

CRANIOFACIAL SURGERY

C O M M E N T S

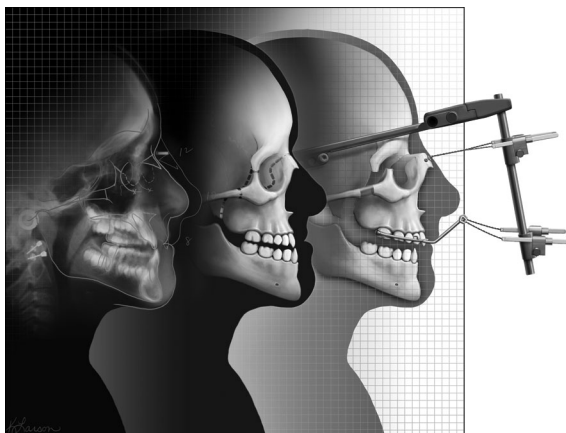
No single discipline has the expertise to fully treat children with a craniofacial anomaly. Consequently, such children require care from many different disciplines. Typically, surgical, medical, dental, and psychosocial specialists are involved. When a large number of professionals are involved in the multifaceted care of patients, healthcare teams develop. Cleft and craniofacial teams, which were formed in response to the complex, life-long, ongoing clinical and psychosocial needs of this patient population, are widely considered an effective means to avoid fragmentation and dehumanization in the delivery of such highly specialized health care.

Team care represents an improvement over splintered, community-based, multispecialty care. The benefits of healthcare delivery by the team approach include the abilities to provide interdisciplinary services, to address the emotional and psychological needs of patients and their family, and to perform multifaceted examinations that enable comprehensive treatment plans to be formed based on the team's recommendations and the family's preferences.

Since 1986 the Barrow Craniofacial Center (formerly the Southwest Craniofacial Center) has been treating children and adults with cleft and craniofacial disorders from Arizona and the Southwest through this essential interdisciplinary team approach. The team helps address parental concerns about the choice and timing of treatment and the coordination of ongoing care. In addition to providing these clinical craniofacial services, the team conducts clinical research and provides education for students of these disciplines.

This issue of the *Barrow Quarterly* reviews established principles of craniofacial care and presents new findings on the genetics of craniofacial deformities, new practices in orthodontic cleft care, and new techniques of distraction osteogenesis. The breadth of the material underscores the broad base of interdisciplinary knowledge and care required to care for patients with these complex deformities—care best provided at centers of excellence such as the Barrow Neurological Institute. We are proud to showcase the efforts of this dedicated team working with these most complicated cases. Please consider using the enclosed self-addressed stamped envelope to make a tax-deductible donation that will help us continue to share the exciting work performed at our institution. Thank you.

Stephen P. Beals, MD
Guest Editor



This issue's cover depicts the key steps to successful craniofacial surgery as they relate to a child with Crouzon syndrome. As a part of the planning process, the doctor traces the projected position of the mobilized bone. A Le Fort III osteotomy is performed to free the bone fragment that will undergo distraction osteogenesis. Once distraction is complete, the rigid fixation device is left to hold the face forward until it heals, obviating the need for bone grafts, plates and screws. See the article by Beals et al. on page 11. The illustration is by Kristen Larson.

Craniosynostosis Syndromes in the Genomic Era[†]

Kirk Aleck, MD

The origin of craniosynostosis is heterogeneous: hereditary, mechanical, teratogenic, and idiopathic. Craniosynostosis is further defined by the suture(s) involved and whether it is syndromic or nonsyndromic. Syndromic craniosynostosis typically involves cranial sutures plus changes in the central nervous system and extracranial skeleton. Nonsyndromic craniosynostosis is usually confined to cranial changes. The most common syndromic synostoses reflect changes in FGFR activity related to mutations in the genes coding for these receptors. Other genes have been implicated in other craniosynostosis syndromes. Several craniosynostosis syndromes are caused by mutation of the same FGFR making the eponymic designation (e.g., Crouzon or Pfeiffer syndrome) less certain. Ultimately, syndrome eponyms may be replaced by designation of the underlying mutation. Neurological complications can include mental retardation, increased intracranial pressure, and cranial nerve abnormalities. Craniosynostosis syndromes require careful physical examination, radiological investigation, and now molecular evaluation to predict outcomes and the risks of recurrence.

Key Words: cranial suture, craniosynostosis, fibroblast growth factor receptor

Abbreviations Used: FGFR, fibroblast growth factor receptor; IQ, intelligence quotient

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Craniosynostosis is the premature closure of a cranial suture or sutures, leading to alterations in head shape. It is a frequently encountered physical finding in children, affecting about 1 in 2500.¹¹ Etiologically and morphologically, craniosynostosis is heterogeneous, often necessitating careful clinical and radiological evaluation to determine its cause. Neurological complications can include increased intracranial pressure with decreased cerebral blood flow, abnormalities of hearing and vision, and intellectual impairment. This review explores the classification, biology, and diagnosis of craniosynostosis.

Origins of Craniosynostosis

There are five major causes of craniosynostosis. The first is premature fusion for unknown developmental reasons. The second is mechanical compaction (external pressure) and fusion of the suture. The third is premature fusion related to metabolic disorders or teratogenic agents. Fourth, the suture may close prematurely because of a lack of underlying brain growth that ordinarily causes the suture to remain separated. Finally, premature fusion may occur as the result of a genetic mutation.

Craniosynostosis is most often an isolated developmental abnormality with no known underlying mechanism. Craniosynostosis has also been reported in more than 100 different genetic syndromes⁷ in which the causative gene may or may not be known. Craniosynostosis is reported in a number of chromosomal disorders as well.⁵ Metabolic disorders include hyperthyroidism, rickets, hypercalcemia, and mucopolysaccharidoses; several hemato-

logical disorders account for a small percentage of craniosynostosis cases.⁵ Several teratogens have also been implicated as causes of craniosynostosis.⁵

Biology of Craniosynostosis

The cranial sutures are an example of the type of bony articulation known as a synarthrosis. In this type of articulation, bones almost contact one another but are separated by fibrous tissue and are relatively immovable. Sutures occur only in the cranium and in no other part of the body. Sutures are the sites of constant deposition and resorption, which allow the rapid changes in size and shape that accompany head and face growth. If a facial suture fails to develop or closes prematurely, compensatory changes in the surrounding bones occur.

The cranial bones originate as a series of ossification centers located in specific areas within the fibrous desmocranium that surrounds the fetal brain.⁵ During this period of rapid growth, the ossification develops and the underlying brain grows equally rapidly. When growth of the brain slows, these ossification centers form sutures at their margins. Eventually the sutures themselves are obliterated as the bones of the calvarium fuse. Appropriate timing of this fusion results in the normal skull shape. With the exception of the metopic suture, which fuses between 2 and 3 years of age,²¹ sutural closure is an adult phenomenon. The major sutures fuse in the third decade. Premature fusion occurs when a suture is obliterated by bone before it can adapt to the rapid growth.

Classification

A morphological classification of craniosynostosis refers to the suture or sutures that exhibit premature closure (Fig. 1). Bilateral coronal synostosis is most often associated with syndromic synostosis and results in a short from front to back (brachy) and tall (turri)-shaped head referred to as brachyturriccephaly. Unilateral coronal synostosis results in an asymmetric head shape referred to as plagiocephaly. Sagittal synostosis results in a long, thin

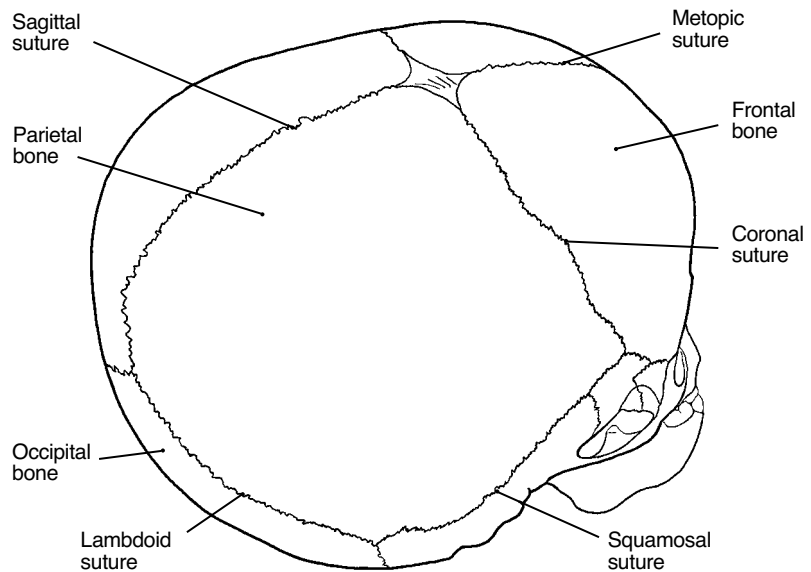


Figure 1. The bones and sutures of the cranium.

Table 1. Classification of Craniosynostosis

Primary craniosynostosis

- Primary syndromic synostosis with known mutations
- Primary syndromic synostosis without known mutations
- Nonsyndromic synostosis

Secondary craniosynostosis

- Mechanical etiology
- Metabolic etiology
- Early sutural closure due to failure of brain growth

head shape referred to as dolichocephaly or scaphocephaly. Lambdoidal synostosis also results in brachycephaly, but the major effect is on the posterior skull. Metopic synostosis creates a ridge on the center of the forehead that extends from the anterior fontanel to the forehead. It often results in a triangular-shaped head (trigonocephaly). Synostosis involving multiple sutures leads to tower skull (acrocephaly). Finally, multiple sutural synostosis with bulging through squamosal or sagittal sutures leads to a trilobed cloverleaf skull (Kleeblattschädel).

Craniosynostosis can be divided into primary and secondary synostosis (Table 1). Primary synostosis, whether as part

of a syndrome or on an isolated basis, is caused by an error in development. Primary craniosynostosis can be divided into syndromic craniosynostosis with known genetic mutations, syndromic synostosis without known genetic mutations, and nonsyndromic craniosynostosis. Secondary synostosis is due to mechanical or metabolic causes that alter a previously normal cranial developmental pattern. Secondary synostosis also is associated with early sutural closure related to failure of the brain to grow.

In the primary syndromes of craniosynostosis, the shape of the skull can be predicted by two general rules: The head will be shorter perpendicular to the closed

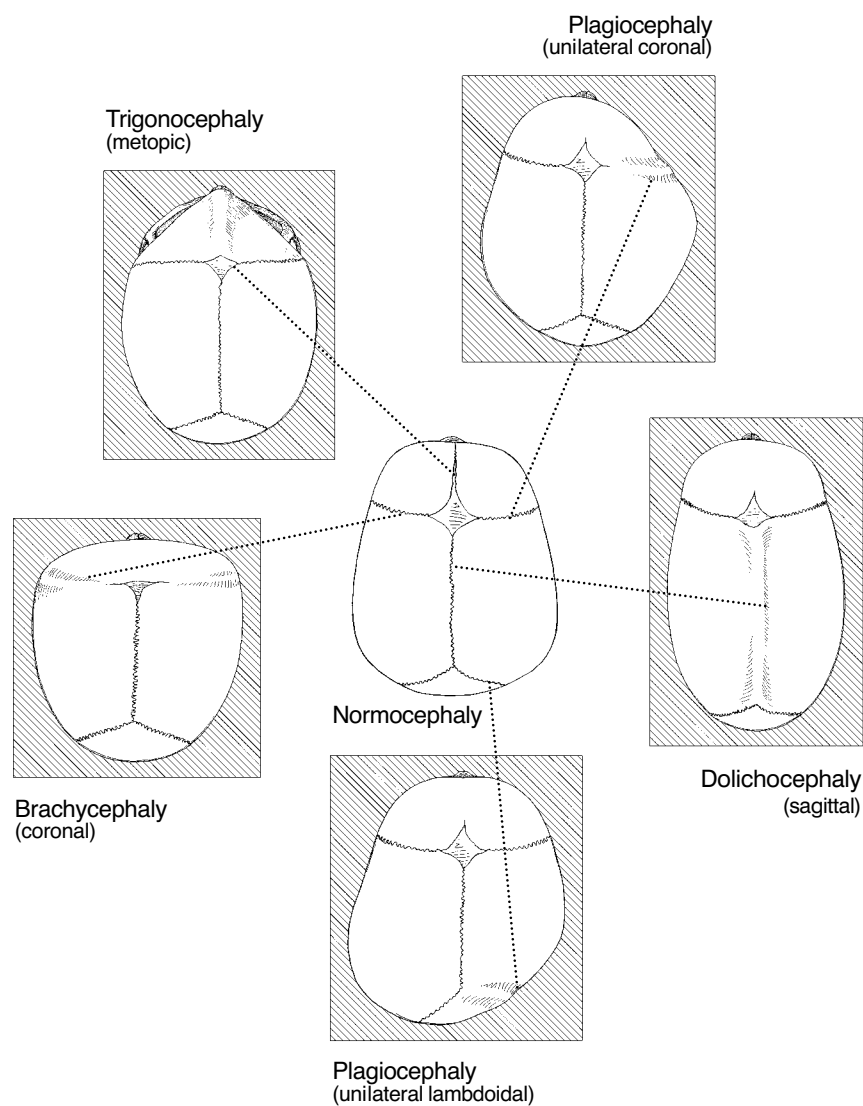


Figure 2. Morphologic effect of cranial suture synostoses.

suture (growth restriction occurs at right angles to the fused suture) and longer parallel to the closed suture (compensatory expansion in the same direction of the fused suture, Fig. 2).

