Patau's Syndrome (Trisomy 13)

Patau's syndrome (trisomy 13) carries a high mortality rate with multiple congenital abnormalities which result in severe physical and mental impairment.

- It is usually due to a free-standing trisomy with an extra number 13 chromosome, instead of the usual pair, in all cells.
- An unbalanced chromosome translocation can also occur - commonly, a Robertsonian translocation, in which an extra copy of chromosome 13 is attached to another chromosome.
- There may also be mosaic variations in which some cells are normal with 46 chromosomes and others have the extra chromosome. Infants with mosaic variations tend to be less severely affected.

Epidemiology

The prevalence of trisomy 13 is between 1 in 5,000 and 1 in 29,000 live births, and it is the third most common autosomal trisomy in newborns after trisomy 21 and trisomy 18[1].

Risk factors

- A personal or close family history of giving birth to an affected child increases the risk.
- Risk rises with increasing maternal age but not as markedly as with Down's syndrome (trisomy 21) or Edwards' syndrome (trisomy 18)[2].

Presentation[1, 3]

Many fetuses never survive until term and are stillborn or spontaneously abort. Features include:

- Intrauterine growth restriction and low birth weight.
- Congenital heart defects: these occur in 80%; they include atrial septal defect, ventricular septal defect, patent ductus arteriosus, dextrocardia.
- Holoprosencephaly: the brain doesn't divide into two halves; this can present with midline facial defects including:
  - Cleft lip and palate.
  - Microphthalmia or anophthalmia.
  - Nasal malformation.
  - Hypotelorism (reduced distance between the eyes) or cyclops.

- Other brain and central nervous system abnormalities, including:
  - Neural tube defects.
  - Other anatomical defects of the brain
  - Severe learning disability.
  - Problems with control of breathing (central apnoea).

- Other craniofacial abnormalities include:
  - Microcephaly.
  - Scalp defects (cutis aplasia: skin missing from the scalp).
  - Ear malformations and deafness.
  - Capillary haemangioma.
Gastrointestinal abnormalities: omphalocele, exomphalos, hernias.
Urogenital malformations: polycystic kidneys, micropenis or hypertrophy of the clitoris.
Abnormalities of hands and feet: polydactyly (extra fingers or toes), small hyperconvex nails and rocker-bottom feet.

Differential diagnosis

- Infants with Patau's syndrome and Edwards' syndrome can have similar features and be difficult to differentiate.
- Pseudotrisomy 13 is used to describe babies with features typical of trisomy 13 but with a normal karyotype.

Investigations and management

Cytogenetic studies and chromosomal analysis will confirm the diagnosis. Organ systems will need specific investigation depending on the abnormality - eg, echocardiography for cardiac abnormalities; skeletal radiography, etc.

Treatment of a 'liveborn' infant is generally supportive but life-sustaining measures are not always carried out. Considerable thought and discussion are recommended before undertaking measures such as surgical correction of abnormalities. Nasogastric or gastrostomy feeding is feasible but the clinician should take into account the wishes of the parents and any potential harms it may cause the infant. Parents will need a great deal of support and counselling.

If Patau's syndrome is due to an unbalanced chromosome translocation or structural chromosomal abnormality, both parents should undergo chromosomal analysis. It may be that the translocation in the infant occurred de novo but a balanced translocation may be found in one of the parents. This has significance for future pregnancies because of a higher risk of recurrence. Other family members may also be affected.

Screening and/or prenatal diagnosis should be offered for future pregnancies. Women who have had a previous trisomic pregnancy, especially those under 35 years of age at the time, appear to be at an increased risk of future pregnancies being trisomic. See also the separate Prenatal Diagnosis article.

Prognosis

- Life expectancy is very limited. Median survival is 2.5 days.
  - About 50% live longer than one week.
  - 5-10% of infants live longer than one year.
- Profound learning disability and developmental delay occur in survivors. Seizures and feeding difficulties are common.
- However, trisomy 13 mosaicism causes a variable phenotype ranging from complete trisomy 13 with neonatal death, to just a few dysmorphic features and prolonged survival.

Screening

- Specific ultrasound findings may suggest trisomy 13 and subsequent cytogenetic studies may therefore be indicated. Findings include increased nuchal translucency, cardiac defects, neural tube defects, facial clefting, renal abnormalities and omphalocele.
- A study from Kings College Hospital showed that at the 11- to 13(+6)-week scan, the measurement of fetal nuchal translucency and fetal heart rate and fetal examination for holoprosencephaly, exomphalos and megacystis can identify >90% of fetuses with trisomy 13.
- First-trimester multiple marker screening (that which is currently offered for Down's syndrome screening) may also help to identify a fetus with trisomy 13 or 18. Markers can include maternal age, nuchal translucency measurement, pregnancy-associated plasma protein A (PAPP-A) and human chorionic gonadotrophin (hCG). Ultrasound at that time may also show fetal anomalies.
- Second-trimester screening tests can be offered if the mother presents later. See the separate Antenatal Screening for Down's Syndrome article for more details about second-trimester screening tests.
- In a UK-based study from 2003, 44 cases of trisomy 13 and 88 cases of trisomy 18 were examined. 64% were first detected by chromosomal analysis because of abnormalities noted on fetal anomaly scanning in the second trimester. 3% of cases were detected through the serum screening programme currently offered for Down's syndrome. 11% of cases were detected postnatally. Of note, in the same study, 12% of couples chose to continue with the pregnancy after prenatal diagnosis.

Prenatal diagnosis

- Amniocentesis or chorionic villus sampling is needed to make a definitive prenatal diagnosis.
- Cytogenetic study of fetal blood may also be carried out.
- Mothers over the age of 35 may choose to go straight to diagnostic testing. Others mothers may choose to have diagnostic testing after a positive screening test.

History

Klaus Patau was a German-born, American human geneticist. Patau et al described the syndrome in 1960. The clinical appearance of trisomy 13 was first described by Erasmus Bartholin in 1657 but he was unaware of the aetiology.
Further reading & references


5. Pseudotrisomy 13 syndrome; Online Mendelian Inheritance in Man

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<th>Author: Dr Colin Tidy</th>
<th>Peer Reviewer: Dr Anjum Gandhi</th>
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