



## ~ Trisomy 13 Facts ~

Trisomy 13 syndrome is a disorder of human chromosomes which occurs in approximately 1 in 10,000 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 such as normal cells and trisomy cells).

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. (Genetic testing must be done to confirm diagnosis.) 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. Cleft lip or cleft palate or both occur in about 60% of children. 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 neo-natal period but on occasion more medically serious heart defects can occur in Trisomy 13.

### **Synonyms:**

- Trisomy 13
  - Patau Syndrome
  - Trisomy 13-15
  - D Trisomy Syndrome
- (the last two terms are usually not used at the present time)

### **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*.

#### **Important and Common Birth Defects 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%

#### **Common Disorders in infants and young children with Trisomy 13**

- Feeding difficulties
- Gastroesophageal reflux
- Slow post natal growth
- Apnea
- Seizures
- Hypertension
- Kidney defects
- Developmental disabilities
- Scoliosis

#### **Routine follow-up care of infants with Trisomy 13**

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

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