Nonsyndromic Isolated Primary Craniosynostosis

Isolated Sagittal Synostosis. Isolated early fusion of the sagittal suture is the most common form, estimated at about 56% of all cases of isolated craniosynostosis.⁹ It results in scaphocephaly. It has a marked predilection for males. Its cause is unknown and it usually has no neurological sequelae. Treatment is surgical, usually

with excellent results. Dolichocephaly without premature sutural closure often occurs in premature infants. Its origin is deformational caused by lying on the sides of the head for prolonged periods.

Isolated Coronal Synostosis. Early fusion of the coronal suture, whether unilateral or bilateral, is the second most common form of craniosynostosis, occurring in about 22% of isolated cases.⁹ As noted, it results in brachyturicephaly. If unilateral, it causes plagiocephaly and facial asymmetry. If unilateral, coronal synostosis occurs, and patient and family must be examined carefully for systemic signs of a

syndrome such as Saethre–Chotzen as described later.

Metopic synostosis is characterized by a bony prominence in the midline, extending from the anterior fontanel to the forehead. It accounts for about 15% of cases of isolated craniosynostosis and is most common in males. It is often accompanied by trigonocephaly and upslanting palpebral fissures. Metopic synostosis is part of several well-described chromosomal syndromes, particularly 9p- and some examples of Opitz C syndrome.⁹ Isolated lambdoidal synostosis, whether bilateral or unilateral, is relatively rare and results in marked flattening of the occiput. Oxycephaly, also rare, is associated with multiple sutural closures. It often results in a tall, pointed (tower) skull with the brain growing in the direction of least resistance. Acrocephaly has an ominous prognosis. Finally, Kleeblattschädel (cloverleaf skull) is an isolated abnormality or part of well-described syndromes such as Pfeiffer II syndrome or thanatophoric dysplasia. Cloverleaf skull is also associated with an ominous prognosis.

Primary Craniosynostosis with Known Mutations. Most of the common genetic causes of craniosynostosis have been traced to mutations. The most common craniosynostosis syndromes involve mutations in the FGFR system (Fig. 3).¹⁸ Mutations in other gene families have also been found. Unfortunately, like most genetic disorders, several mutations of the same gene are associated with a particular syndrome. For example, more than 15 mutations of the FGFR2 gene are associated with Crouzon syndrome.¹⁵ Thus genetic testing for Crouzon syndrome must involve sequencing the entire gene for malformations rather than looking for a single mutation causing all examples of Crouzon syndrome.

Moreover, mutations in different genes occasionally cause the same phenotypic syndrome. For example, Pfeiffer syndrome has been associated with mutations in FGFR1 and FGFR2.²⁰ Thus a specific diagnosis of Pfeiffer syndrome that might be useful in prenatal diagnosis would require screening both FGFR1

and FGFR2. An additional confusion, almost unique to the craniosynostosis syndromes, is that the same specific mutation can cause different craniosynostosis syndromes. For instance, the identical mutation has been associated with Crouzon syndrome in some families and with Pfeiffer syndrome in others.¹⁹ This feature is still unexplained but presumably is due to other genes altering the expression of a specific gene.

FGFR Mutations

Mutations of three FGFRs account for most causes of syndromic craniosynostosis.¹⁸ These include Crouzon, Crouzon with acanthosis nigricans, Pfeiffer, Apert, Muenke, Beare-Stevenson, and Jackson-Weiss syndromes. Crouzon syndrome is an autosomal dominant disorder characterized by variable, but usually symmetric, synostosis of the cranial sutures. Although the coronal suture is

the suture most commonly involved, all sutures in various combinations have been involved. Ocular proptosis (exorbitism), which occurs in all cases, is the hallmark of the disorder.⁹ Findings are usually confined to the head and neck although occasional findings involve the extremities. Neurological findings include visual alterations (decreased visual acuity, strabismus, and optic nerve hypoplasia) and acoustic abnormalities (conductive hearing loss). Seizures occur in 12% of cases and headache in 29%; hydrocephalus and jugular foramen stenosis with venous obstruction also occur.⁹ Intelligence is usually normal. Mutations in the FGFR2 gene are believed responsible for all cases of Crouzon syndrome, but to date only 50% of patients have known mutations.¹⁸ Crouzon syndrome with acanthosis nigricans is a separate disorder that features both conditions. It is sometimes referred to as the Crouzon dermoskeletal syn-

drome. This disorder is genetically distinct from Crouzon syndrome and involves the specific mutation A391E (alanine mutated to glutamate at the 391st amino acid of FGFR3).¹⁶

Pfeiffer syndrome tends to be a more severe disorder and has been divided into three subtypes. It is associated with mutations in FGFR1 and FGFR2.⁶ In all three types of Pfeiffer syndrome, brachy-turriccephaly is accompanied by abnormalities of hands and feet. The hands feature broad, medially deviated thumbs and variable brachydactyly (short fingers) while the feet exhibit broad, medially deviated great toes and variable toe brachydactyly. The broad, medially deviated thumbs and great toes are the hallmarks of this disorder. Individuals with Type I Pfeiffer syndrome usually have normal intelligence, moderate to severe midfacial flattening, exorbitism, typical hands and feet, and often hearing loss and hydro-

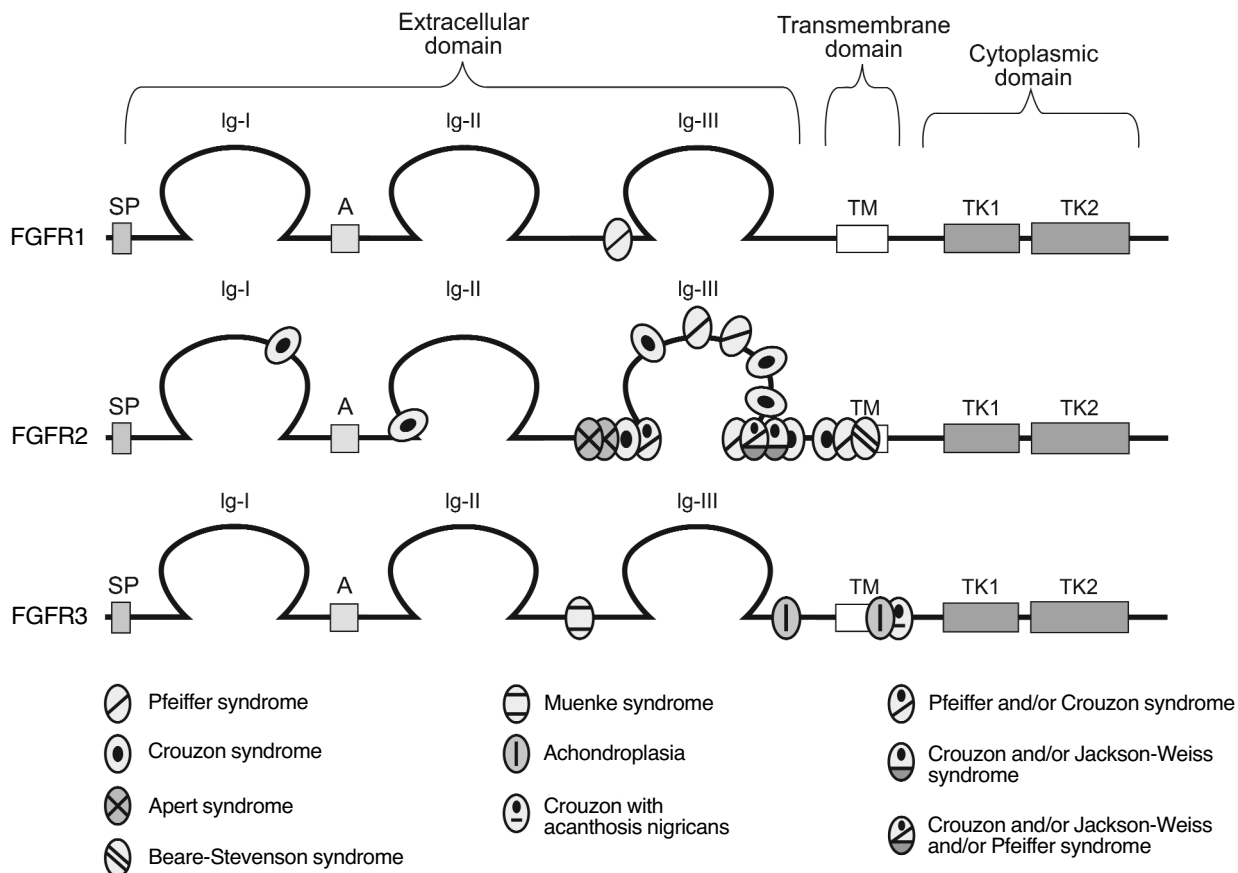


Figure 3. Schematic diagram showing the FGFRs and known sites of mutations in specific disorders.

cephalus. About 5% of patients with Type I have mutations in FGFR1 and 95% have mutations in FGFR2.¹⁸ Type I Pfeiffer has the most favorable prognosis of the subtypes.

Type II Pfeiffer syndrome is caused by mutations in FGFR2 and is associated with mental retardation, typical hand and feet changes, cloverleaf skull malformation, ankylosis of the elbows and knees, exorbitism with inability to close the eyes completely, hydrocephalus, seizures, and the risk of early death. The specific distinguishing feature of this disorder is the cloverleaf skull malformation.⁹ Type III Pfeiffer syndrome is caused by mutations in the FGFR2 gene. These patients are similar to those with Type II but lack the cloverleaf skull.⁹

Apert syndrome features brachyturri-cephaly and various degrees of exorbitism. The hands are distinguished by soft tissue and bony syndactyly (mitten hand), which is the hallmark of this disorder. The thumbs can be free or fused to the fingers. The feet exhibit similar changes. Intelligence varies, with at least 50% exhibiting mental retardation.⁹ Apert syndrome is caused by mutations in FGFR2, and more than 98% of the cases have identifiable mutations.¹⁸

Muenke craniosynostosis (also referred to as FGFR3-associated coronal synostosis syndrome) is a variable craniosynostosis syndrome in which patients can have bilateral or unilateral coronal synostosis or no craniosynostosis at all.¹⁷ Some mutation-positive individuals exhibit megacephaly as their only feature. The extremities in Muenke are also variable, but carpal tarsal fusion is typical. All cases of Muenke craniosynostosis are caused by mutations in FGFR3.

Jackson-Weiss and Beare-Stevenson syndromes are rare causes of craniosynostosis related to mutations in FGFR2. Jackson-Weiss syndrome is distinguished by relatively mild craniosynostosis and typical foot findings, including broad and medially deviated great toes, as well as various abnormalities of the tarsal bones.¹⁰ Beare-Stevenson syndrome is remarkable for cutaneous findings. In addition to mild craniosynostosis, these patients exhibit redundant skin (cutis gyrata) and

acanthosis nigricans.³ All exhibit mental retardation. Many exhibit unusual somatic features, including pyloric stenosis, an anteriorly placed anus, and genital anomalies.

