Patau Syndrome (Trisomy 13) Fact Sheet

Patau syndrome due to trisomy 13 is a medically complex condition of human chromosomes that occurs in approximately 1 in 10,000-25,000 live-born infants. Syndrome refers to a recognizable pattern of physical findings. Trisomy refers to three copies of a chromosome instead of the normal two. Therefore in Patau syndrome there is the presence of an extra 13th chromosome, trisomy 13.

There are 3 types of chromosome findings in persons with Patau syndrome. About 90% of infants will have a **full or complete trisomy** in all body cells. The remainder will have a **partial trisomy** due to a rearrangement of part or all of one chromosome (usually attached to another chromosome) called a **translocation** or have **mosaicism** which is a mixture of two different cell lines (usually some normal cells and some trisomy 13 cells).

Most often a diagnosis is suspected by findings seen on fetal ultrasound or screening by maternal blood tests like noninvasive prenatal screening (NIPS). Diagnostic testing such as amniocentesis or chorionic villus sampling can be performed during the pregnancy to confirm a diagnosis. Otherwise testing can be done after the infant is born.

Three different categories of challenges are summarized below: birth defects and other conditions, child development, and survival.

Birth Defects

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. Almost all children born with Patau syndrome will have birth defects; however each child is different, the occurrence is highly variable, and no child has all of these findings.

Cardiac (80% of infants)
- Ventricular septal defect (VSD)
- Atrial septal defect (ASD)
- Patent ductus arteriosus (PDA)
- Dextrocardia

Other
- Small head (microcephaly)
- Holoprosencephaly- 60%
- Cleft lip with or without cleft palate- 60%
- Ocular (eye) abnormalities- >50% such as small or absent eyes
- Kidney defects
- Skin defects of the scalp
- Omphalocele

Common Disorders/Illnesses

- Small size compared to individuals of the same age
- Developmental delays
- Feeding difficulties
- Gastroesophageal reflux
- Chronic constipation
- Hearing loss
- Visual difficulties
- Seizures
- Scoliosis
- Urinary Tract infection
- Obstructive sleep apnea (OSA)
- Hypertension

Child Development

Children will have developmental delays meaning that they will meet younger developmental milestones at older ages. Positive support, quality educational programs, stimulating home environment, and supportive therapies lead to increased enrichment of the children's lives. This leads to a wide range of developmental skills in language and communication in addition to gross and fine motor skills.

Survival

Survival in infants, children, and adults is constantly changing and improving. In the past it was said that only 5-8% would survive past 1 year. Recent studies now report increased survival in Patau syndrome with medical interventions; respiratory, nutritional, cardiac, and surgery when appropriate. The largest trisomy 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] The largest trisomy 13 cardiac surgery study in the USA showed an In-hospital mortality of 27.6% but the median survival post discharge 14.8 years. [Peterson et al., 2017] There was another recent survey of over 350 parents of children with trisomy 13. Most parents in the study chose comfort care for their infant and 30% lived over 1 year. In the study of the infants who received interventions 50% lived over 1 year. [Janvier et al., 2012]

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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, et al for that 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 170A:825–837.
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.
Cortezzo D , Tolusso LK , Swarr DT 2022 Perinatal Outcomes of Fetuses and Infants Diagnosed with Trisomy 13 or Trisomy 18 J Pediatr 247:116-123.e5. doi: 10.1016/j.jpeds.2022.04.010.

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