

a guide to understanding
apert
syndrome

a publication of children's craniofacial association

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This parent's guide to Apert syndrome is designed to answer questions that are frequently asked by parents of a child with Apert syndrome. It is intended to provide a clearer understanding of the condition for patients, parents and others.

how can children's craniofacial association (cca) benefit my family?

CCA understands that when one family member has a craniofacial condition, each person in the family is affected. We provide programs and services designed to address these needs. A detailed list of CCA's programs and services may be found on our Web site at www.ccakids.com or call us at 800.535.3643.

The information provided here was written by a member of the Medical Advisory Board of the Children's Craniofacial Association.

This booklet is intended for information purposes only. It is not a recommendation for treatment. Decisions for treatment should be based on mutual agreement with the craniofacial team. Possible complications should be discussed with the physician prior to and throughout treatment.

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what is apert syndrome?

apert syndrome is a congenital condition and falls under the broad classification of craniofacial/limb anomalies. Apert syndrome causes abnormal growth of several bones in the body, primarily the skull, midface, hands, and feet.

how do I recognize this condition in my child?

the skull is usually severely affected, as well as the entire face, especially the eyes and jaw. The skull bones are prematurely fused and the skull is unable to grow normally; the midface (that area of the face from the middle of the eye socket to the upper jaw) appears retruded or sunken; as a result the eyes bulge out and the eyelids tilt downward. The face can have acne. The hair may be unruly. Cleft palate and deafness are common. Patients also have syndactyly, which means joining of fingers and toes. Many children also have mental delays that can vary from mild to severe. In many cases, speech development and behavioral problems become more obvious as the child gets older. Apert syndrome is named for the French physician who first described it, E. Apert, in 1906.

what causes apert syndrome?

apert syndrome is a result of genetic mutation. The syndrome can be inherited from a parent who has Apert's, or may be a fresh mutation. It occurs in approximately 1 per 160,000 to 200,000 live births. When you have Apert syndrome, you have a 1 in 2 (50%) chance of passing this condition to your child. This is because each of us gets 1/2 of our genetic makeup from each parent. However, Apert's is not a recessive trait, which means that the normal child of a parent with Apert syndrome is no more likely to have a child with Apert's than any other person; also, if you have a child with Apert's and you do NOT have Apert's, YOU are no more likely to have another child with Apert's than anyone else in

the population. Studies have shown that Apert syndrome occurs more often in children of older fathers.

Recently studies were conducted at Oxford University and they managed to identify the actual genetic change which occurs in Apert syndrome. The following is a quote from a letter sent to the test families by Oxford.

“A total of 86 children and adults affected with Apert syndrome have been seen. From the blood samples which have been donated for research, we have identified the genetic change that causes the condition. The change is in a gene on chromosome number 10 called 'Fibroblast Growth Factor Receptor 2' (FGFR2 for short). We all have two copies of this gene (one from mother, one from father), which is composed of a string of about 2000 of the chemical building blocks that make up the genetic material called DNA. When Apert syndrome occurs, just one particular building block in one of these two gene copies has been exchanged for another. The other gene copy is entirely normal. This one tiny change in the FGFR2 gene results in the physical features of Apert syndrome.”

who is involved in the treatment of Apert syndrome?

Ideally, treatment of Apert syndrome begins at birth with accurate diagnosis, identification of the child's individual needs, and the proper facilities to administer what is needed. Treatment for these children requires careful planning with multiple surgeries ranging from minor to complex. Treatment from many different specialists working as a team is necessary and can help in avoiding complications.

A craniofacial team may consist of a craniofacial surgeon, neurosurgeon, ENT, audiologist, speech pathologist, oral surgeon, psychologist, ophthalmologist, and an orthodontist. The team approach is used by these physicians to determine the best collaborative corrective plan for the deficiencies of the child.

what treatment is available for apert syndrome?

In a normal newborn child, the skull is made up of several “plates” which remain loosely connected to one another, gradually growing together to form the adult skull. In Apert syndrome, these plates fuse too early, restricting brain growth, and causing increased pressure in the brain as it grows. This is known as craniosynostosis. Early surgery to detach the plates from each other relieves the pressure. During this early surgery, which usually takes place within the first year of life, some “cranial remodeling” may be done by your surgeon to give the child a more normal appearance.

The “retrusion” or lack of development of the midface is what could be described as concave or dished in profile. As the skull grows, the middle third of the face grows slower, resulting in a more pronounced retrusion over time. A surgical procedure known as the LeFort III can be used to correct this condition. The procedure is usually done after substantial growth is complete (preadolescence) and may be repeated as necessary. The LeFort procedure involves detaching the facial bones from mid eye to upper jaw and spacing this area out with bone grafts so that a proper alignment is made. If the forehead has not grown well either, a procedure called “monoblock” may be used.

In the last few years, many surgeons have come to prefer “distraction” of the bones using either the Rigid External Distraction (RED) system or internally placed distractors. With this procedure, the operation remains the same but now the bone is gradually pulled forward instead of moved at once during surgery. This leads to formation of new bone over time.

In addition, your child may need a frontal-orbital advancement within the first twelve months to increase space within the skull and the size of both orbits (the part of the skull which holds the eyeball), a facial bi-partition to widen the upper jaw, derotate the orbits, and to narrow the upper

face, and/or during the teen years an osteotomy (cutting through the bone of the upper and lower jaw) to correct further problems.

The severity of your child's Apert syndrome determines whether he/she needs some or all the procedures described here.

how are the fingers and toes affected?

The fusion of the fingers and toes along with the craniofacial problems mentioned above is what really separates Apert from other similar syndromes. This condition is called syndactyly. It always involves fusion of the soft tissues of the first, middle, and ring fingers, and often there is fusion of the bones themselves. The joints usually do not move well, if at all. The thumb may be fused into the hand, or may be free. Surgery is used to separate the fingers as needed to obtain better function. The feet and toes are affected similarly, but surgery is usually only done in cases where the ability to walk would be impaired.

can children with apert syndrome have other problems?

The following problems have been observed in some children with Apert syndrome. However, whether or not they were caused by Apert syndrome is uncertain.

- Various heart defects
- Cleft palate
- Dextrorotation
- Pulmonary Arteries
- Patent Ductus Arteriosus (PDA)
- Tracheoesophageal Fistula
- Pyloric stenosis
- Polycystic kidneys
- Bicornate uterus
- Hydrocephalus

- Ear infections which can cause hearing loss
- Sleep Apnea, small nose and airway passage make breathing difficult
- Severe acne, hyperactive sweat glands
- Increased incidence of eye injuries, imbalance of eye muscles

can complications occur?

Yes. Minor complications include infections of the skin, around the stitches, collections of blood under the skin, and hair loss. Orthodontic care is almost always required especially if the jaw is moved. Tooth loss may occur. Scalp and face numbness is common, especially after surgery. This may or may not get better with time. Bruising and swelling always occur to some degree.

Major complications may require surgery or a hospital stay. More severe infections may occur, especially if distractors to move bone are used. Severe bleeding may need blood transfusions or more surgery. Double vision and other vision problems may occur that may need eye surgery. Blindness is extremely rare. Brain damage and death are also extremely rare. Fortunately, with the multi-team approach that most craniofacial centers have, complications are kept to a minimum.

Remember, new advances and procedures concerning Apert syndrome are constantly being developed, so ask questions and be an advocate for your child!



children's craniofacial association

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