



Apert Syndrome

A guide to diagnosis
and treatment



**Boston
Children's
Hospital**

Craniofacial Anomalies
Program

Having a child born with Apert syndrome may feel overwhelming. Knowing your child will need medical care from a large team of medical providers is a lot for any parent to process. We hope you find comfort knowing that the Craniofacial Anomalies Program at Boston Children's Hospital cares for more than 500 patients every year with a range of craniofacial diagnoses. As one of the oldest such programs in the world, our extensive experience and commitment to innovative and compassionate care have established us as a national leader in the treatment of children with Apert syndrome.

At Boston Children's Hospital we use a collaborative, team, and patient-centered approach for our patients. Our surgeons are experienced in multiple surgical techniques, allowing us to develop and tailor a specific treatment plan to best fit your child's individual needs.

Apert syndrome is a genetic disorder, characterized by anomalies of the skull, face, and extremities that impacts an individual's health, daily function, and social interaction.

Successful treatment of patients with Apert syndrome requires an interdisciplinary medical team, including:

- plastic surgeons
- oral surgeons
- hand surgeons
- neurosurgeons
- otolaryngologists
- (ear, nose and throat)
- pediatric dentists
- orthodontists
- geneticists
- speech pathologists
- audiologists
- physician assistants
- nurses
- complex care pediatrics
- neuropsychologists

Boston Children's Hospital Craniofacial Anomalies Program incorporates each of these specialists with unique skills to deliver the very best care for patients and their families. Since Boston Children's Hospital is the only hospital in Massachusetts focused exclusively on children, our entire team is specially trained to optimize treatment for young patients and their families.



John Meara, MD, DMD
Plastic Surgeon-in-Chief

Overview of Apert Syndrome

Apert syndrome, also known as “acrocephalosyndactyly,” is a rare complex genetic disorder that may affect a child’s face, skull, teeth, hands, and feet. The impact on each child can be different as there is a wide range of clinical expression and severity. It occurs in one out of 100,000 newborns.

Clinical features of Apert syndrome

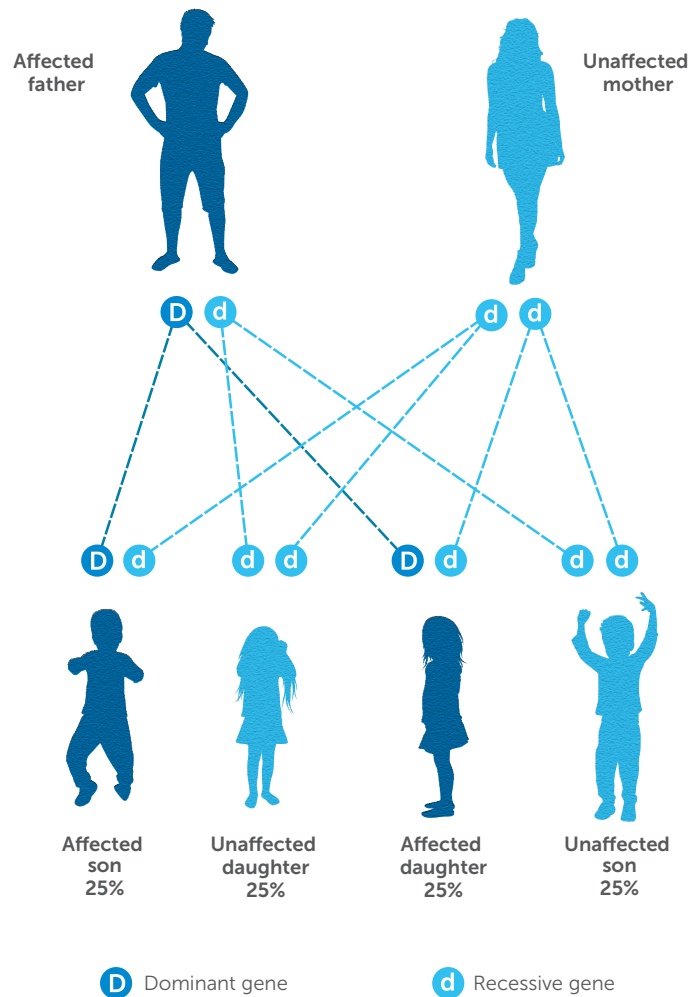
- craniosynostosis: early closure of growth plates of the skull, resulting in changes in the shape of the head and possible increased pressure on the brain
- midfacial hypoplasia: decreased growth of the central face causing a sunken facial appearance, serious breathing problems, or sleep apnea.
- syndactyly: fusion of the fingers and/or toes
- other features may include: crowded teeth, cleft palate, hearing loss, fused spinal bones, and acne

