Craniosynostosis: A Case Report

Abstract

Craniosynostosis is the premature fusion of cranial sutures which can result in neurological abnormalities, facial and skull deformations and increased intracranial pressure. It can be diagnosed by physical examination, the use of plain film x-rays, CT exams, or with the use of ultrasound. If caught within the first few months of life, or prenatally, craniosynostosis can be treated endoscopically with minimal invasiveness. Those cases that are discovered after the first 6 months of life can be treated surgically with cranial vault remodeling. Both procedures have an excellent success rate and a low recurrence rate. Craniosynostosis can be an isolated occurrence or it can be part of a syndrome. There are several different types of craniosynostosis dependant on the suture, or multiple sutures, that have fused; each with a unique presentation of the skull cap and facial bones.

Introduction

Craniosynostosis is the fusion of cranial sutures before brain growth is complete. This results in the abnormal growth of the skull and can cause multiple deformities in the shaping of the head and face. It is estimated to affect one in every 2000 children born.\(^1\) Growth of the brain is continually stimulating the bones in the skull to expand and create new bone at the suture lines. This continues to happen until around the fourth decade of life, but is most valuable in the first year when brain growth quadruples. The normal range for infant fontanelles to fuse is from 4 to 26 months, with semi permanent fusing by the age of 12, and permanent fusing occurring by age 40 or 50.\(^1\) Craniosynostosis is usually present at birth but may not be diagnosed until the early months of the child’s life.\(^1\)
Fusion typically involves one or more of the five major infant sutures which are: metopic, coronal, sagittal, lambdoidal, and squamosal (see Figure 1). The sagittal suture is most commonly fused, followed by the coronal, metopic, and lambdoidal, which is the most rare.\textsuperscript{1,3,4} It can be a single diagnosis, or part of a more complex syndrome such as Apert, Crouzon, Carpenter, Pfifer, with other symptoms including delayed brain growth and trunk deformities.\textsuperscript{5} More than 200 types of syndromes involve craniosynostosis.\textsuperscript{4}

The cause of craniosynostosis is for the most part is unknown. Cases can be classified as either primary, which is an intrinsic defect in suture or secondary, which is “premature closure of normal sutures because of another medical condition such as deficient brain growth.”\textsuperscript{3} (p.1)

Primary craniosynostosis, from an etiologic view, can either be idiopathic or familial.\textsuperscript{3} Familial cases usually present as a type of craniofacial syndrome and can be the result of a genetic mutation.\textsuperscript{3} These known mutations happen in the fibroblast growth factor receptors, which modulate cell proliferation, differentiation and migration.”\textsuperscript{3} These mutations are associated with Pfeiffer, Apert, Crouzon, Beare-Stevenson, Jackson-Weiss and Muenke syndromes.\textsuperscript{3,6}

Secondary craniosynostosis is developed from a known underlying disorder. These disorders can be systemic and metabolic and include conditions such as “hyperthyroidism, hypercalcemia, hypophosphatasia, vitamin D deficiency, renal osteodystrophy, Hurler’s Syndrome, sickle cell disease and thalassemia.”\textsuperscript{3} (p.1)

**Diagnosis**

Children are often referred to a pediatric neurosurgeon for an abnormally shaped head or head deformity. Diagnosis of craniosynostosis is done by physical examination. The child’s head is examined and the suture lines are palpated. The presence of a ridge where the suture lines are
located is a sign of craniosynostosis, as this is not present with normal sutures. The physician will also check for symmetry in the shape of the head, also the location of the ears on the skull and symmetry between the eyes. The shape and slope of the forehead are also noted, as well as the size and shape of the fontanelles. The physician may ask if the child’s milestones are being met, and pay special attention to the child’s alertness. The physician will also pay special attention to the hands and feet for congenital anomalies which can represent several syndromes associated with craniosynostosis.