Other Disorders Featuring Craniosynostosis and NonFGFR Mutations

Saethre-Chotzen syndrome is a common and variable craniosynostosis syndrome caused by mutations in the TWIST gene.⁸ Typical manifestations include facial asymmetry, a low anterior hairline, small ears, ocular proptosis, and asymmetric craniosynostosis. Digital abnormalities include two to three cutaneous syndactyly and brachydactyly. Intelligence is usually normal although mild mental retardation has been reported.⁹ Boston-type craniosynostosis is a rare form with a highly variable phenotype that can include fronto-orbital recession, frontal bossing, brachyturri-cephaly, and a cloverleaf skull or nonpenetration. It has been related to a mutation on the MSX2 gene.¹³

Antley-Bixler syndrome is a rare condition characterized by craniosynostosis, radiohumeral synostosis, joint contractures, and femoral bowing. Recently, it has been shown to be due to mutations in the P450 oxoreductase gene. The same phenotype occurs in fluconazole embryopathy and is due to inhibition of the P450 oxoreductase gene by high doses of the antifungal fluconazole during pregnancy.¹

Primary Craniosynostosis Without Known Mutations

As noted, there are more than 100 well-established genetic syndromes with known modes of inheritance. Among the most common are Carpenter syndrome, craniofrontonasal dysplasia, Baller-Gerold, Philadelphia-type craniosynostosis, Shprintzen-Goldberg, and Opitz C syndrome.

Secondary Craniosynostosis

Craniosynostosis can occur pre- or postnatally on a nongenetic basis. Compression of the fetal skull in the third trimester can cause closure along a specific suture. Compression can be caused

by fetal positioning or by extrinsic compressive forces such as uterine anatomy or a twin. Craniosynostosis can also be due to metabolic conditions such as hyperthyroidism, mucopolysaccharidosis, or rickets.⁵ Craniosynostosis has been associated with various syndromes caused by teratogens including diphenylhydantoin, valproic acid, aminopterin, retinoic acid, and fluconazole.⁵ Finally, craniosynostosis can occur when the brain fails to grow as in extreme microcephaly.

Molecular Biology of the Craniosynostosis Syndromes

Mutations in three FGFRs have emerged as central to the common craniosynostosis syndromes. FGFRs are receptors for circulating FGFs. Seventeen FGRs are known regulators of cell proliferation, differentiation, and migration.¹⁴ Each stimulates a cell membrane-associated FGFR. Surprisingly, there is no specificity between the stimulating FGF and the stimulated FGFR. Consequently, any FGF can stimulate any FGFR.

The FGFR molecule has three domains: an extracellular domain, a transmembrane domain, and a cytoplasmic domain. The extracellular domain consists of three immunoglobulin-like domains. The binding of the FGF to the FGFR occurs at the second and third immunoglobulin-like domains. After an initial binding of FGF and FGFR, the FGFR binds to another FGFR and forms a dimer. As noted, any FGF can bind with any FGFR and the dimerization can occur with any FGFR.

The extracellular binding and dimerization stimulate the third domain of the FGFR, a split cytoplasmic tyrosine-kinase domain. Once the tyrosine-kinase domain is stimulated, it stimulates a complex sequence of intracellular signaling. The intermediate domain, the intramembranous domain, anchors the receptor to the cell. However, mutations in this domain can be associated with malformation syndromes, particularly achondroplasia, a mutation of FGFR3 associated with decreased limb growth.

Remarkably, the FGFRs are responsible for restraining growth. Mutations that enhance the activity of these receptors can cause decreased growth, as seen in achondroplasia. Mutations that decrease the activity of these receptors would be less likely to restrain growth and are said to be “hypermorphic.” Early fusion of a cranial suture is a hypermorphic event.

Mapping the three involved FGFRs shows that Pfeiffer syndrome has been associated with mutations in FGFR 1 and 2. Strictly speaking, this finding suggests what is suspected clinically: Pfeiffer syndrome is heterogeneous. Crouzon syndrome is associated with mutations in all three immunoglobulin-like domains of FGFR2. Apert syndrome is associated with mutations between two immunoglobulin-like domains. Remarkably, two separate families, one with Crouzon and one with Apert, have been associated with the same mutation. This phenomenon is still unexplained.

Neurological Considerations in Craniosynostosis

Neurologic alterations are common in syndromic craniosynostosis and less common in isolated craniosynostosis. Not surprisingly, patients with substantial alterations in skull shape have neurological abnormalities. However, not all neurological abnormalities can be attributed to the bony changes. Some neurological changes are primary defects that accompany the synostosis rather than the consequence of the craniosynostosis. This final section reviews the major neurological alterations associated with craniosynostosis syndromes.

Mental Retardation

Mental retardation is a common complication of craniosynostosis. In primary nonsyndromic craniosynostosis with only one fused suture, the rates of retardation are low. Estimates range from 2.4 to 4.8%,⁴ which is similar to the general population. When a second or third suture is involved, retardation becomes more common. With the complex and multi-

ple craniosynostosis associated with oxycephaly and cloverleaf skull, retardation is very common.

There are no reliable figures for the incidence of mental retardation associated with syndromic synostosis although reasonable estimates have been offered. Individuals with Crouzon syndrome appear to be among the least likely to exhibit mental retardation, with only 3% exhibiting “marked” retardation.⁹ Saethre-Chotzen syndrome is much like Crouzon syndrome in that affected individuals are typically normal.

Type I Pfeiffer syndrome is usually associated with normal intelligence, but individuals with Types II and III typically exhibit severe retardation. Mental retardation is likely overestimated in Pfeiffer syndrome by a factor of two or three because of the severe visual and auditory abnormalities in these individuals. The frequency of retardation associated with Apert syndrome is variable. The mean IQ of such individuals is 70.⁹ Few individuals have an IQ higher than 100, but occasionally patients with Apert syndrome have a high IQ.⁴ Thus Apert syndrome and normal or high intelligence are compatible. The intellectual performance of individuals with FGFR3 (Muenke) craniosynostosis is relatively mildly affected, but 37% are said to have mild developmental problems.⁹

The cause of mental retardation is not readily explained, but it is often attributed to the increased intracranial pressure caused by hydrocephalus or decreased venous drainage or to anatomic changes related to brain distortion. Primary comalformations of the brain are only rarely considered a cause of retardation.

Increased Intracranial Pressure

Increased intracranial pressure is also relatively common in the craniosynostosis syndromes. The increased pressure tends to be low grade and chronic. Severe headaches occur in 30 to 50% of cases of syndromic synostosis,⁴ and these headaches may be correlated with increased intracranial pressure. Intracranial hypertension can also lead to papilledema, which can cause optic atrophy and even

blindness. Optic atrophy can also result from local factors such as foramina stenosis or closure.

Hydrocephalus

Hydrocephalus is most common in patients with multiple sutural synostosis, and its incidence appears to be highest in Crouzon syndrome. The site of obstruction is most often at the basal cisterns, but aqueductal stenosis is also reported. Increasingly, jugular venous obstruction is being recognized as the cause of the hydrocephalus.

Cranial Nerve Dysfunction

The optic and auditory nerves appear to be the cranial nerves most commonly affected in craniosynostosis. As noted, dysfunction of the optic nerve is a consequence of increased intracranial pressure or of stenosis of the optic foramina.

Some degree of optic atrophy occurs in as many as 20% of patients with Crouzon syndrome,¹² and this incidence has been estimated to be as high as 80%.² The frequency of optic atrophy is lower in almost all craniosynostosis syndromes and occasionally in nonsyndromic synostosis. Again, the greater the number of sutures involved, the more likely it is that optic atrophy will develop. Other ocular problems include strabismus and irritation of the cornea from exposure caused by exorbitism.

Abnormalities of the auditory nerve are also important findings that necessitate audiological evaluation of all patients with craniosynostosis. Although conductive hearing loss is the most common cause of hearing in these syndromes, nerve deafness also occurs.

Diagnostic Evaluation

An abnormal head shape or size suggests the presence of craniosynostosis. Typically, the normal shape is exaggerated either with a very brachycephalic or dolichocephalic head. An asymmetric head is associated with unilateral sutural involvement or with positional plagiocephaly. When a markedly brachycephalic, dolichocephalic, or plagiocephalic head is encountered, the possibility of

craniosynostosis must be considered. A very microcephalic head should also raise the possibility of early sutural closure. Finally, very unusual head shapes, such as acrocephaly or cloverleaf skull, also suggest craniosynostosis.

The initial evaluation for craniosynostosis can be challenging. First, careful palpation of the skull sutures for patency or "heaping-up" is undertaken. If the suture is nonpatent at an age when it should be patent or if there is a raised area along the suture (heaping-up), craniosynostosis should be considered. Experienced clinicians can often distinguish between patent and nonpatent sutures, but imaging techniques may be needed. Standard x-ray evaluation can be helpful, but standard or three-dimensional computed tomography is often necessary.

Once the presence of craniosynostosis is confirmed, the patient should be examined for extracranial findings that might indicate a specific syndrome. The eyes and extremities are most helpful in this regard. The presence of hypertelorism or exorbitism is typical of syndromic synostosis. Alteration of the hands and feet, either with cutaneous or bony syndactyly, or broadening of the thumbs or great toes is particularly common.

After the patient is examined, a detailed pregnancy and family history is necessary. Exposure to teratogens during early pregnancy or a family history of unusually sized or shaped heads can indicate an etiology for the patient's abnormalities. It also can help determine the patient's prognosis and the risks of recurrence for subsequent pregnancies.

Although some patients exhibit clear-cut syndromic diagnoses, it is often still necessary to test the patient with molecular probes. Thus, if a patient has what appears to be Crouzon syndrome, the physician should still attempt to identify the specific mutation involved and to determine the possibility that one parent is very mildly affected and the associated risk for subsequent pregnancies. Moreover, if a specific mutation is identified in a child, this information can be used to test subsequent pregnancies for the same al-

teration. If a patient exhibits an unclassifiable pattern of changes, molecular probes can be used in an attempt to define the specific mutation involved.

Molecular diagnostic laboratories now offer testing for specific gene alterations (i.e., FGFR1, 2 or 3 or the TWIST mutation). Clinicians can also request a craniosynostosis panel that evaluates all involved genes. Clinicians often encounter patients with syndromic synostosis without a specific mutation identified, especially in Crouzon syndrome. With a strong family history of craniosynostosis and no identified gene, it is sometimes possible to identify mutations with complete sequencing of the known genes. This option is usually available on a research basis.

Conclusion

Craniosynostosis is a relatively common disorder of children, and its etiology is heterogeneous. Causes may be genetic, developmental, teratogenic, metabolic, or idiopathic. Recently, the molecular biology of several craniosynostosis syndromes, particularly the FGFR-related disorders, has been elucidated. This understanding, however, has introduced new levels of complexity that are not yet clearly understood.

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Principles of Craniofacial Surgery[†]

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The essential principle of care for those affected by a craniofacial anomaly is the interdisciplinary approach. The craniofacial healthcare team evaluates and plans treatment using established protocols individualized for each patient's unique expression of his or her syndrome. The axioms of craniofacial surgery, established by Dr. Paul Tessier, have their roots in lessons learned from the treatment of facial injuries in World War II. Ideal timing of surgery, meticulous planning, use of facial skeletal osteotomies, autogenous bone grafts, rigid fixation, distraction osteogenesis, and soft tissue repair are the essential elements of craniofacial surgical reconstruction.

Key Words: cranial grafts, craniofacial surgery, craniofacial syndromes, craniofacial team

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The German, Max Picard, wrote that "A man is as good as his face appears to be. The essence of man dwells in the front of his face." Jean Cocteau wrote "If there is a defect on the soul, it cannot be corrected on the face. But, if there is a defect on the face and one corrects it, it can correct the soul." Indeed, deformities of the face significantly affect a person's life, especially young impressionable children trying to define themselves as individuals.

Throughout history, and even among medical professionals, facial deformity has been equated with mental retardation. This concept has been perpetuated by the movie and advertising industries. Indeed, the unfortunate child born with a severe facial deformity has little chance of developing healthy self-esteem, which is required to lead a normal life. One needs only to meet one such child to realize the severe inner torment that is endured each day.

Craniofacial centers, using an interdisciplinary approach, have been established to treat such children. The development of craniofacial techniques has made possible osteotomies of all facial bones and repositioning in almost any direction necessary, including repositioning the brain within the cranium. The result has often improved the appearance, behavior, achievement, and self-esteem of patients.⁷ This article briefly reviews the history of craniofacial surgery and considers how an interdisciplinary team approach helps optimize outcomes for patients.

