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 website at www.ccakids.com or call us at 800.535.3643.

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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.

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?

Apert syndrome is named for the French physician who first described it, E. Apert, in 1906. You or the delivery team will likely recognize this condition immediately at birth, if not before during prenatal visits, using ultrasound imagery. Based on the skull shape and syndactyly of the hands and feet, Apert Syndrome has very recognizable features. 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. Cleft palate and deafness are common. Patients also have syndactyly, which means joining of fingers and toes. Some children also have developmental delays that can vary from mild to severe. In many cases, speech and behavioral differences become more obvious as the child gets older.

what causes apert syndrome?

Apert syndrome is a result of genetic mutation. The syndrome can be inherited from a parent who has Apert, or may be a new, sporadic 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 is not a recessive trait, which means that the unaffected child of a parent with Apert syndrome is no more likely to have a child with Apert than any other person; also, if you have a child with Apert and you do NOT have Apert, YOU are no more likely to have another child with Apert than anyone else in the population.

Apert syndrome is caused by a change (mutation) in the fibroblast growth factor receptor-2 (FGFR2) gene. This gene plays a critical role in skeletal development. Genes provide instructions for creating proteins that play distinct roles in our body. When a mutation of a gene occurs, the protein product may not work as it should. In Apert syndrome, mutations in FGFR2 result in these receptors not properly communicating with fibroblast growth factors. This affects the formation of normal sutures in the

and can obstruct the development of many other structures in the body. This improper formation is what causes the malformations seen in Apert syndrome.

In almost all reported patients, the disorder has been caused by one of two specific mutations of the FGFR2 gene. (These mutations are designated “Ser252Trp” and “Pro253Arg.”) These mutations may cause slightly different presentations, including the severity of syndactyly. Different mutations in the FGFR2 gene may cause several other related disorders, including Pfeiffer syndrome, Crouzon syndrome, and Jackson-Weiss syndrome.

From: NORD, National Organization for Rare Disorders, 2023

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, otolaryngologist (ENT : ear, nose, and throat surgeon), plastic surgeon, hand surgeon, 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 child.

what treatment is available for apert syndrome?

In an unaffected 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 improve the shape of the child's head.

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 properly developed, 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 patient's 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?

All children born with Apert syndrome have fusion of the fingers and toes. This condition is called syndactyly and its consistent occurrence distinguishes Apert syndrome from similar craniofacial conditions. The syndactyly typically involves the all the fingers, and may involve the thumb as well. There is almost always some bone involvement and the joints in the fingers do not move. There is motion in the joints between the hands and the digits. The thumb is short and deviates away from the hand. Despite these differences, surgical correction can

be performed early in life to allow the child to explore their environment with their hands, gain independence in performing their tasks of daily living, as well as participate in sports and other hobbies.

Although syndactyly releases are usually performed within the first three years of life, thumb lenghthening and straightening is typically deferred until school age. Additional corrective straightening procecdures may also be performed later during adolescence. Routine amputations are rarely necessary and should be avoided, although functional outcomes are similar even when a digit is amputated.

Surgery to separate the toes may be performed at the same time as hand surgery. Although the procedure predictably creates a foot with 5 toes, no significant functional gain is achieved. Moreover, other foot and ankle issues may become significant later in childhood regardless of whether the toe syndactyly has been addressed.

can children with apert syndrome have other problems?

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

- Need to monitor cranial pressure
- Various heart defects
- Cleft palate
- Dextrorotation
- Pulmonary Atresia
- Patent Ductus Arteriosus (PDA)
- Tracheoesophageal Fistula
- Pyloric stenosis
- Polycystic kidneys
- Bicornate uterus
- Hydrocephalus
- Ear infections, which can cause hearing loss if untreated

- Sleep apnea, small nose and airway passage make breathing difficult
- Severe acne, hyperactive sweat glands (hyperhydrosis)
- Increased incidence of eye injuries, imbalance of eye muscles
- Neurodivergence and learning/behavior differences

can complications occur?

Yes. Most often complications occur during surgery or recovery. 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 surgical repair. Double vision and other vision problems may occur that may require 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!
If you need any support, please contact us at CCA.

can my child lead a "normal" life?

Absolutely. The best thing you can do for your child is raise them like you would raise an unaffected child, with just a little care on your part to build a strong, supportive network, to learn about medical stress and trauma, and to advocate for their inclusion in school, your community, and activities they enjoy.

children's craniofacial association

empowering and giving hope to individuals and families affected by facial differences.

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