Diagnosing Apert syndrome

- A trained craniofacial team including a geneticist should evaluate your baby.
- In some cases the provider may order x-rays and/or a computed tomography (CT) scan to help confirm the diagnosis.
- Additionally, genetic testing may be performed using a blood or saliva sample.

Will my child have developmental limitation as a result of Apert syndrome?

Some children with Apert syndrome have developmental delays. However, this is not true for all children with the condition. The needs of each child are carefully assessed and individually managed by our collaborative interdisciplinary team. Some children benefit from receiving speech therapy, physical therapy, or occupational therapy. Our craniofacial neuropsychologist will evaluate your child and provide access to developmental or cognitive interventions that might prove beneficial.



Why was my child born with Apert syndrome?

Apert syndrome is caused by a specific change (mutation) in the Fibroblast Growth Factor Receptor 2 gene (FGFR2). This gene is responsible for facilitating proper bone development and when altered can cause premature fusion of cranial bones (craniosynostosis) and maldevelopment of extremity bones (syndactyly). The FGFR2 gene is also involved in similar craniofacial syndromes, including Crouzon, Pfeiffer, and Jackson-Weiss.

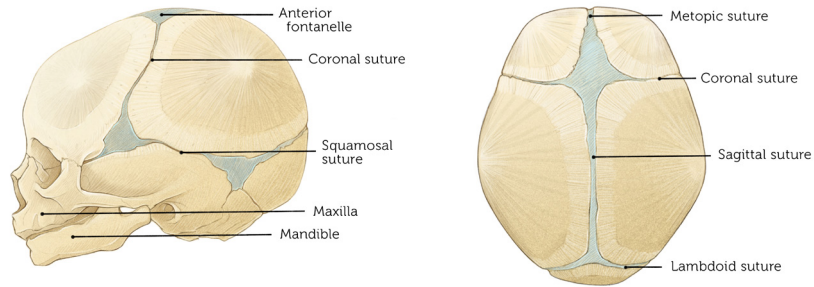
If I have other children, will they also have Apert syndrome?

In most cases, there is no family history of Apert syndrome. The risk of having another child affected with Apert syndrome is low but not zero. This is due to the very remote possibility that the genetic change occurred in more than one of the parent's germ cells (sperm or egg). In such rare instances, there would be a 50 percent chance of passing it on to another child.

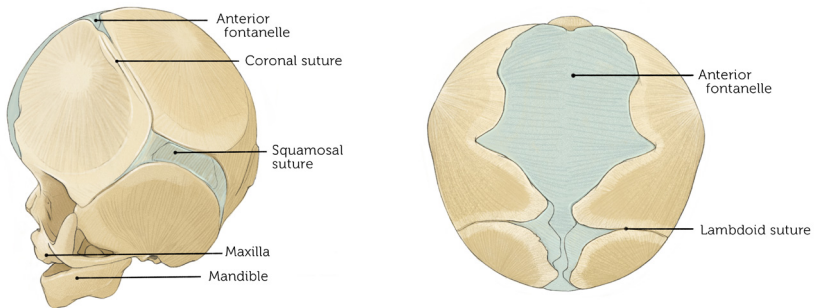
When my child becomes an adult and has children of his or her own, what are the chances they will also have Apert syndrome?

If your child has Apert syndrome each of his or her children will have a 50 percent chance of also having the condition.