History of Craniofacial Surgery

The first efforts to correct facial skeletal deformities began during World War I,

which produced severe traumatic deformities that required repositioning of fractured facial bones or reconstruction of missing parts. Techniques and concepts developed further during World War II. Subsequently, these techniques were applied to the correction of congenital craniofacial deformities.

Many surgeons have helped advance the field of craniomaxillofacial surgery. Three surgeons, however, have made major contributions. Sir Harold Gillies made the transition between reconstruction of traumatic deformities to correction of major congenital deformities. Professor Hugo Obwegeser developed techniques for correcting the lower facial skeleton. Dr. Paul Tessier, however, is the true pioneer of contemporary craniofacial surgery (Fig. 1).¹

In 1957 Tessier saw a 20-year-old patient with severe exorbitism and a grotesque craniofacial configuration. He later recognized this set of deformities as Crouzon disease and realized that the orbital maxillary and facial abnormalities must be treated simultaneously. As he studied the problem, he realized that a radical solution was necessary. He practiced on cadavers at the Department of Anatomy in Nantes. Later, at surgery, he advanced the entire facial skeleton 2.5 cm using bone grafts to fill the spaces. In the late 1950s and 1960s, he treated other patients with craniosynostosis and orbital hypertelorism. He waited 3 years before operating on his first case of orbital hypertelorism, obviously a reflection of the respect that he had for this radical surgery.

In 1967 in Montpellier, Tessier presented his first case at the meeting of the French Society of Plastic Surgery. His presentation was well received, but its full impact was not appreciated until he reported his work at the International Congress in Rome in 1967.^{2,16} Subsequently, Tessier invited about 20 people, including several leading plastic surgeons and specialists in other disciplines, to observe his techniques. He requested their criticism and stated that he would stop this surgery immediately if they thought the procedure was too radical. The group was amazed and en-

thusiastic about his techniques, and he continued.¹¹

Violating many of the conventional surgical principles of the day, Tessier proved that his radical approaches allowed severe, previously untreatable deformities to be corrected. His guiding fundamental principle was as follows: If a craniofacial defect was caused by an underlying skeletal deformity, the only solution was to reposition the skeleton or to reconstruct it with autologous bone grafts.

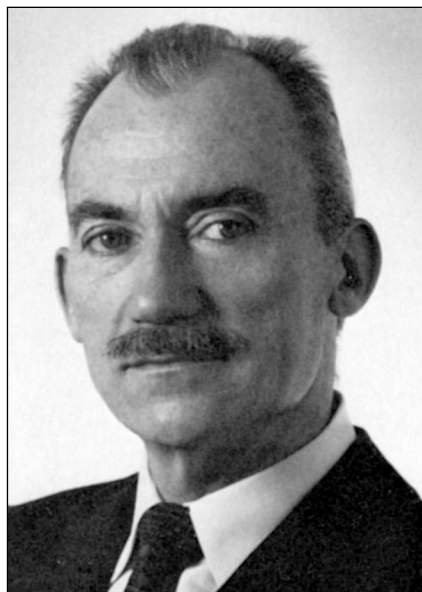


Figure 1. Paul Tessier. From Jackson IT, Munro IR, Salyer KE, Whitaker LA (eds): *Atlas of Craniomaxillofacial Surgery*, St. Louis: Mosby, 1982. Copyright Elsevier, 1982.

Based on this principle, Tessier developed three techniques previously considered impossible. First, extensive areas of the craniofacial skeleton could be devascularized completely by stripping the periosteum and repositioning the bones. Second, circumferential mobilization of the orbital contents allows the eye to be moved in any of the three planes without affecting vision. Finally, simultaneous intracranial and extracranial surgery, which would allow radical movement of the orbit and skull for reconstruction, could be performed. These three principles remain fundamental to craniomaxillofacial surgery.

The Team Approach

From the inception of contemporary craniofacial surgery, Tessier worked with many specialists of the head region to help him evaluate and treat these patients. Professionals of all disciplines who are interested in head and neck diseases have many overlapping areas of expertise. In the 1960s it was established that children with clefts are best treated by well-coordinated teams of specialists. In the 1970s craniofacial deformity teams evolved from teams treating patients with clefts of the lip and palates.^{5,6} The rationale for such a team is well accepted: Patients with craniofacial deformities and their families have a complex interplay of physical, mental, and emotional factors that require expertise not possessed by a single health-care discipline.

Today this "healthcare team" is the dominant organizational structure for the delivery of care to patients with craniofacial differences. The contemporary craniofacial team is based on an interdisciplinary model. The team meets regularly to assess and plan, and each professional performs an individual assessment of the patient before sharing findings and agreeing on recommendations.¹⁴ The keys to success for the interdisciplinary team are vigilant concern for the patient, shared input, and respect across professional lines. Ultimately, the overall physical and emotional health of these complicated patients depends on the clinical expertise of each team member and on meaningful participation from the patient and family.¹³

Several factors created the need for a team approach to provide patients with craniofacial deformities with the appropriate comprehensive treatment. First, the nature of craniofacial deformities requires that specialized professionals from the many disciplines related to the head evaluate and treat these patients (Table 1). The role of each team member is well described in the literature.^{2,4-6} Second, the rarity of these deformities does not allow a sufficient number of cases for specialists in every medical center to gain the necessary experience and skill to manage these patients safely. Third, the establishment of craniofacial centers allows

Table 1. Disciplines Represented on a Craniofacial Team

Anesthesiology	Orthodontics
Anthropology	Otolaryngology
Audiology	Pediatrics
Computer sciences	Pedodontics
Genetics	Photography
Medical illustration	Plastic surgery
Neuroradiology	Prosthodontics
Neurology	Psychiatry
Neurosurgery	Psychology
Nursing	Radiology
Ophthalmology	Social services
Oral surgery	Speech pathology

longitudinal study of patients to document long-term outcomes of treatment. Such follow-up is essential to foster the development of new ideas and to evaluate the effect of these radical procedures on the growth of the craniofacial skeleton. Finally, the reality of health care demands that this interdisciplinary care be performed as cost effectively as possible. Increasingly, third-party payers and state agencies for disabled children favor centralization of care.^{4,6}

To be successful, a craniofacial team requires an adequate number of patients, enthusiastic input from each participating professional, and strong organization. The goals of the team are to diagnose, to plan and execute treatment, and to provide long-term follow-up that considers all the ramifications of the disorder and its treatment.

Craniofacial teams often have a medical director dedicated to the treatment of craniofacial disorders. A parallel non-medical administrator or coordinator is responsible for organizing and coordinating activities among the various professionals, agencies, patient, and family. The team meets regularly to assess patients. The results of these evaluations must be communicated to the patient, responsible agencies, and primary-care providers.¹⁷ Good organization and open communication are essential to insure that patients receive the appropriate care.

For the patient with an acquired or congenital facial deformity, the team ap-

proach provides the best opportunity for gathering, analyzing, and assembling information. In turn, this process provides the basis for the most accurate and complete diagnosis, prognosis, and treatment plan.¹³

Planning

Craniofacial team members must be trained to think about major skeletal deformities. When correction of a craniofacial deformity is planned, facial proportions must be emphasized. In the lower face, there are many ways to achieve normal occlusion. Not all of these methods, however, would provide aesthetically pleasing facial proportions. It is also important to understand the natural history underlying the growth and development of a given deformity in relationship to a normal face.

Evaluation begins with clinical assessment by the craniofacial surgeon. Problems are noted and analyzed individually. The craniofacial surgeon must organize a preoperative assessment of each problem prepared in conjunction with the other team members. The neuroradiologist has the key role of delineating the craniofacial skeletal anomaly. Three-dimensional computed tomography is essential for this purpose. When a patient's orbits must be moved during surgery, the neuro-ophthalmologist must provide a detailed assessment. The orthodontist evaluates the patient's occlu-

sion and must plan the corrections. Members of the psychosocial disciplines are especially important in evaluating the patient's mental capacities, emotional milieu, and the impact of the deformity on the patient and family. These professionals make projections about the effect of the corrective surgery, which may be indicated or contraindicated on the basis of emotional considerations alone. The unique perspective of these team members helps keep the other members aware of the larger picture.³

After the patient's assessment is completed, each participating member makes recommendations at the team meeting. The craniofacial surgeon then assimilates these findings and determines the final treatment plan.⁹

Timing of Surgery

In patients whose deformity merits surgical treatment, four factors affect the timing of their procedure: their age, the effect of further growth, the severity of the deformity, and the psychosocial effects on patient and family.²

As a general guideline and if technically feasible, it is preferable to operate on patients at as young of an age as possible.¹ Growth does not improve skeletal deformities. Indeed, some problems (exorbitism and malocclusion) become worse with growth. Delaying treatment can require more radical surgery with less chance of achieving the best outcomes.

Psychosocial concerns, however, often override all other factors. Children must develop a healthy self-esteem to become well-adjusted adults. Children who spend their formative years among peers tormented about their disfigurement are unlikely to develop a good self-image. Thus, it is better to treat certain deformities surgically at an early age even though some procedures may need to be repeated as growth alters the corrections that were achieved. Indeed, it is easier for patients to cope with their deformities if treatment markedly improves their appearance and they know that future surgery will improve their appearance even more.⁸

With these concepts in mind, general guidelines have been established to determine the timing for surgical correction of some deformities. Patients born with craniosynostosis should be treated within the first 6 to 9 months, preferably within the first few months of life. Likewise, encephaloceles should be corrected as early as possible. Orbital hypertelorism or dystopia, which requires moving the entire orbit, is difficult to correct technically before the age of 2 years. Advancing the infant's forehead may help prevent exorbitism. At the age of 5 or 6 years, the skeleton is mature enough to advance the forehead and maxillary bones simultaneously. Exorbitism severe enough to cause visual changes or corneal exposure can force early intervention. Patients with severe maxillary retrusion can develop sleep apnea because their nasopharyngeal space is severely compromised. These patients also may require early maxillary advancement.

Patients with Treacher-Collins syndrome should undergo periorbital correction before they begin school. Their jaw and occlusal problems are best treated after their secondary dentition has erupted and they have completed growth as an adolescent. Patients with craniofacial microsomia have a wide range of expressivity. Patients with severe cases should undergo earlier skeletal correction and construction of absent bone than those with mild cases.²

Typically, problems are not corrected surgically until after facial growth is completed. Again, however, psychosocial considerations may dictate earlier intervention in patients with severe problems even though they may need surgery later.

Surgery

Anesthesia

The anesthesiologist and surgeon must have a close working relationship, each understanding the potential problems of the other. The anesthesiologist must be familiar with the techniques of neuroanesthesia, pediatric anesthesia, and

hypotensive anesthesia. Preoperatively, the patient requires a clinical evaluation, and rapport should be established with the patient and family. The airway and potential difficulty with intubation must be assessed. Maxillary, mandibular, and cervical spine anomalies often must be considered. Blood must be cross-matched for 1.5 times the estimated loss of blood volume.

Controlled hypotensive anesthesia is an asset to craniofacial surgery. Mean arterial pressure is reduced to about two-thirds of the patient's normal value. This technique greatly facilitates surgery. It not only decreases the amount of blood loss and thus the amount needed for transfusion, it also reduces the length of the operation.

Postoperatively, severe swelling is common and airway precautions dominate. Patients with intermaxillary fixation may need to remain intubated from several hours to several days. Postoperative pain is usually minimal in the head and neck; it is primarily associated with rib and iliac bone graft donor sites. The administration of analgesics should be minimized after intracranial procedures.¹²

Basic Principles

The keys to successful craniofacial surgery are careful preoperative planning, skillful technical execution of the osteotomies, and rigid fixation. The latter allows rapid bony healing and prevents relapse.⁹

The use of bone grafts is an important tool in craniofacial surgery. They are used to fill gaps of advanced segments, to construct missing segments, and to correct an abnormal contour by onlay grafting.² Three different sites can be used: iliac, rib, and cranial bone grafts. Although still preferred by many surgeons, iliac bone grafts have lost popularity because of their associated morbidity. Nevertheless, it is a good source of bone when indicated. Furthermore, it is the best source of cancellous bone, which is used to graft alveolar cleft defects. Rib grafts and split cranial grafts are the most common donor sites in craniofacial surgery. Rib grafts have the advantage of

easy harvest and ease of contouring in areas such as the orbit and zygoma. Costochondral bone grafts are used to reconstruct the ramus and temporomandibular joints in patients with hemifacial microsomia. They are also used for nasal reconstruction.

