What is Craniosynostosis?

Craniosynostosis is defined as the premature closure of the cranial sutures (what some people refer to as soft spots). This results in restricted and abnormal growth of the head. This restriction is frequently associated with eye and ear problems. Occasionally, defects with hands, feet, mouth, spine, heart, and kidneys are also seen. In some cases, this condition is part of a genetic syndrome. What this means is that the patient will have a certain appearance, which allows a geneticist to classify the constellation of problems. Recent advances in molecular biology around the country have determined that in many of these syndromes there are defects in specific regions of the patients DNA, which are associated with this problem An example is a defect in a growth factor receptor in the developing head called the fibroblast growth factor receptor (FGFR). There are several subclasses of this receptor that, when altered, lead to craniosynostosis syndromes such as Apert's and Crouzon's.

Our team is one of the few in the country specializing in Endoscopic Craniosynostosis repair. This minimally invasive surgery is used for help correcting several types of craniosynostosis with less scarring and without the need for metal plates and screws.

Dr. Stelnicki also performs Spring Skull reconstruction and Distraction Osteogenesis of the skull on selected patients. The springs can be placed in the skull, prior to one year in life, for the treatment of sagittal craniosynostosis. Springs are less invasive than open cranial vault remodeling, but more invasive than the endoscopic repair. They can be used on the patient who is older than 5 months of age, but less than one year old. The springs expand the elongated scaphocephalic skull slowly as the child grows. (see figure) These are placed through a relatively small incision with little skull dissection at the time of surgery. They are left in place for several months and then removed once the child has had correction of the scaphocephaly. No helmets are required for this therapy, making it useful for families who cannot come in frequently for helmet adjustments. Ask Dr. Stelnicki if this operation is the right one for your child.

Dr. Stelnicki also uses distraction Osteogenesis of the Skull in the treatment of craniosynotosis. During infancy, Dr. Stelnicki will utilize skull distraction when necessary for the treatment of coronal, metopic, and syndromal craniosynostosis as an adjunct to the endoscopic repair. The use of distraction osteogenesis at the time of endoscopic surgery involves the placement of a specialized distractor.

Endoscopic Cranial Vault Remodeling

Dr. Stelnicki is the only surgeon in Florida routinely performing endoscopic craniosynostosis repair. In addition, our center is only one of a few in the entire country to perform this technique on a variety of craniosynostosis types. Our practice

was recently honored by being able to present the first series of endoscopic craniosynostosis repair vs. traditional repair. This paper, which given at the prestigious International Craniofacial Society meeting in Monterey, California was the first to carefully analyze the advantages of endoscopic repair over the traditional open technique. This new technique for treatment of craniosynostosis is minimally invasive. It creates incisions smaller than 2 cm in length that are used to access the fused and deformed skull. A specialized set of endoscopic instruments designed by Dr. Stelnicki, are then inserted through these holes and used to completely reshape of the skull without performing a large ear-to-ear incision.

The advantages to endoscopic surgery are many. The surgery has less blood loss than the traditional repair and, in many cases, prevents the need for blood transfusion during the surgery. The endoscopic technique also results in less postoperative swelling, decreases operative time, and decreases hospital stay by several days. Postoperative pain is also significantly reduced by this operation. The other advantage to endoscopic surgery is that there are no internal screws or plates that are required to hold the bone in place after surgery. This decreases the risk of plate induced pain, infection, swelling, or allergic reaction in your child. Dr. Stelnicki would be glad to discuss the advantages of endoscopic craniosynostosis surgery versus the classic craniosynostosis repair with you at time of the initial consultation.

Endoscopic and surgical correction is available for any patient, under 5 months of age, with a craniosynostosis. Initially endoscopic craniosynostosis corrections were restricted to single suture fusions, such as sagittal craniosynostosis, coronal craniosynostosis, metopic craniosynostosis, or lamdoid craniosynostosis. However, we now know that is also a useful adjuvant in the treatment of multi-suture craniosynostosis. When possible, the endoscopic approach is used in the first stage of these patient's repairs.