Normal skull



Apert syndrome skull



Care and treatment of Apert Syndrome

While the prospect of multiple operations over your child's lifetime can seem overwhelming, it's important to remember that each of these procedures are very successful. In order to understand the overall management of a child with Apert syndrome, it is helpful to have a general outline of when the various procedures would be needed.

Please see page 18 for a comprehensive timeline of your child's care.

Craniofacial Treatment

Which craniofacial findings are commonly associated with Apert syndrome?

Since growth plates (also called "sutures") in the skull have closed too early, children with Apert syndrome have misshapen heads that are typically too wide and tall. The eyes appear very prominent resulting from shallow eye sockets, and the middle part of the face appears flat or sunken. The nose is short and has a beak-like prominent shape. The teeth are crowded and there may be an associated cleft palate or blockage of the breathing passages (called "airway obstruction").

Treatment options for breathing problems

If your child is born with airway obstruction, a multidisciplinary consultation with our providers specializing in otolaryngology, sleep medicine and/or pulmonology will be sought soon after birth. Clinical evaluation of your child's breathing problems may include examination of the airway with a small camera or obtaining a sleep study. There are several options for treating airway obstruction in children with Apert syndrome. These options include wearing a Continuous Positive Airway Pressure (CPAP) mask, tonsillectomy and/or adenoidectomy, and/or mid-facial advancement (also called a "Le Fort III"). In severe cases, placement of a tracheostomy (a surgical procedure creating an opening through the neck into the airway) may be needed until your child is old enough to undergo other operations.

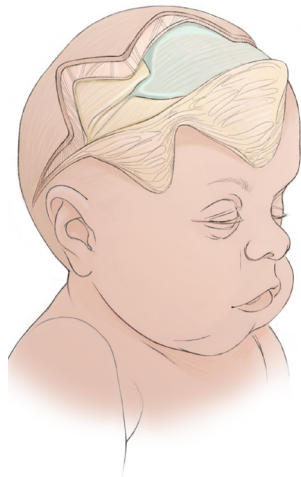
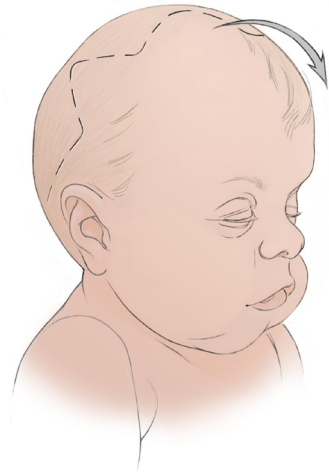
Care and treatment of Apert syndrome

What is the typical treatment for a child's face and skull?

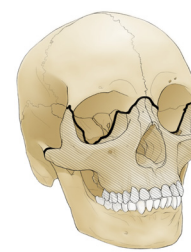
At Boston Children's Hospital we offer an innovative minimally invasive approach to treating craniosynostosis. Whenever possible, endoscopic (minimally invasive) release of the fused cranial sutures is performed early in life, usually when the child is between 2 and 4 months old. By performing this procedure endoscopically followed by helmet therapy, we can significantly reduce the skull shape deformity in children with Apert syndrome. More importantly, our center advocates for an early endoscopic suture release to ensure that the child's brain has adequate room to grow and minimizes the risk of brain injury.

For older patients where endoscopic treatment is not possible, a procedure such as a calvarial vault remodeling or a fronto-orbital advancement is usually performed when the child is between 9 and 11 months old. The type of procedure performed depends on the specific needs of your child and which of his or her cranial sutures are fused. These operations involve a plastic surgeon and a neurosurgeon who work together to carefully release the fused sutures and reshape the skull. Our surgeons use a zigzag incision to camouflage or hide the scar entirely within the hair-bearing scalp.

Incision for craniofacial procedure



Le Fort procedures



Le Fort III



Le Fort I

Your child may need a procedure called a midfacial advancement (Le Fort III procedure). This operation moves the middle part of the face forward to open the airway and protect the prominent eyes. We use a technique called distraction osteogenesis to slowly bring the midfacial bones forward and to provide the most stable results. The timing of this operation depends on the specific needs of your child.