The use of split cranial bone represents one of the recent advances in craniofacial surgery. The bone can be harvested through well-concealed scalp incisions by one of two techniques. If a relatively small amount of bone is needed and a craniotomy is unnecessary, the outer table can be harvested with the use of burs and osteotomes. The inner table is left intact to protect the brain. If larger amounts of bone are needed or a craniotomy is otherwise necessary, the bone segments can be removed as a full thickness piece and split using a power saw. The outer table is replaced to reconstruct the cranium.¹⁵

For many craniofacial surgeons, a split cranial graft is the first choice because of the ease of harvesting, minimal morbidity and postoperative pain, and minimization of operative sites. Furthermore, cranial bone is membranous bone, as is a large proportion of the craniofacial skeleton; therefore, it is thought to undergo less resorption than other types of grafts.¹² Compared to the use of rib grafts, another advantage is that a large sheet of cranial bone can be harvested. The sheet allows reconstruction of major facial segments without the need to combine many smaller segments as is required when rib grafts are used. The procedure is then less time-consuming, and the graft provides more stability to the reconstruction during the early stages of healing. The one disadvantage of split cranial bone that remains to be solved is that it is much less pliable than rib grafts. Thus, it may not always be applicable when this feature is necessary, such as in orbital reconstruction.

Over time, the preferred surgical incision in craniofacial surgery has changed. In 1957 when Tessier performed his first correction of Crouzon syndrome, he used multiple facial incisions. Bicornal incisions provided excellent exposure to the cranium and upper facial skeleton.

As the specialty has developed, the number of facial incisions has decreased. Almost all procedures can now be performed through a bicoronal incision (Fig. 2), intraoral incision, or a combination of the two. These incisions are often even adequate to correct orbital hypertelorism. Formerly, a disfiguring midline incision over the nose was considered necessary.

Complications

The primary major complications associated with craniofacial surgery include death, brain damage, blindness, and infection. A large combined report of 793 operations performed at six centers detailed some of the problems and complications associated with craniomaxillofacial surgery.¹⁸ The complication rate was 16.5%, and the mortality rate was 1.6%. The rate of blindness was less than 1% and that of brain damage was 0 to 0.5%. Infection occurred in 4.4% of cases. Factors that reduced morbidity and improved outcomes were hypotensive anesthesia, short operative time, rigid stabilization of the mobilized bones at the end of the operation, few incisions, and extensive antibiotic therapy.

Munro and Sabatier reported their experience of more than 12 years with 2,019 craniomaxillofacial procedures performed in 1,092 patients.¹⁰ Their overall mortality rate was 0.64%; the mortality rate directly related to surgery was 0.18%. Major complications occurred in 14.3% of patients, but few had permanent sequelae. Infection, the most common complication, occurred in 5.3% of major operations. The authors noted that the incidence of complications was partially related to inexperience with craniomaxillofacial surgery. As they gained experience, their annual incidence of complications decreased to 5.4%. Experience creates familiarity, which is a major factor in reducing the length of surgery. This learning curve lends further support to the essential concept of performing craniomaxillofacial surgery at a major center where the craniofacial team obtains experience to develop and maintain expertise in performing these complicated procedures.

Conclusion

Craniofacial surgery requires patients to be treated by a large specialized multidisciplinary team that can obtain adequate experience to maintain their surgical expertise. A multidisciplinary approach increases the safety of the procedures, improves technique, and allows longitudinal study of large groups of patients. The timing of surgical intervention depends on the patient's growth, functional problems, and psychosocial considerations. The team must consider that young children need to develop self-esteem to function normally as adults. The keys to technical success of craniofacial surgery are good planning, precise execution of the osteotomies, and rigid fixation. The development of patients' social skills, inter-

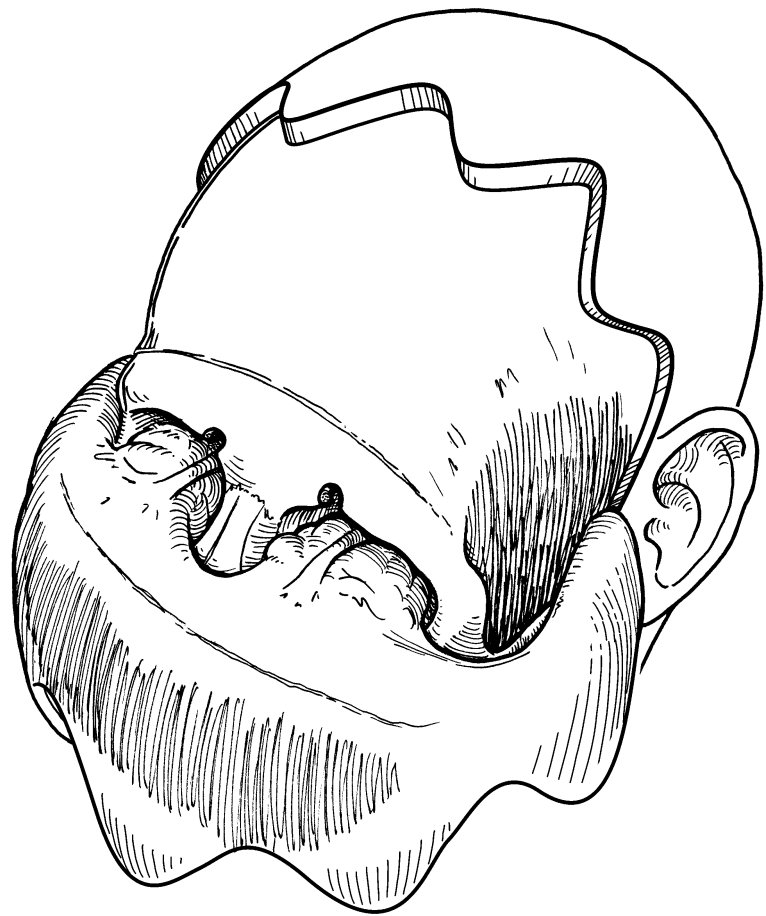
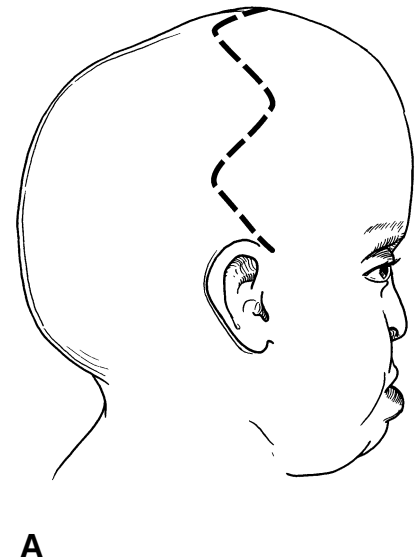


Figure 2. (A) The bicoronal incision provides good exposure of the (B) entire upper craniofacial skeleton.

personal relationships, opportunities for education and career, personal expectations, and goals in life are all affected by how they perceive themselves and by how others perceive and react to them in return. Appropriate craniofacial surgery performed by an experienced interdisciplinary team can help optimize outcomes for patients with craniofacial deformities.

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Distraction Osteogenesis of the Craniofacial Skeleton

Davinder J. Singh, MD

Since the early 1990s, distraction osteogenesis, a bone-lengthening technique, has evolved for use in craniofacial surgery. This article reviews the applications, advantages, and disadvantages of distraction osteogenesis of the craniofacial skeleton.

Key Words: bone remodeling, craniofacial surgery, distraction osteogenesis, hemifacial microsomia

Distraction osteogenesis, a technique for lengthening bone, uses the body's natural healing mechanisms to generate new bone. An osteotomy is made in the area of bone deficiency, and a device is placed that slowly elongates the bone to its new dimension while natural ossification produces new bone at the site of distraction.¹ Before the advent of distraction osteogenesis, bone lengthening or expansion was performed using osteotomies and bone grafts. Expansion was limited by the constriction of soft tissue and the vascularity of the graft.

In 1905 Codivilla described the first use of osteodistraction in orthopedic surgery.¹ As discussed in his historical overview,³ Ilizarov further developed the technique for use in the lower extremities in 1949. In 1992 McCarthy et al. reported the first application of distraction osteogenesis to lengthen the human mandible.⁵ This publication in the English literature began the era of craniofacial distraction, which is now used at every level of the craniofacial skeleton. Distraction osteogenesis is most often used for congenital hypoplasia or growth restriction, but it has also been used for bone remodeling to fill gaps from posttraumatic defects or cancer resection.⁹

The advantage of distraction osteogenesis is that it mimics the normal physiologic process, allowing time for soft tissue to remodel and adapt. Expansion of the soft tissue envelope is the single most beneficial aspect that enables greater skeletal distraction than can be achieved in traditional operations. Distraction osteogenesis also shortens surgical times and lengths of hospital stay,

Abbreviations Used: CT, computed tomography; HFM, hemifacial microsomia

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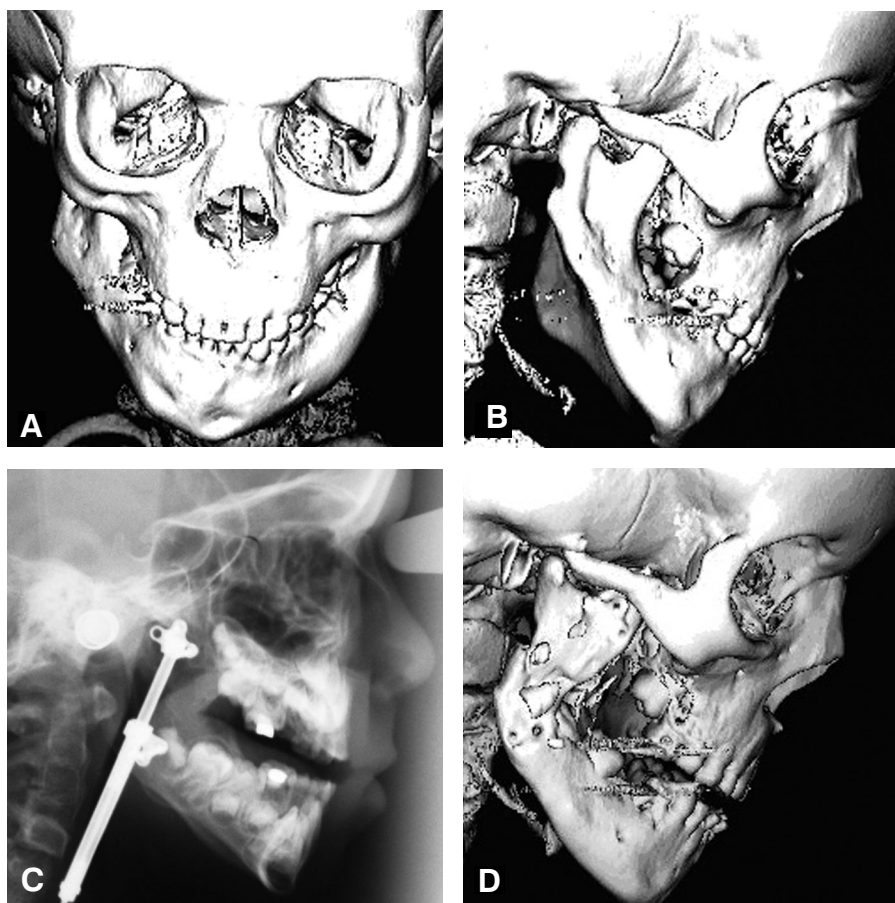


Figure 1. Three-dimensional (A) frontal and (B) lateral CT scans of a 8-year-old boy with right HFM and a Pruzansky Type IIB mandible. (C) Lateral cephalogram shows semi-buried mandibular distractor after full activation and lengthening. (D) Postoperative three-dimensional CT scan shows the increase in the length and volume of the right mandibular ramus.

obviates the need for bone grafting, reduces the likelihood of relapse, reduces the likelihood of transfusions, and offers considerable postoperative control over the distraction of one or more bone segments.^{6,7,9}

Process of Distraction Osteogenesis

The process of distraction osteogenesis begins with preoperative planning, continues through several stages, and culminates with removal of the device. Preoperative planning entails deciding on the location of distraction on the craniofacial skeleton and obtaining appropriate imaging studies such as three-dimensional CT, models, and cephalography. To some extent, the patient's

age and size dictate the rate of distraction. Devices can be uni-, bi-, or multidirectional, and they can be internal or external.⁶ Before the device is placed, especially in the case of unidirectional devices, the vector must be determined. Vector placement determines the direction of elongation or movement.² After preoperative planning has been completed and the device has been selected, the osteotomies are performed and the device is placed. After surgery, distraction osteogenesis proceeds according to the phases of healing.

