival of 11.5% at 1 year, and 9.7% at 5 years. [Meyer et al., 2016]

Largest Trisomy 18/13 cardiac surgery study in the USA-Trisomy 13 outcomes: In-hospital mortality 27.6%
Median survival post discharge 14.8 years [Peterson et al., 2017]

Often reported surgeries for Trisomy 13

Gastrostomy tube placement
Fundoplication
Cardiac surgery
Tracheostomy
Cataract surgery and Glaucoma
Cleft lip and/or Cleft palate repairs
Cataract surgery and Glaucoma
Gastrointestinal, various repairs

Resources: John C Carey, MD, MPH, Medical Advisor for SOFT, Professor of Pediatrics and Genetics, University of Utah
Janvier A, Farlow B, Wilfond BS. 2012. [The Experience of Families With Children With Trisomy 13 and 18 in Social Networks](#) .Am Academy of Pediatrics, doi:10.1542/peds.2012-0151
Meyer RE, Liu G, Gilboa SM, Ethen MK, Aylsworth AS, Powell CM, Flood TJ, Mai CT, Wang Y, Canfield MA, for the National Birth Defects Prevention Network. 2016. Survival of children with trisomy 13 and trisomy 18: A multi-state population-based study. Am J Med Genet Part A 9999A:825–837. <http://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.37495/full>
Peterson JK, Kochilas LK, Catton KG, Moller JH, Setty SP. [Long-term Outcomes of Children with Trisomy 13 and 18 After Congenital Heart Disease Interventions](#). *Ann Thorac Surg*. 2017 Apr 26. pii: S0003-4975(17)30375-2. doi: 10.1016/j.athoracsur.2017.02.068. [Epub ahead of print]
Translated from English to Spanish by: Nadin Koharic, certified Literary and Scientific translator (Universidad del Salvador, Bs As, Argentina)
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Contact: SOFT, 2982 South Union St. Rochester, NY 14624, phone 800-716-7638 www.trisomy.org