

APERT SYNDROME

Description

Apert Syndrome, first described in 1906 by E. Apert, a French physician, is characterized by malformations (abnormal growth) of the skull and face accompanied by fusion (webbing) of the fingers and toes. The shape of the head appears long and pointed at the top. Wide-set, bulging eyes that tilt down at the sides, a sunken-in looking mid face (middle of eye socket to upper jaw) and dental crowding are often found.

Skulls of newborns are comprised of sutures or plates that are loosely connected and eventually join together to form the skull. In children with Apert's, these sutures are prematurely fused (joined together). This fusion (craniosynostosis) can cause increased pressure on the brain as it develops causing the head to grow upwards.

Children with Apert's may also have one or more of these abnormalities:

- Visual problems due to eye muscle imbalance
- Hearing loss due to frequent infections
- Severe acne
- Mild mental retardation
- Cleft palate
- Hyperactive sweat glands

Prevalence/Causes

Apert syndrome occurs in approximately 1:160,000 live births. It is caused by a genetic mutation (change). Neither parent is responsible for this mutation *first* occurring in a family line. However, a parent *with* this syndrome has a 50% (1:2) chance of passing the condition onto a child. Offspring *without* Apert's have NO greater likelihood of having a child with Apert's. This syndrome is unlikely to occur in future offspring if neither parent of a child with Apert's has the condition.

Treatment

The treatment of Apert syndrome is surgery-intensive:

- Skull reshaping and frontal-orbital advancement to increase space within skull at 3-4 months
- Mid-face advancement – 4-5 years
- Fingers/toes separation
- Osteotomy (upper/lower jaw surgery) – teen years
- Genioplasty (chin/cheek surgery)
- Rhinoplasty (nose surgery)
- Eyelid surgery

A craniofacial team approach is recommended because of the varied nature of the problems (plastic surgeon, neurosurgeon, orthodontist, geneticist; pediatric ophthalmologist, dentist and otolaryngologist; psychologist, speech therapist).

Associated Conditions

Breathing: Children with Apert syndrome breathe noisily because their noses may be smaller or their airways narrower.

Eyes: The area housing the eyeball (the orbit) is too small preventing the eyelids from closing completely.

Moisturizers can help.

Ears: Prevalent ear infections may decrease hearing ability. It may be recommended that tubes be placed in the ears to prevent such loss.

Fused Fingers/Toes: Fused fingers can be surgically corrected though once separated, the fingers tend to grow crookedly requiring additional surgeries. Fused toes do not impede walking but for cosmetic purposes are separated.

Skin: Hyperactive sweat glands are most apparent during sleep. Acne, especially during puberty is often a problem.

Psychosocial

Because of the various factors involved with Apert syndrome, many of them visually apparent, children with these conditions need to be encouraged to excel in any area that gives them satisfaction and a sense of accomplishment. Moreover, often teased through their early and middle school years, it is crucial that children with Apert's are helped to develop positive self-esteem so that they have strong coping mechanism.

Skin: Hyperactive sweat glands are most apparent during sleep, but can be a problem during adolescence when the importance of one's physical presentation becomes central. Acne, especially during puberty is often a problem, but at least this is a problem that is commonly shared across the peer group.