Distraction osteogenesis entails three main healing phases: latency, activation, and consolidation. Latency is the period that follows the osteotomy and application of the device; it ranges from 1 to 7 days depending on the patient's

age. The latency period is followed by the activation phase in which the distractor device is activated to lengthen the gap between the bones. The device lengthens the gap 1 mm/day. This rate allows callus to form and the soft tissue envelope to expand slowly. After the appropriate length has been reached, the consolidation phase lasts twice as long as the time required for activation. During consolidation, the device provides temporary fixation as the bone heals in its new position. After the bone has healed, the device is removed.^{5,9}

Applications of Distraction Osteogenesis to the Craniofacial Skeleton

Distraction osteogenesis can be used in most parts of the craniofacial skeleton and has been described for mandibular distraction, maxillary distraction at the Le Fort I level, maxillary distraction at the Le Fort III level, frontal facial (monoblock) distraction, and calvarial distraction.⁹ Distraction osteogenesis is most often used for distracting the mandible or maxilla because it is ideal for correcting hypoplasia of these areas.⁷

Mandibular hypoplasia is associated with both functional and aesthetic concerns. Functional concerns include airway compromise, feeding difficulties, speech problems, and dentition.⁸ Mandibular distraction is indicated in patients with hemifacial or bifacial microsomia, Treacher-Collins syndrome, Pierre Robin sequence, and posttraumatic growth disturbance.⁶

HFM is one of the most common congenital anomalies with an incidence of about 1 in 5000 births.⁴ In numerous cases of HFM, the mandible has been distracted with adequate bone to lengthen (Fig. 1). In cases of severe hypoplasia, bone grafting is performed first. Then the graft is lengthened via distraction as the child grows. In a growing child, this strategy offers the opportunity to improve function and appearance without leaving permanent hardware, which poses issues with growth restriction.⁵ Devices may be internal or external based

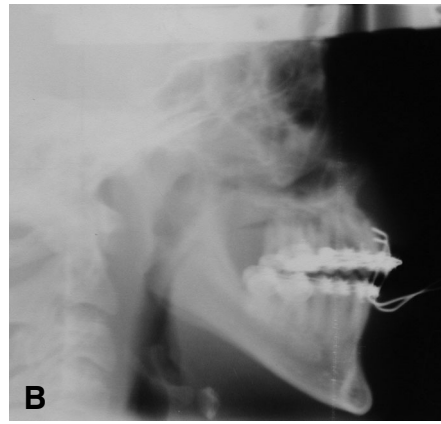
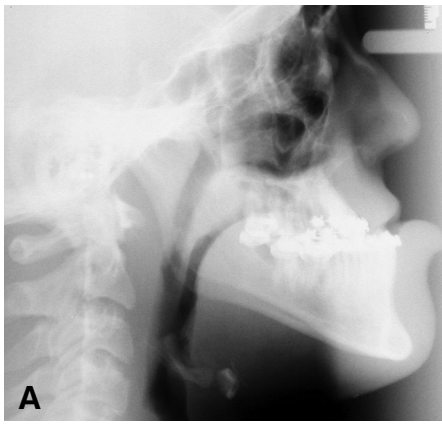


Figure 2. (A) Lateral cephalogram of a 17-year-old male with a unilateral cleft lip and palate and maxillary hypoplasia and retrusion. (B) Lateral cephalogram after external distraction at the Le Fort I level with a halo device shows Class I occlusion.

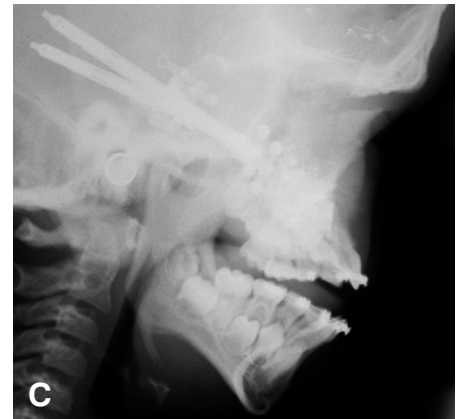
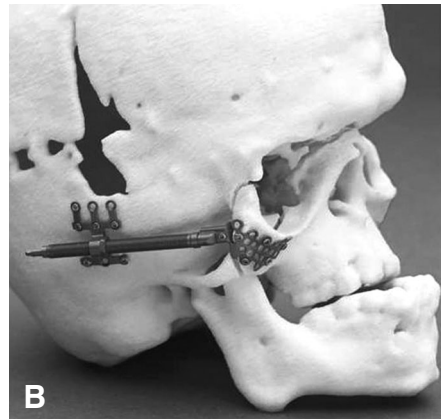
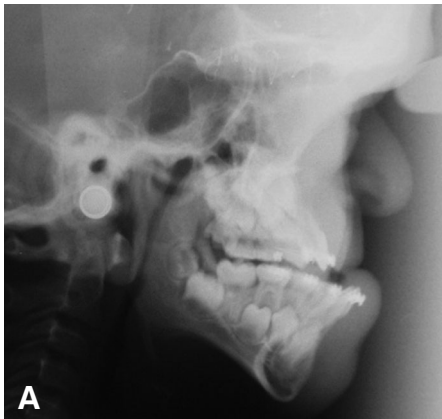


Figure 3. (A) Lateral cephalogram of a 7-year-old boy with Apert syndrome and midfacial hypoplasia who underwent advancement at the Le Fort III level for exorbitism and Class III malocclusion. (B) Three-dimensional model of this patient showing placement of internal distractor appliances for advancement at the Le Fort III level. (C) Lateral cephalogram at the completion of distraction shows the advancement of the midface and the improved proportions of the face and occlusion.

on the patient's age, size, and degree of deformity.

Midface deficiency can also be corrected with distraction. Indications include severe maxillary hypoplasia and the need for more than 10 mm of distraction. Patients with a cleft lip and palate who have significant palatal scarring are candidates, as are patients with syndromes such as Apert and Crouzon who have severe midface retrusion.⁹ Advancement of the maxilla at the LeFort I (Fig. 2) and III (Fig. 3) levels improves airway, mastication, speech, and appearance.

Distractor devices can be either internal or external. Internal devices improve cosmetic appearances and reduce the likelihood of dislodgement. However, not all patients are candidates for internal devices. The more hypoplastic the bone is, the less room there is for internal devices

and the greater is the need for the use of external devices. Postoperatively, external devices offer more vector control and fine tuning of the distraction. In contrast, the internal device has a fixed vector that must be decided intraoperatively.

Complications related to the procedure are hardware infection and exposure, dislodgement of the device, device failure, premature consolidation, malunion, and nonunion.^{7,9} Most patients tolerate the distraction process well. However, distraction requires a high degree of compliance by the patient and family because it necessitates daily maintenance and limits activities.

Conclusion

Compared to traditional orthognathic procedures, distraction osteogenesis of

the craniofacial skeleton allows superior advancements in treatment and reduces the likelihood of relapse. Not all patients require such a large amount of distraction, and a role for conventional, single-stage procedures still exists. However, in severely affected patients and in patients who are still growing, distraction osteogenesis has revolutionized the craniofacial surgeon's ability to improve function and aesthetics in patients at a younger age than was once possible.

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Oral Manifestations of Selected Craniofacial Conditions

Patricia Glick, DMD

Children born with clefts of the lip and palate; craniofacial syndromes such as Apert, oculoauriculovertebral, and Treacher-Collin syndrome; and chromosomal abnormalities such as Down syndrome and 22q11.2 deletion syndrome experience significant dental morbidity. Their treatment is best planned by an interdisciplinary team approach. In this and other countries, coordinated care among surgeons, dental professionals, and other specialists is considered the standard of care. Because these children have high rates of oral anomalies, early screening, careful monitoring, and coordination of related services are important components of their comprehensive care.

Key Words: 22q11.2 deletion syndrome, Apert syndrome, cleft lip, cleft palate, cleidocranial dysplasia, craniosynostosis, hemifacial microsomia, Treacher-Collins syndrome

In 1998 the United States Maternal and Child Health Bureau defined “children with special health care needs” as “children who have or are at an increased risk for chronic physical, developmental, behavioral, or emotional conditions, and who also require health and related services of a type or amount beyond that required by children generally.”²⁸ This group includes but is not limited to children born with clefts of the lip and palate, craniofacial syndromes such as Apert and Treacher-Collins syndrome, and chromosomal abnormalities such as Down syndrome and 22q11.2 deletion syndrome. In addition to multisystem organ involvement, many of these patients experience significant dental morbidity. Consequently, they require coordinated dental, orthodontic, and surgical treatment plans. Coordinated care among surgeons, dental professionals, and other specialists is the standard of care for individuals with clefts of the lip and palate and craniofacial conditions. This article describes the oral and associated manifestations of a select group of patients with special health care needs.

Cleft Lips and Cleft Palates

Cleft formation is a complex embryonic process. Isolated clefts of the palate involve the uvula minimally and can involve the entire secondary palate (up to the incisive foramen). Unilateral clefts of the lip and palate involve the primary palate (lip to incisive foramen) as well as the secondary palate (uvula to incisive foramen). Bilateral clefts of the lip and palate involve the primary and secondary palates. Furthermore, clefts can be complete, as described above, or incomplete

Abbreviation Used: HFM, hemifacial microsomia

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involving less tissue. Genetically and embryologically, clefts of the primary palate and clefts of the secondary palate are distinct entities.

The prevalence of complete clefts of the lip and palate (unilateral and bilateral) is higher than that of isolated clefts of the palate. Although the prevalence varies among populations, on average 1 in 750 infants is born with a unilateral or bilateral cleft of the lip and palate. Interestingly, the prevalence of isolated cleft palates, which is 1 in 2000, is constant across populations. The frequency of systemic malformations associated with unilateral and bilateral clefts of the lip and palate is 26.2% while that of isolated cleft palates is 51.7%.⁵ Boys are affected with clefts of the lip and palate twice as often as girls, but girls are affected with isolated clefts of the palate twice as often as boys.

The clefting process and subsequent surgical interventions both contribute to clinically observable oral manifestations. Complete unilateral cleft lip and palate is a risk factor for poor midfacial growth and frequently results in severe anteroposterior jaw disharmony. There is a general retrusion of the entire profile, including the nasal bone and both upper and lower jaws. Class III malocclusion is commonly observed (Table 1). The cranial base angle is more obtuse in a unilateral cleft population than in the unaffected population. The vertical height of the anterior upper midface is reduced while that of the anterior lower midface is increased. The result is an increase in the total height of the face. Hypoplasia of the posterior maxilla contributes to overeruption of posterior permanent teeth and undereruption of anterior teeth. The combined result is reduced overbite and overjet.³⁴

The profile of an adolescent with a complete unilateral cleft lip and palate is characterized by a relatively large nose and a retrusive lip emphasized by the underlying maxillary hypoplasia.¹⁰ In an adolescent with a complete bilateral cleft lip and palate, the premaxilla protrudes and Class II malocclusion is often observed. This configuration improves with growth, and in adulthood the maxilla often achieves the same anteroposte-

Table 1. Classification of Dental Occlusions

Class	Maxilla	Mandible	Overjet	Clinical presentation
I	Within normal limits	Within normal limits	Within normal limits	Maxillary incisors in contact with mandibular incisors
II	Prognathic	Retrognathic	Excess	Maxillary incisors anterior to mandibular incisors
III	Retrognathic	Prognathic	Reverse	Mandibular incisors anterior to maxillary incisors

rior position as in a noncleft individual.³⁰

In individuals with a bilateral cleft lip and palate, the nasal bone is longer and the base is wider.³⁹ The length of the anterior cranial base is disproportionately smaller than the total length of the cranial base. The length of both the maxilla and mandible is reduced.⁴³ Similar to individuals with a unilateral cleft lip and palate, the height of the posterior maxillary alveolus is hypoplastic. Concomitantly, the posterior permanent teeth overerupt.⁴³ The maxillary incisors are overerupted and retroclined. They can be improved with orthodontic treatment and growth but usually retain some retroclination.