Will my child need further craniofacial operations as he or she ages?

When your child has finished growing he or she may also need an upper jaw (maxillary) advancement (also called a Le Fort I procedure). This helps to correct the way the teeth come together. Also, many children benefit from improvement in the appearance of the forehead in adulthood.

Dental treatment

What dental concerns are associated with Apert syndrome?

The abnormal shape of the face can also affect the position of the teeth and, in particular, the way the upper and lower teeth come together (called “occlusion”). Many children have an underbite, as well as misaligned teeth. Delayed tooth eruption is also common for children with Apert syndrome. Children should visit a pediatric dentist when the teeth start to come in—no later than 2 to 3 years of age.

What is the typical treatment approach for correcting the teeth and bite?

The decision to treat occlusion should be made with input from a pediatric dentist and orthodontist experienced in treating children with craniofacial disorders. In addition, a treatment plan should be created early to avoid unnecessary tooth loss. Removal of primary teeth may be needed to allow for more normal positioning of incoming adult teeth.

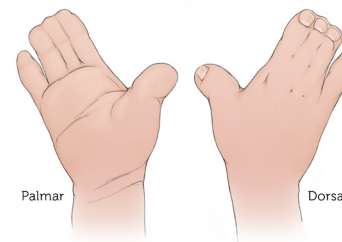
Your child will likely require one or more phases of jaw surgery combined with orthodontic therapy to help correct the bite and improve jaw appearance and function.

Hands and feet treatment

What hand and foot problems are associated with Apert syndrome?

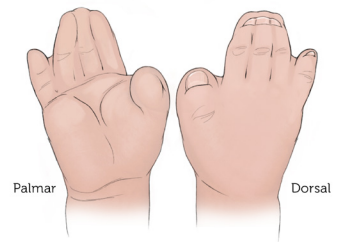
All children with Apert syndrome have some degree of fusion of the fingers and toes, a condition called syndactyly. The hands of patients with Apert syndrome can be classified into three categories (Upton classification) based on the degree of soft tissue and bone involvement. For most children with Apert syndrome, multiple procedures will be required throughout childhood.

Upton classification



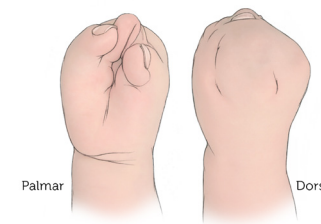
Type I

The index, middle and ring fingers are fused, but the thumb is free. The palm of the hand is flat. This type is commonly referred to as “spade” hand.



Type II

The thumb is fused to the index finger, in addition to the fusion of the three middle fingers. The palm of the hand is concave. This type is commonly referred to as “mitten” hand.



Type III

All digits of the hand are fused, with one nail. The thumb of the hand is turned inward, and it is often difficult to tell the digits apart. This type is commonly referred to as “rosebud” hand.