There are several diagnostic tests that can be performed to definitively diagnose craniosynostosis, which include x-rays, a computed tomography (CT) exam, and ultrasound. Most often a three dimensional (3D) CT exam is done to assess the severity and exact location of the fusions, and if there is viscerocraniam involved. A 3D exam will highlight the skull features, the location of the sutures, and if there are any other deformities present. It can demonstrate the relationships of the bony structure of the skull and help preoperatively in planning the exact location for osteotomies. With the use of CT the resulting shape of the head is known when the specific fused suture has been identified.

The use of ultrasound is becoming more prevalent in the diagnosing of craniosynostosis prenatally. In a case study presented at the International Society of Ultrasound in Obstetrics and Gynecology World Congress, the ultrasounds of patients that were diagnosed postnatally with craniosynostosis were examined and were found to have observable brain shadowing. This was evident when the sutures were fully closed and even partly closed. This was performed without the use of high definition 3D transducers. Usually diagnosis of fetal craniosynostosis is difficult due to the nature of ultrasound exams, especially if there is no family history or it includes a sagittal fusion. However a brain shadowing sign is easy to identify without having to have a
specific fetal position. The disadvantages to ultrasound are that craniosynostosis usually cannot be diagnosed until the second or third trimester. Ultrasound has advantages such as no radiation dose and early detection; however, CT is still the standard of care because of the accuracy of the scans.

Types of Craniosynostosis

Dolichocephaly and scaphocephaly

Dolichocephaly and scaphocephaly are synostoses that involve the sagittal suture of the skull cap, and presents in an anterior-posterior elongated skull. This fusion is more prevalent in males than it is in females, and any neurological or elevated intracranial pressures are unlikely. Scaphocephaly is known to form distinct ridge on the top of the head, and usually presents with frontal and/or occipital bossing (see Figure 2).

Brachycephaly and anterior plagiocephaly

Brachycephaly and anterior plagiocephaly are the fusion of bicoronal or unicoronal sutures. It is more prevalent in females and results in biparietal growth. “Bilateral coronal synostosis results in a prominent frontal bone, flattened occiput and anterior displacement of the vertex.” (see Figure 3) There are secondary facial deformities that are due to underdeveloped supraorbital ridges creating oblong, elliptical orbits. Unicoronal synostosis presents as a flattened frontal bone on the fused side, and an enlarged frontal eminence on the unfused side (see Figure 4).
Turricephaly and posterior plagiocephaly are bilateral and unilateral fusions of the lambdoidal suture; they are rare and can often be mistaken for positional plagiocephaly which occurs postnatally as the result of infants spending too much time positioned on their backs. With bilateral lambdoidal fusions the head presents as a *tall cranium* with growth continuing upward and the posterior fossa being underdeveloped (see Figure 5). ³ This type of fusion is often associated with syndromes that do not occur with other fusions. Turricephaly and unilateral lambdoid synostosis presents as posterior plagiocephaly.

**Trigonocephaly**

Trigonocephaly is the fusion of the metopic fusion (see Figure 6). This presents in a “triangular, pointed forehead with flattened frontal bones and bossing of the parieto-occipital regions.”³⁽³⁾ This form of synostosis is often associated with a syndrome and includes other midline anomalies with the brain and palate. Recently there have been cases of trigonecephaly which are more mild and are termed *metopic ridge*, this condition is usually not surgically treated.³

**Oxycephaly**

Oxycephaly is the severe fusion of the sagittal and coronal sutures. This is the most severe of the synostosis conditions and is often associated with neurologic impairments and raised intracranial pressure.³ These fusions present as a variety of shortening and bossing of the skull depending on where the fusions have occurred (see Figure 7).

**Kleeblattschadel**
Kleeblattschadel or cloverleaf synostosis is associated with a combined sagittal, coronal, and lambdoid suture fusion. This condition is severe and often patients experience severe neurological impairment. This form of stenosis presents with the skull taking a clover-like shape with bossing in the temporal and parietal bones (see Figure 8).