Besides the obvious discontinuity of tissue surfaces and scarring associated with surgical repair, many oral manifestations are associated with clefting. Natal or neonatal teeth are far more prevalent in the cleft population than in the noncleft population. Their prevalence is 2% in infants with a unilateral cleft and 10% in those with a bilateral cleft. Unlike the noncleft population, neonatal teeth in the cleft population are usually present in the maxillary incisor region. Neonatal teeth in the cleft population are most often found in male babies.⁸

In the case of a child with a unilateral cleft lip and palate, the lateral incisor, when present, erupts distal to the cleft in both the primary and permanent dentition 82.4% of the time.⁴⁴ Approximately

half of all children with a unilateral cleft lip and palate with a primary cleft side lateral incisor are missing the permanent cleft side lateral incisor. In rare instances a child with an absent cleft side primary lateral incisor has the permanent cleft side lateral incisor.⁴⁰ The cleft side permanent lateral incisor is absent 50.2% of the time. In 10.9% of patients with a complete unilateral cleft lip and palate, the noncleft side permanent lateral incisor is missing.²³ After the cleft side lateral incisor, the maxillary left second premolar is the tooth most often absent.²³ One in four patients with a unilateral cleft lip and palate has an absent or supernumerary tooth not involving the teeth adjacent to the cleft.⁴⁴ Typically, more severe clefts are associated with absent teeth.

After absence of the cleft side lateral incisor, the most common abnormalities involve the size and shape of the cleft side tooth. Enamel defects of cleft side central incisors are significantly more prevalent when compared to the noncleft side. Permanent cleft side incisors have more enamel defects than primary cleft side incisors. In both primary and permanent dentitions, the defect is most often a yellow opacity of the incisal third.²⁵ The obvious prevalence of associated oral anomalies in individuals with clefts of the lip and palate underscores the need for early observation and carefully planned intervention.

Apert Syndrome

Craniosynostosis, the premature fusion of one or more calvarial sutures, occurs in 1 in 2500 births.²⁹ This congenital defect is mostly an isolated finding. However, it is also part of about 100 different syndromes, including Muenke, Crouzon, Jackson-Weiss, Pfeiffer, Beare-Stevenson, and Apert syndrome among others.

Apert syndrome occurs in 1.5 of 100,000 births.⁴² It is characterized by midface hypoplasia, bilateral symmetric hand and foot syndactyly, and craniosynostosis. The syndrome has an autosomal dominant mode of transmission. Most cases are from sporadic mutations.

Individuals with Apert syndrome have severe malocclusions caused by significant craniofacial and dental abnormalities that require the coordinated services of a craniofacial team to treat. Minimally, at birth the coronal sutures are fused, and a wide midline calvarial defect is present. Cranial-base abnormalities include reduced length of the anterior cranial base and platybasia.³ These abnormalities contribute to midface deficiency and an anterior open bite. Orbital hypertelorism, shallow orbits, ocular motility disturbances, a depressed nasal bridge, a "bird-beak"-shaped nose, and a trapezoidal-shaped mouth are also common findings.

An isolated cleft palate is observed in about a third of patients. In the absence of a cleft, the soft palate is long and thick, a feature that sometimes contributes to respiratory distress. In contrast, the hard palate is short. Clinically, the palate is high and arched. It is typically described as having a "byzantine" shape. A deep midline palatal furrow accentuates with age as mucopolysaccharides accumulate. Subsequent compression of the dental arch and midface hypoplasia combine to produce a V-shaped maxilla that is deficient in the transverse dimension.

Ectopic eruption of the maxillary first permanent molars and palatal eruption of the maxillary second premolars are common. Supernumerary teeth and absence of teeth have been reported. Dental delay of one year is present by age 6 and increases with time.¹⁹ Severe crowding and

Class III malocclusion with a posterior cross bite are universal findings. Patients with Apert syndrome always require careful dental monitoring and frequently require staged orthodontic care.

22q11.2 Deletion Syndrome

The deletion of chromosome 22q11.2 is one of the most common human genetic syndromes with an estimated incidence of 1 in 4000 live births.⁶ This deletion accounts for almost all cases of velocardiofacial, DiGeorge, and conotruncal anomaly face syndromes.⁹ Significant phenotypic variability is associated with the 22q11.2 deletion syndrome, but the basis of this variability remains unclear. The wide range of clinical features includes but is not limited to congenital heart disease, palatal abnormalities, neurologic manifestations, psychiatric findings, learning disabilities, immune dysfunction, feeding difficulties, hypocalcemia, renal abnormalities, characteristic facies, and oral manifestations.²⁷ These features have many clinical implications including the possible need for antibiotic coverage and the likelihood of reduced salivary flow related to systemic medication side effects.

A dysmorphic facies is common in childhood and decreases with growth into adulthood. Characteristic findings include hypertelorism with downslanting palpebral fissures; low-set and malformed ears; and a nose with a large root, narrowed alar base, and short tip. Children have thin lips and microstomia.¹³ Vertical maxillary excess and general facial muscle hypotonia may contribute to a long face. Platybasia (obtuse cranial base angle) may explain the observed facial flatness and mild retrognathia.¹

Palatal dysfunction is manifested by velopharyngeal incompetence (29%), submucosal cleft palate (15%), overt cleft palate (11%), bifid uvula (5%), and complete cleft lip and palate (1%). An additional 22% have suspected but undocumented palatal dysfunction. Only 16% of patients exhibit no palatal dysfunction.²⁷

Children with a 22q11.2 deletion have an increased incidence of oral abnormal-

ities. Enamel irregularities in the primary and permanent dentition have been reported, and hypomineralization of enamel has been linked to the patient's medical condition.²⁰ Hypodontia has been noted in 13% of patients and dysmorphic crown formation in 15%.²¹ A resultant increase in dental caries has been observed. Impaired oral health has been reported in 28% of patients. Dental crowding, a single maxillary incisor, a missing mandibular central incisor, and delayed eruption and formation of permanent teeth have all been reported.^{14,16,21} Due to the high prevalence of oral anomalies in children with the 22q11.2 deletion syndrome, careful dental management and optimal preventive services are suggested.

Oculoauriculovertebral Spectrum

The oculoauriculovertebral spectrum of anomalies includes Goldenhar syndrome and HFM. After cleft lip and palate, HFM is considered to be the most common craniofacial anomaly. It is characterized by mandibular hypoplasia and by ocular and auricular anomalies. Its prevalence is 1 in 5600, and it affects males and females in a 3:2 ratio.¹² Goldenhar syndrome is at the more severe end of this spectrum of anomalies. In addition to the above anomalies, epibulbar dermoids and vertebral abnormalities also occur.

The origin of oculoauriculovertebral anomalies remains uncertain. It is known to affect the formation of structures derived from the first and second branchial arches. There is evidence for vascular disruption and a genetic contribution. Currently, its origin is considered multifactorial. Historically, the oculoauriculovertebral spectrum of anomalies has been described as a unilateral deformity. However, evidence suggests that both sides of the mandible and face are affected to some degree.³ Major organ involvement includes cardiac, central nervous, pulmonary, renal, gastrointestinal, and skeletal systems.

In addition to hypoplasia of the mandible, the malar and temporal bones

and associated soft tissues are underdeveloped. There is an association between the bony malformation and underdevelopment of the muscles of mastication.⁴¹ On the most affected side, the parotid gland may exhibit hypoplasia or aplasia. Unilateral or bilateral cleft lip and palate and cleft palate are documented in 10% of patients, and macrostomia is present in 62% of cases. Reduced palatal width with asymmetric lifting of the soft palate, velopharyngeal incompetence, and hearing loss are frequent findings.¹² Depending on the severity of the deformity, the temporomandibular joint may be functionally normal or completely absent. There is a resultant Class II asymmetric skeletal malocclusion. Consequently, a dental midline deviation, occlusal plane cant, and crossbite are usually present.

Patients with oculoauriculovertebral anomalies have dental delay on the most affected side.¹¹ Hypodontia is more common in patients with oculoauriculovertebral anomalies than in the unaffected population. Its incidence has been reported to range from 6.7% to 26.9%.^{22,26} Enamel defects have been observed. On the most affected side, the mesiodistal diameter of the mandibular primary molars and of the mandibular permanent molars is reduced.¹⁸ Of greater interest perhaps is that the width of both the maxillary and mandibular permanent and primary molars is reduced on the less affected side.³⁶ Patients with oculoauriculovertebral anomalies show significant craniofacial and dental abnormalities, which highlight their need for coordinated surgical, orthodontic, and dental care.

Treacher-Collins Syndrome

Treacher-Collins syndrome, also known as mandibulofacial dysostosis and Franceschetti syndrome, is a disorder involving structures derived from the first and second branchial arches. The causative gene, which has been identified, is transmitted as an autosomal dominant trait with variable expressivity and penetrance. The prevalence of Treacher-Collins syndrome is 1:50,000.³⁵ More than half of new cases are considered new mutations. Hallmark features include severe man-

dibular hypoplasia, eyelid colobomas, and characteristic facies.

The distinctive appearance of a patient with Treacher-Collins syndrome results from severe hypoplasia of facial bones, including the mandible, portions of the frontal bone, supraorbital ridges, hypertelorism, and possible overt clefting of the malar bones. Eyelid colobomas form in the outer third of the lower lid. Cilia are partially absent, and the palpebral fissures slope downward. The ears are often microtic, malformed, and malpositioned; hearing loss is common. Because facial features are underdeveloped, the nose appears large.

Cephalometric analysis reveals a reduced cranial-base angle that decreases further with age.³¹ This reduction has important implications for obstructive sleep apnea, which is a significant complication of Treacher-Collins syndrome. The mandible has a short ramus, short corpus, and poorly developed symphysis.² The gonial angle is obtuse with severe antegonial notching of the lower border. The mandibular plane is particularly steep. The maxilla is vertically deficient and sometimes posteriorly positioned with a clockwise rotation of the occlusal plane.^{33,38} Resultant skeletal open bite, long lower facial height, and Class II and III malocclusions are common.

Significant oral manifestations include cleft palate or, less frequently, cleft lip and palate in 35% of patients and macrostomia in 15% of patients. In the absence of a cleft palate, a high, arched palate is common. The most frequent dental anomaly is agenesis, primarily mandibular second premolars. Hypoplastic enamel is found in 20% of patients, and the maxillary first permanent molars erupt ectopically in 13.3% of patients.⁷ The parotid glands may be hypoplastic. Patients with Treacher-Collins syndrome have a high incidence of dental anomalies and should be screened carefully.

Cleidocranial Dysplasia

Cleidocranial dysplasia is a rare inherited disorder often characterized by persistent open fontanelles, aplasia or hypoplasia of the clavicles, short stature,

multiple supernumerary teeth, and other skeletal abnormalities. Hypertelorism, small or absent nasal bones, and a broad nasal base are observed. Lateral cephalography shows true wormian bones and reduced or absent paranasal sinuses. The height and width of both the maxilla and mandible are underdeveloped.¹⁷ Closure of the mandibular symphysis is often delayed, and a relative mandibular prognathism can result. Clinically, the face is brachycephalic with reduced height of the upper anterior face.¹⁵ Class III malocclusion is most often observed.⁴

Clefts of the hard and soft palates and submucosal cleft palates have been documented.⁴⁵ The noncleft palatal configuration is often described as high and arched. Significant morbidity is associated with major dental abnormalities. Primary dentition develops and erupts normally. However, it does not exfoliate normally because the roots of the primary teeth are not restored. Multiple supernumerary permanent teeth and noneruption of permanent teeth are almost always observed. Crown morphology of the supernumerary teeth is often abnormal. The eruption of permanent dentition has been delayed 2 to 3 years, and permanent first molars have erupted without surgical or orthodontic intervention.³⁷ The mandibular molar root can be significantly longer than normal in patients with cleidocranial dysplasia.³⁶ Supernumerary teeth are most often observed in the mandibular premolar area and in the maxillary incisor and canine area.³² Cellular cementum of permanent teeth is lacking, and the acellular cementum is hyperplastic.²⁴ Abnormalities of root morphology are common. A significant handicapping malocclusion often necessitates a long-term, coordinated orthodontic and surgical treatment plan.

Conclusion

The oral health of an individual is interrelated to systemic health and overall well being. Children with special health care needs experience oral manifestations at a higher rate than unaffected children. These documented oral manifestations are often responsible for significant mor-

bidity. Early screening, careful monitoring, and coordination of related services are important components of the comprehensive care of children with special health care needs.