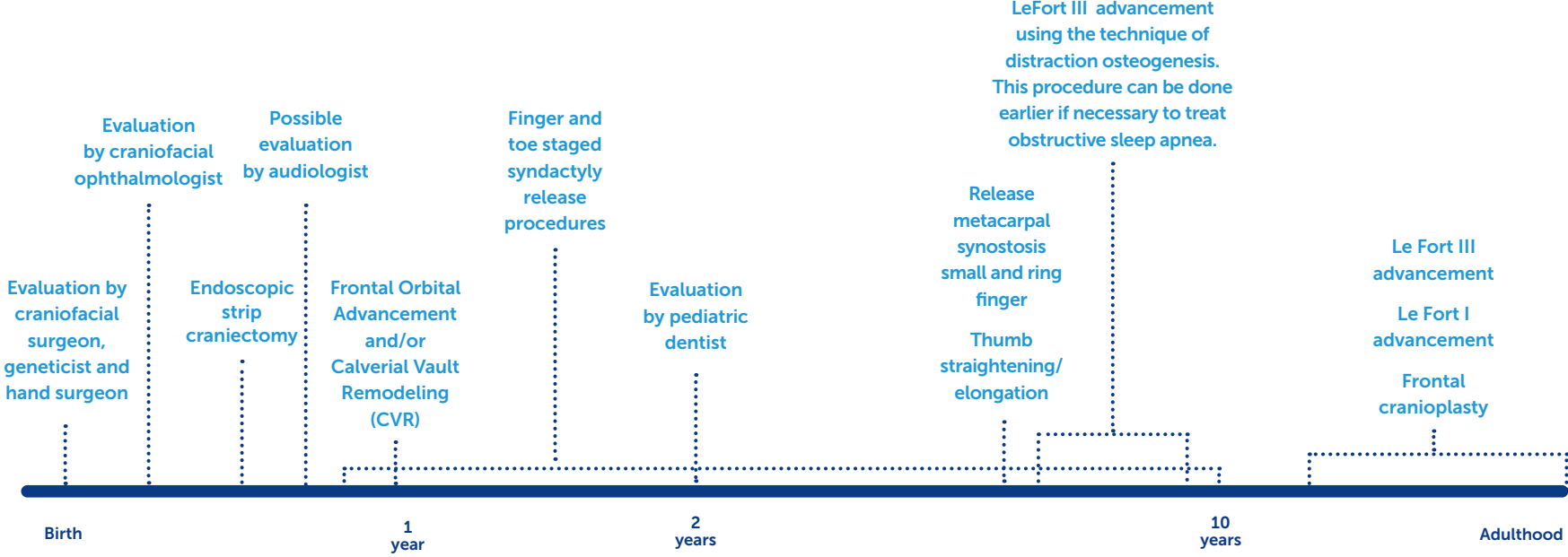
What is the typical time schedule for treatment of hand malformations?

If your child's thumb is involved repair will begin between 6 to 9 months of age to obtain optimal function. If the thumb is not involved, the other fingers will be separated between 9 months and 2 years. Hand surgeons are usually able to separate the fingers to give most children four fingers and a thumb, even in the most severe and challenging cases. This requires multiple complex operations, many of which involve separating and straightening fingers. Children with Apert syndrome have fewer normal functioning joints in their fingers, so they will likely not develop fine object manipulation skills. Nevertheless, these children function like most other children and do very well in life.

What is the typical treatment approach for fused toes?

Toes that are fused have very little effect on a child's ability to walk or run. You may, however, want to have your child's toes separated to improve the appearance of the feet to help them feel less self-conscious. The separation of fused toes can be often performed in conjunction with the release of fused fingers. As children grow they can also develop prominent bones in the feet that may cause difficulty with walking—these issues may also need to be addressed operatively.

Timeline of treatment



Glossary

Apert syndrome: rare complex genetic disorder that may affect a child's face, skull, teeth, hands, and feet. It is caused by a mutation in the FGFR2 gene.

Calvarial (or "cranial") vault remodeling: procedure used to release prematurely fused sutures, expand, and reshape the skull

Cranioplasty: surgical repair of a defect or deformity of the skull

Craniosynostosis: premature closure of the growth plates of the head, resulting in deformity of the skull and possible increased pressure on the brain

Distraction osteogenesis: procedure where a division is created in bone and then a medical device is used to slowly move the cut edges apart, inducing new bone formation, and gradually lengthening the bone

Endoscopic strip craniectomy: minimally invasive procedure to remove fused cranial sutures and provide the brain with necessary space for expansion.

Exorbitism: shallow eye sockets that cause the eyes to protrude. This is related to craniosynostosis and midfacial hypoplasia.

Fronto-orbital advancement: procedure where the forehead and supra-orbital rim (the bone deep to the eyebrow) is moved forward to make room for the brain and reshape the skull

Maxillary ("Le Fort I") advancement: operation to move the upper jaw forward to correct the way the teeth come together

Midfacial ("Le Fort III") advancement: operation to move the middle part of the face forward, opening up the airway, and protecting prominent eyes

Midfacial hypoplasia: decreased growth of the central face, causing a sunken facial appearance and potential airway obstruction and/or sleep apnea

Proptosis: protruding, unprotected eyes

Occlusion: relationship between the upper and lower teeth

Syndactyly: fusion of the fingers and/or toes

Sleep Apnea: disorder characterized by pauses in breathing or abnormally shallow breathing during sleep

Tracheostomy: surgical procedure to create an opening between the skin of the neck and the airway to facilitate breathing.