Treatment

Surgery for patients with primary craniosynostosis is needed to avoid a range of future problems from body image insecurities and other cosmetic reasons to increased intracranial pressure. The age range for successful surgery is 9-12 months and a secondary surgery need for primary craniosynostosis is rare. The surgery consists of “a combined intracranial and extracranial approach” in which a “bi-frontal craniotomy is performed.” The skull pieces are then reshaped by segmenting and reforming the skull; correcting any bossing or narrowing that may have existed and allowing for future growth of the brain. The incisions are large and usually span from one ear to the next (see Figure 9). The surgery is extensive and can last up to eight hours. Complications include facial swelling, blood loss, as the patients are small and have little reserve they often have to undergo a blood transfusion, and difficulty in maintaining an airway, which can be more problematic with syndromic craniosynostosis cases. The patient is typically then admitted to the Pediatric Intensive Care unit for a night, and then moved to a pediatric floor for an additional 3-5 nights.

The use of endoscopic treatment is also an option for some infants who qualify. The infant is usually between the ages of 3 to 6 months and their deformity is less complex. With this technique the surgeon makes several small incisions and is able to endoscopically resect the closed suture, and insert an absorbable plate (see Figure 10). The patient then wears a helmet to
further reshape the skull for 6 to 8 months after surgery. This surgery is a lot less invasive and involves little blood loss, almost completely eliminating the need for a blood transfusion. The patient typically spends one night in the hospital and then is allowed to go home.

**Prognosis**

The reconstructive surgery for primary craniosynostosis has a very good prognosis with no seen cognitive problems and a need for a second surgery is often very rare. The endoscopic surgery also has a very high success rate and although the recurrence rate is slightly higher than the cranial vault reconstruction surgery it is still very low. A diagnosis of craniosynostosis which is part of a known syndrome has a higher incidence of a repeat surgery needed.

**Case Study**

Davutoglu et al discuss the case of a child that presents with a narrow shaped forehead that is small and triangular in appearance. The patient had been referred to the clinic for microcephaly, and was diagnosed with both anterior and posterior fused fontanelles. The child’s blood work and gestational period were unremarkable, but softness of the head was noted when palpated, along with a low weight percentage. A cranial CT revealed there were several thinned areas of the cranial vault, but could not accurately portray the sutures that were fused. “The patient underwent thin-slice high resolution cranial CT examination using multislice CT scanning and reconstruction image were obtained for three dimensional analysis.” The use of this 3D scan revealed that there were multiple fusions; including a fusion of the metopic suture, an almost complete fusion of the sagittal suture, and fusions of both coronal sutures. It also revealed “multiple exaggerated convolutional marking in the form of lacunar skull…"
The cranial CT scan had not been able to visualize the fusions of both the coronal sutures and the lambdoidal suture could not be evaluated. The more complete diagnosis of compound craniosynostosis was given due to the findings of the 3D CT scan.

**Conclusion**

Craniosynostosis is a fairly prevalent disorder, occurring in 1 out of every 2000 births. It consists of the premature fusion of 1 or several cranial sutures, resulting in misshapen skull and facial bones and the possibility of impeding on the growth and natural shaping of the brain. The fusion occurs in the fetus and usually isn’t diagnosed until the first few months of life. If the disorder is suspected early, an ultrasound can be utilized to view the suture lines and evaluate the brain shadowing even prenatally. This allows the patient a lower dose than an x-ray or CT exam and if found prenatally can allow the family time to prepare. If the disorder is found after birth, the standard of care is a 3D CT exam. This allows the surgeons to know the exact location and extent of the fusions. This exam can also be helpful in planning the surgical vault remodeling.

There are several types of craniosynostosis which depend on the suture or sutures that are involved. They are: Dolichocephaly, Scaphocephaly, Brachycephaly, Anterior Plagiocephaly, Turricephaly, Posterior Plagiocephaly, Trigonecephaly, Oxycephaly, and Kleeblattschadel. These types of craniosynostosis can be isolated or can be part of secondary to a known syndrome.