Endoscopic craniosynostosis repair must be combined with the use of a molding band in order to normalize skull shape after the surgery. Immediately after the surgery, you will note a significant improvement in your child's head. However, the final shape will not been seen until after 6-8 months of helmet therapy. The corrective helmets used in helmet therapy are FDA approved devices for molding and protecting the developing head. They have been used on multiple patients throughout the country following endoscopic craniosynostosis repair.

Dr. Stelnicki is also a member of an elite set of surgeons who have designed & created their own endoscopic craniosynostosis instruments. These tools, which bear our name and are sold by KLS Martin, are now available to any surgeon around the world for the treatment of this condition. Our research has helped in the creation of these instruments and will aid in developing even newer instruments to better treat children with craniosynostosis in a minimally invasive fashion.

Multisutural Craniosynostosis and related syndromes:

- Apert's syndrome
- Crouzon's syndrome
- Pfeiffer's syndrome
- Saethre Chotzen syndrome
- Other forms of syndromic craniosynostosis.

Although most forms of craniosynostosis are sporadic and affect no other part of the developing infant, there are some rare forms of craniosynostosis that are syndromic. These patients have a very characteristic appearance. Their craniosynostosis usually involves multiple sutures, and in every case, more than just the shape of the skull is affected. These patients should always be treated by a craniofacial team, as their syndrome involves multiple regions of the body. Without a good team approach of addressing each problem in an organized way, the likelihood of a good treatment result is decreased.

Apert's Syndrome:
Apert's syndrome is an autosomal dominant form of congenital craniosynostosis. It is thought to result from a mutation in a Fibroblast Growth Factor Receptor (FGFR I-IV) that is essential for proper head and extremity development.

Patients with Apert's syndrome typically have premature fusion (craniosynostosis) of multiple cranial sutures. The ones most commonly affected are the coronal sutures that extend from ear to ear. As a result of this fusion, the head of these patients is shortened from front to back (brachycephalic) and elongated from top to bottom (turricephalic). The eyes of these patients appear to "bulge out" due to the fact that their skull base and mid-face fails to grow in a normal fashion. The palate of these patients is typically high arched and narrows. Dental eruption is typically delayed.

Apert's children also always have some degree of hand and foot webbing called polysyndactyly. This can be of a minor type that only requires skin separation, or a more severe type, where there is actual fusion of bone within the hands and feet.

Most Apert's children have normal intelligence. However, varying degrees of mental retardation have been seen with this condition.Effected areas in Apert's syndrome include:
Cranial sutures
Skull growth
Skull base
Maxilla
Cleft palate
Eye movement
Orbit position
Dental development is delayed.

Webbed hands and wrist problems
Webbed toes and foot problems
Speech abnormalities
Mental development
Psychological development
Chromosomal changes
Crouzon's Syndrome:
Patients with Crouzon's Syndrome have a similar facial appearance to those with Apert's syndrome. However, there are several important distinctions. Crouzon's patients seldom have macrocephaly, in spite of the fact that they have multiple cranial suture fusions. They have a variable degree of midfacial deficiency and exorbitism. Their hands are completely normal. Their mental development is variable. And their teeth tend not to have the delayed eruption seen in patients with Apert's syndrome.

Like Apert's, children with Crouzon's typically have a genetic mutation in the FGFR gene. Mutations in FGFR I-IV have been seen, but FGFR I seem to be the most common. The phenotype of children with Crouzon's is highly variable. In some instances, only mild brachycephaly and exorbitism are seen. In other cases, sever brachy-turricephaly is evident and the eyes are wide set (hypertelorism).

Pfeiffer's Syndrome:
The patient with Pfeiffer's syndrome is similar to the Apert's and Crouzon's patient. These patients also have a defect in the FGFR gene, which lead to a multi-suture craniosynostosis. However, their midface is usually more severely effected. In many cases, these patients will have severe exorbitism that puts them at risk for corneal exposure and damage. A full ophthalmologic evaluation by an experienced pediatric ophthalmologist is needed, and in some cases these patients need the lateral aspect of their eyelids sewn together to provide adequate protection for the eye (tarsorhaphy). Patients with Pfeiffer's syndrome also have thumb and great toe anomalies, which occasionally have to be treated by an experienced hand or foot surgeon. All Pfeiffer's patients seen by our team are carefully evaluated by our pediatric hand surgeon and podiatry staff to ensure that proper growth is occurring in these areas.