Upton classifications: categories describing the degree of soft tissue and skeletal involvement in Apert patient hands

- **Spade hand (type I):** index, middle finger, and ring finger are fused, but the thumb is free and the palm is flat
- **Mitten hand (type II):** thumb is fused to the index finger, the three middle fingers are fused, and the hand is concave
- **Rosebud hand (type III):** all digits are fused with one common nail, the thumb is turned inward, and it is difficult to tell the digits apart

Insurance and resources at Boston Children's

Insurance information

Children with Apert syndrome require long-term care, so understanding your insurance plan is important. Ask your individual insurance provider what you can expect for coverage and what your out-of-pocket expenses will be in the short- and long-term.

You should also understand the referral and coverage process your insurer requires with regards to treatment of the teeth and jaws, which may be considered dental and not medical in nature. Please enroll your child in your dental insurance plan, as dental procedures are typically not covered by medical insurance.

If you have further questions, contact your insurance provider for assistance. Most providers list a customer service phone number on the back of the insurance card.

Resources at Boston Children's

Craniofacial Anomalies Program

617-355-6309

bostonchildrens.org/craniofacial

Center for Families

617-355-6279

The Center for Families helps families locate the information and resources they need to better understand their child's particular condition and take part in their care. All Boston Children's patients, families and health professionals are welcome to use the center's services at no extra cost.

Additional resources and support

AmeriFace

ameriface.org

AmeriFace provides information and emotional support to people with facial differences and their families.

Apert International Inc.

apert-international.org

The Annual Apert Conference is open to families who have been affected by Apert syndrome or any other similar craniofacial difference. The organization's website connects families to an online network that provides support resources.

FACES: The National Craniofacial Association

faces-cranio.org

The association provides financial support for non-medical expenses to patients traveling to a craniofacial center for treatment. Eligibility is based on financial and medical need.

Children's Craniofacial Association

ccakids.org

This is a national organization that addresses medical, psychosocial, emotional, financial, and educational concerns and provides support for individuals with craniofacial anomalies.

Foundation for Faces of Children

facesofchildren.org

This is a New England-based organization providing clear, accurate information and other resources to children born with craniofacial conditions and their families. It was started by Boston Children's Hospital plastic surgeon John B. Mulliken, MD, and a group of parents whose children were receiving treatment at Boston Children's Hospital.

Facebook groups and pages

Apert USA

Apert Syndrome

Apert Owl

What parents have to say:

"The care our daughter receives at Boston Children's Hospital has exceeded our expectations. After a bad experience at another hospital, we quickly learned the importance of having doctors with experience with Apert syndrome care for our daughter. We love the way Boston Children's treats her as an individual and how decisions regarding care are based on her needs."

–Tara Lafleur, mother of Giuliana Lafleur

"The day we started going to BCH was truly the greatest day of my family's collective lives. The folks at BCH on our team are always one step ahead of me. BCH saved my son's life and future and for that we are eternally grateful. BCH is two hours from where we live, but I wouldn't go to another facility ever! We use them for everything because they are truly amazing."

–Jeremy Gold, father of Jackson Gold

"A family shouldn't be discouraged if they live far away from Boston and want to receive treatment here. Boston Children's Hospital works closely with our local doctors, and the communication is excellent. We travel to Boston for her surgeries and get testing done in our hometown as needed. The approach to our daughter's care is a team effort with a unique treatment plan for her needs. I feel very comfortable with BCH and believe the doctors treat our child the way they would treat their own."

–Tambra Milot, mother of Madilynn Milot

To make an appointment:

bostonchildrens.org/craniofacial

617-355-6309



Where the world comes for answers