

The Support Organization for Trisomy 18, 13 and Related Disorders (SOFT)



~ Trisomy 18 Facts ~

Trisomy 18 syndrome is a disorder of human chromosomes which occurs in approximately 1 in 7,000 live born infants. Trisomy refers to three copies of a chromosome instead of the normal two and in trisomy 18 there is a presence of an extra #18 chromosome. Over 90% of infants with Trisomy 18 syndrome will have a full trisomy in all body cells while the remainder will have a trisomy due to a rearrangement of part or all of one chromosome (attached to another chromosome), called a translocation, or have mosaicism (two different cell lines such as some normal cells and some trisomy cells).

Infants born with Trisomy 18 usually are small in size at birth. There is 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). These physical findings are not medically significant but provide clues. They include: prominence to the back part of the head, short eyelid fissures, small mouth and jaw, external ear variations, clenched fist with index finger overlapping the third, and 5th finger overlapping the 4th, small fingernails, underdeveloped or altered thumbs, short sternum (breastbone), club feet and redundant skin at the back of the neck.

Synonyms:

- Trisomy 18
- Edward Syndrome
- Trisomy E
- Trisomy 16-18

(the last two terms are usually not used at the present time)

The major impact of Trisomy 18 is a predisposition to congenital malformations (birth defects), a high incidence of infant mortality, and developmental and motor disability in older infants and children.

Birth Defects

The congenital malformations involve the medically significant findings mentioned above as well as the presence of some internal or external birth defects. The most common and important is a defect of the heart. Over 90% of children with Trisomy 18 will have a congenital heart malformation; these include:

- Ventricular septal defect (VSD) which is 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 heart defect involving the lack of closure of the channel that usually closes near the time of birth and thus is a persistence of the opening of this channel.

In addition, children with Trisomy 18 usually have an alteration of one of the four heart valves. This combination is referred to as a ventricular septal defect with polyvalvular dysplasia.

The majority of heart lesions are usually not those that cause death in the neonatal period but about 10% of children with Trisomy 18 will have a life-threatening heart defect noted before or soon after birth. These include a double outlet right ventricle and hypoplastic left heart.

Medical Problems

The increased occurrence of infant mortality is related to a combination of factors but most importantly *central apnea*, where the brain does not give the message to breathe. Other complicating factors include difficulty feeding with aspiration, and a predisposition to *aspiration pneumonia*, and under-development of the lungs. The *heart defects* can play some role in this but are usually not the only cause of this increased mortality.

Important and common birth defects seen in Trisomy 18:

- Congenital heart defects 90%
- Multiple joint contractures 10%
- Spina bifida
- Hearing loss >50%
- Radial aplasia (underdevelopment or missing radial bone of forearm) 5-10%
- Cleft lip 5-10%
- Birth defects of the eye 10%

Common Disorders in Trisomy 18

- Feeding difficulties
- Gastroesophageal reflux
- Slow post natal growth
- Apnea
- Seizures
- Kidney defects
- Urinary tract infections
- Developmental disability
- Scoliosis

Routine follow-up of infants with Trisomy 18

- Routine child care/anticipatory guidance
- Cardiac evaluation
- Eye evaluation

- Hearing test
- Infant/pre-school program early intervention
- Ongoing Support
- Routine ultrasound for Wilms tumor
- Routine immunization
- Referral for feeding clinic if appropriate
- Scoliosis check

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