The treatments that are available for craniosynostosis are both highly effective. If the diagnosis is early in life, an endoscopic option is available which offers minimal invasiveness, little bleeding, quick recovery, and little scaring. If the diagnosis is made later in life, the surgical option is a cranial vault reconstruction. This can involve bleeding, swelling, highly invasive, and
a sizeable amount of scarring. Both options offer a good prognosis with excellent success rates and rarely a need for a second procedure. These options make craniosynostosis when known as a primary deformity completely recoverable.

References


**Figures and Captions**
Figure 1. Normal sutures. 3DCT volume rendered images. Vertex (A) and lateral (B) views. (a) Metopic suture; (b) coronal sutures; (c) sagittal suture; (d) lambdoid suture; (e) squamosal suture; (f) anterior fontanel; (g) posterior fontanel; (h) sphenoidal fontanel; (i) mastoid fontanel. Cranial vault bones usually ossify from the center to periphery, which results in this “widened” appearance of the sutures in the newborn. Endocranial skull base view (C) shows portions of the occipital bone and sutures: (j) Basioccipital; (k) paired exoccipital; (l) supraoccipital; and (m) interparietal. Associated synchondroses are (n) spheno-occipital; (o) anterior intra-occipital; (p) posterior intra-occipital; (q) petro-occipital; (r) occipitomastoid; (s) and mendosal sutures. Note that o, k, p and s are paired structures. Vertex view (D) shows the lambda (point of intersection of the sagittal and lambdoid sutures) and bregma (point of intersection of the coronal and sagittal sutures. Endocranial skull base view (E) shows the basion (located on the basiocciput, at the midpoint of the anterior margin of the foramen magnum) and opisthion (located on the occipital bone, at the midpoint of the posterior margin of the foramen magnum). Image courtesy of Khanna P, Thapa M, Iyer R, Prasad S. Pictorial essay: the many faces of craniosynostosis. Indian J Radiol. 2011;21(1):1-9. doi:10.4103/0971-3026.76055.
**Figure 2.** Dolichocephaly and scaphocephaly. 3DCT volume rendered images. Left anterior (A), posterior (B) and endocranial (C) images show fusion and prominent ridging of the sagittal suture (white arrows) with frontal and occipital bossing (*). Also note the increased anterior-posterior dimension and restricted biparietal diameter. Image courtesy of Khanna P, Thapa M, Iyer R, Prasad S. Pictorial essay: the many faces of craniosynostosis. *Indian J Radiol.* 2011;21(1):1-9. doi:10.4103/0971-3026.76055.
Figure 3. Bilateral (A-D) and unilateral partial (E) coronal synostosis 3DCT volume rendered images (A-C, E) and coronal CT scan (D). There is complete fusion of the coronal suture (white arrows) with a prominent frontal bone and flattened occiput. Coronal reconstruction (D) demonstrates prominent bilateral elliptical orbits, known as the “harlequin eye” deformity. Note the early partial fusion of the right coronal suture (arrowheads in E). Image courtesy of Khanna P, Thapa M, Iyer R, Prasad S. Pictorial essay: the many faces of craniosynostosis. Indian J Radiol. 2011;21(1):1-9. doi:10.4103/0971-3026.76055.
**Figure 4.** Anterior plagiocephaly. Anterior (A), vertex (B) and endocranial (C) 3DCT volume rendered images show right unicoronal synostosis (white arrows) with ipsilateral frontal flattening and contralateral frontal prominence (*). An incompletely fused metopic suture is also seen (arrowhead), probably compensatory. Image courtesy of Khanna P, Thapa M, Iyer R, Prasad S. Pictorial essay: the many faces of craniosynostosis. *Indian J Radiol.* 2011;21(1):1-9. doi:10.4103/0971-3026.76055.
Figure 5. Turricephaly. Lateral (A) and posterior (B) 3DCT volume rendered images show turricephaly secondary to bilateral lambdoid fusion (arrows). Note the small, underdeveloped posterior fossa (*), and the “tall” cranium (double-headed arrow). Image courtesy of Khanna P, Thapa M, Iyer R, Prasad S. Pictorial essay: the many faces of craniosynostosis. Indian J Radiol. 2011;21(1):1-9. doi:10.4103/0971-3026.76055.