There are several other syndromes involving craniosynostosis. Whether it is Saethre-Chotzen (a mutation in the Twist gene), Carpenters syndrome, or another rare form of early suture closure, our team carefully evaluates each patients to make sure that all their medical and developmental needs are met.

Treatment of Syndromic Craniosynostosis Patients:

The treatment of these syndromes begins at birth. Because the midface fails to grow, Apert's patients may have problems breathing. As a result, they should be watched closely at birth for signs of apnea or desaturations. If the breathing problem is severe,

early intervention is required. Our nurse practitioner/feeding specialist works closely with each child to ensure that nutrition is adequate. Our staff geneticist performs a full genetic evaluation, including FGFR receptor analysis to complete the diagnosis.

The child is then evaluated carefully by our pediatric neurosurgeon for signs of hydrocephalus or increased intracranial pressure. Based on this evaluation, plans are made to begin the skull remodeling surgery. Our goal is to normalize skull shape by the patient's first birthday. This is done by Dr. Eric Stelnicki working closely with one of the pediatric neurosurgeons. Care is taken to sequentially mold the front and back of the misshapen head, while decreasing intracranial pressure and allowing room for growth.

If a cleft palate is present, this is typically closed between 12-18 months of age. The decision to close the cleft is based on a close consultation between the Craniomaxillofacial surgeons, the team of speech therapists, the orthodontic staff, and the patient's family. The goal of this surgery is to close the hole in the roof of the mouth, while maximizing speech development and maxillary growth.

Treatment of the hands and feet begins as early as 2-3 months of age. Our team hand specialists are dedicated hand surgeons who specialize in the treatment of pediatric hand abnormalities. The type of surgery performed is based on the type of hand abnormality present in the patient. Severe deformities usually begin with a separation of the thumb so that the infant can hold objects. We also have hand therapists who will work with the child to maximize their function thoughout their development. Our team is one of the few in the country, which recognizes the importance of treating feet abnormalities in these patients. The syndactyly of the toes is treated by trained orthopedic and podiatric surgeons who understand the need for proper foot care and development. These are typically done in conjunction with the hand surgeries in order to decrease the total number of operations a patient with Apert's has during their lifetime. Other hand and foot abnormalities are addressed and treated by the team in an organized, staged fashion that optimizes form and function.

A Team Approach for the Continuum of Care

Speech development is carefully followed by our speech therapist. Early intervention is given as needed to get the best results. Occasionally speech surgery is required to normalize the production of sound. However, this is only utilized as a last resort when conservative measures have not been successful.

Dental development is followed closely by our pedodontic and orthodontic teams. Treatment is adjusted based on the pattern of dental growth and the type orthodontic care needed to normalize tooth and gum position. Children with these syndromes frequently require a surgery to put the underdeveloped "mid-face" into a proper position. This is performed at age 5 using the newest, less invasive methods of distraction osteogenesis when possible. This technique maximizes facial position with the least amount of risk.

A careful eye exam is given at birth. Corneal exposure problems are treated immediately. Problems with eye muscle movement (strabismus) are treated by our pediatric ophthalmology specialist in order to normalize eye position and visual development. In patients with very wide set eyes (orbital hypertelorism) the position of the eyes is normalized at 4 years of age. This is done in conjunction with Dr. Stelnicki, the pediatric neurosurgeon, and the ophthalmologist in order to get the best final result under the safest conditions.

In the teen-age years, nasal and jaw deformities are again addressed by the team. Patients with these syndromes usually require orthognathic surgery to correct jaw position. This is done in conjunction with our orthodontic staff, which works closely with the patient to maximize dental growth and development. Any additional nasal surgery is also performed at this time. Recently, we have been utilizing virtual reality techniques to plan our operations at this stage. A CT scan of the patient's facial bones and skull is compared to a "normal" template. Based on this comparison, the facial bones are segmented and moved into their proper location. All of which significantly normalizes facial appearance.