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The Rotating Stool Test: Diagnosis and Management of Benign Positional Molding

Harold L. Rekate, MD

The Rotating Stool Test was developed to assess the value of a maneuver for the diagnosis of BPM. A management scheme was derived from the test based on the assessment of children referred for evaluation of abnormal head shapes. During the Rotating Stool Test, the examiner holds a child being assessed for abnormal head shape while the parent (usually the mother) sits in a fixed position across the room. As the examiner's stool is rotated from side to side, the child attempts to continue to make eye contact with the parent. This test detects subtle limitations in the child's neck mobility. No child with BPM had a normal test. Invariably, children with BPM are unable to rotate their head away from the flattened side of the head. They either break eye contact with the parent and look at the examiner, or they rotate their entire body toward the parent. All patients with BPM have restricted neck movement as part of the pathophysiology of the condition. Mechanical treatments that concentrate on neck mobility are an important part of the management of this type of skull deformity.

Key Words: benign positional molding, occipital plagiocephaly, rotating stool test, skull deformity

Abbreviations Used: BPM, benign positional molding; CT, computerized tomography; SCM, sternocleidomastoid; SIDS, sudden infant death syndrome

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Infantile skull deformity from mechanical factors is known by various names, including occipital plagiocephaly, BPM, and nonsynostotic deformational plagiocephaly. Each term has disadvantages and advantages. In this discussion, I use the term BPM. BPM involves distortion of the infantile head with flattening of the occiput on one side and milder compensatory contralateral frontal flattening and bulging of the ipsilateral forehead and contralateral occiput. The ear on the side of the occipital flattening is displaced forward and inferiorly.

Two synostotic conditions, both of which may require surgical management, must be distinguished from BPM. Frontal plagiocephaly is caused by unilateral coronal craniosynostosis. It involves an upward and lateral distortion of the orbit called the "harlequin eye" deformity. The prominent deformity of the forehead and mild-to-moderate deformation of the occiput are specific to this condition.

Unilateral lambdoid craniosynostosis may be the rarest form of single suture craniosynostosis and the most difficult to distinguish from BPM. The craniofacial program at the University of Washington in Seattle has done considerable work to make this differential diagnosis easier to make. Unilateral lambdoid synostosis involves an ipsilateral mastoid bulge. The involved occiput is flattened, but the other skull changes are quite different from BPM. In true lambdoid synostosis, the flattened occiput is on the same side as the flattened forehead. Therefore, the shape of the head is that of a trapezoid rather than the parallelogram associated with BPM.^{3,4,9}

Radiographic studies, particularly CT, can confirm the presence or absence of true craniosynostosis. However, BPM is primarily diagnosed by clinical examination. The finding of a parallelogram-shaped head when viewed from the vertex position, as described above, almost certainly reflects mechanical distortion of the head. During the examination of many babies who were referred for this condition, I noticed that the infants had difficulty maintaining eye contact with their parent when they were turned away from the side of the flattened occiput. This simple test also confirms the mechanical nature of the condition and that the abnormality is related to a disturbance of the musculature of the neck. This test, not previously published, has been adopted by the American Academy of Pediatrics as part of its guidelines for the diagnosis and management of skull distortion in infants.^{14,18}

This article describes the Rotating Stool Test, how it is performed, how it leads to the diagnosis of BPM, and its value in emphasizing the importance of neck-stretching exercises in the management of this condition. Although the literature emphasizes the role of shortening of the SCM muscle, shortening of the trapezius muscle may be responsible in a significant percentage of cases.

The Rotating Stool Test

This test evolved spontaneously in my office and reflects my personal approach to the examination of these babies. From the pediatric surgical literature, I was aware of the presence of sternomastoid "tumors."^{11,12,16} Consequently, I examined the neck of each child but rarely, if ever, palpated a mass or lump. Typically, I begin the examination of a baby by picking up and holding the infant in my arms to examine the shape of the head and to palpate the fontanel. I usually sat on a rotating stool facing the parent, typically the mother. As the conversation proceeded, I tended to move the stool around and to rotate it from side to side. When I rotated the stool toward the side of the occipital flattening, it soon became obvious that the baby had no trouble

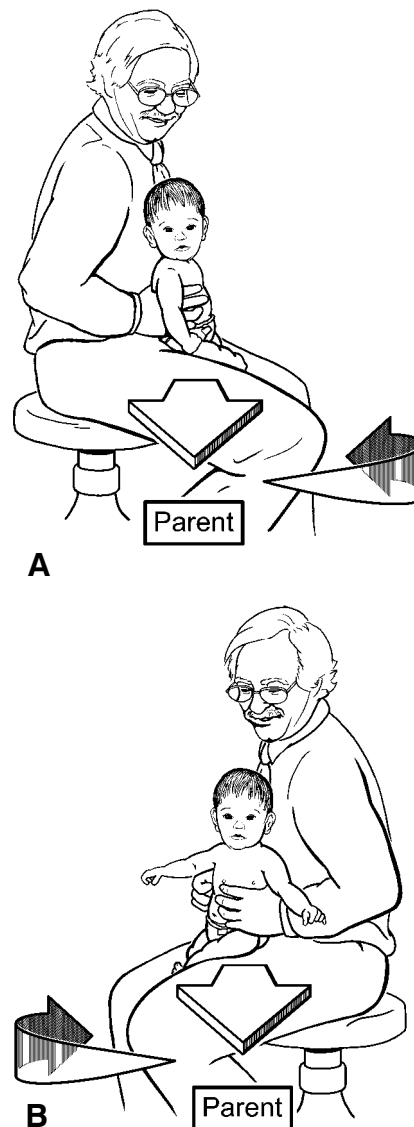


Figure 1. Artist's representation showing how to perform the Rotating Stool Test. (A) When the child is rotated toward the side of flattening, the neck easily rotates to bring the chin to the shoulder. (B) When rotated away from the side of the flattening, the child needs to break the contact or rotate the trunk to maintain it.

maintaining eye contact with the mother and could move the chin all the way to the ipsilateral shoulder (Fig. 1A). In contrast, when I rotated the chair in the other direction, the baby had difficulty maintaining eye contact. The baby either turned toward the flat side and looked up at me or attempted to maintain contact with the mother by rotating the entire upper body (Fig. 1B).

This test has now been performed on more than 300 babies referred for diagnosis and treatment of this condition. All babies had significant difficulty maintaining eye contact with their parent when the stool was rotated away from the side of the flattening.

The condition known as sternomastoid torticollis leads to this form of occipital plagiocephaly. This muscular torticollis is considered responsible for about a third of the cases. When a specific set of tests of sternomastoid function was performed, the coexistence of SCM dysfunction associated with BPM increased to 76% while cervical muscular torticollis was diagnosed in only 12%.⁷ Direct examination of the neck for muscular torticollis is insufficiently sensitive to detect subtle asymmetry of neck rotation. When the rotating stool test is used, 100% of babies with BPM will be found to have subtle problems with neck mobility.

Clinical Implications

What is the value of this simple clinical assessment? First, it is very unlikely that babies with true craniosynostosis will have limited neck mobility. Therefore, this test is an excellent way to confirm the diagnosis of a mechanical condition rather than a genetically determined sutural closure. Second, and certainly more importantly, the parents recognize that the problem is mechanical and related to neck mobility. In my opinion, the most important form of treatment for this condition should focus on the underlying cause. Hence, the parents need little further encouragement to perform the needed neck-stretching exercises.

Initially, all babies with BPM should be managed with neck-stretching exercises. Other authors have emphasized laying the babies prone when awake ("tummy time") and restructuring the child's environment to encourage the child to change positions. However, limited neck mobility often renders these forms of treatment futile.^{10,15} In contrast, my protocol for the management of BPM strongly emphasizes stretching the muscles of the neck (Fig. 2).

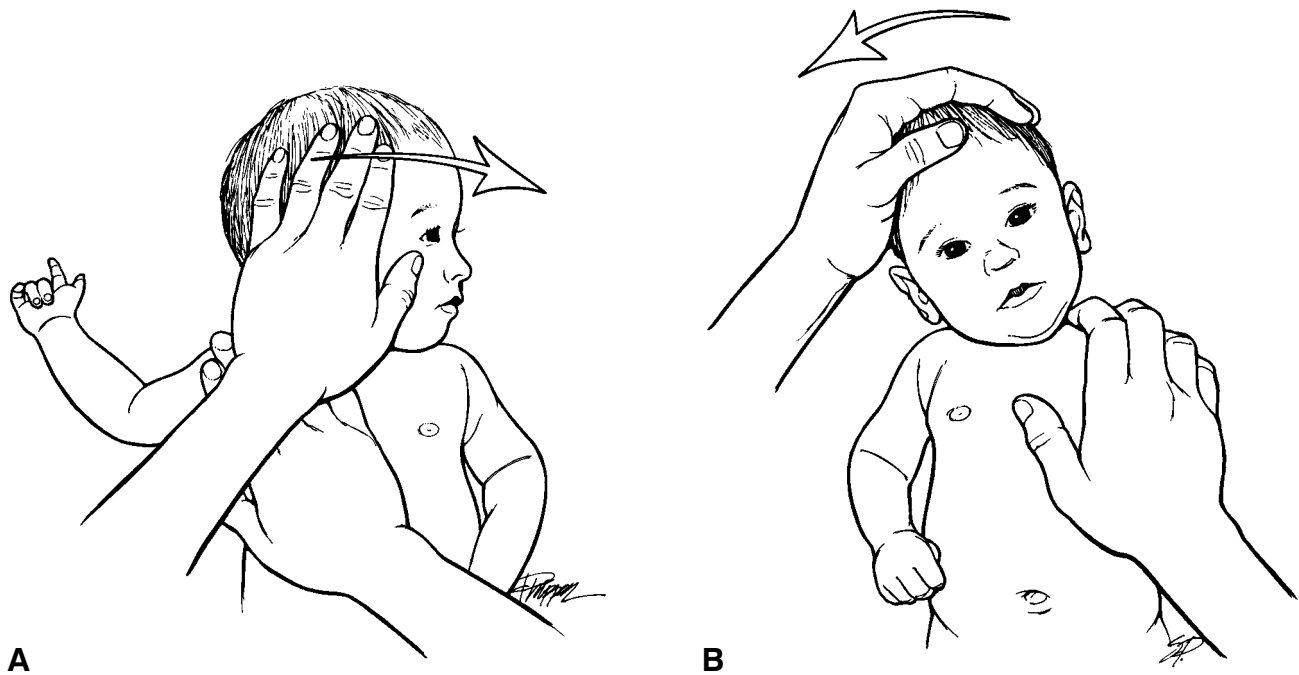


Figure 2. Artist's representation of the neck-stretching exercises derived from the Rotating Stool Test. (A) Neck rotation for assessing and stretching the SCM muscle. (B) Neck tipping for assessing and stretching the trapezius muscle.

To stretch the SCM muscle, the child is positioned on the changing table. The person changing the diaper holds the child's chest gently in position with the nondominant hand while holding the child's forehead with the dominant hand. The head is then firmly rotated until the chin lies on the shoulder. This position is held for 10 seconds (count one 1000, two 1000, and so on). The head is then rotated in the opposite direction and maintained for 10 seconds. Next, attention is paid to the trapezius muscles. The head is tipped to the side until the ear lies against the shoulder, and this position is held for 10 seconds. The maneuver is then repeated on the contralateral side. These four exercises are repeated two more times each so that each stretch is performed three times.

I believe that it is essential to do these stretches frequently. Consequently, I ask that all caretakers learn how to do them and for the exercises to be performed at each diaper change. The babies tend to be frightened for the first few days, but the exercises soon become part of their normal routine.

A baby always resists one of these exercises more than the other three. This tendency makes it possible to diagnose the specific muscle that is involved. As mentioned, 76% of cases of BPM can be explained by a shortening or tightening of the SCM muscle. In my experience, about a third of the babies needing these neck-stretching exercises have specific tightness of the trapezius muscle rather than the SCM muscle. The trapezius extends the head, unilaterally rotates the head up, and tips the head to the contralateral side. Cases of BPM that cannot be explained by abnormal function of the SCM muscle probably involve the trapezius muscle. All BPM likely stems from in utero constriction and shortening of the strap muscles of the neck.

Back to Sleep and BPM

The baseline prevalence of BPM is uncertain. Skull deformity has been found in skeleton remains of historic societies.¹⁹ Before the *Back to Sleep* program was instituted by the American Academy of Pediatrics, about 3% of ba-

bies were found to have BPM; most of these cases resolved within the first few months of life.^{1,2} Two processes have now converged to make the diagnosis and treatment of this condition more common. The first was the dissemination of information as sophisticated craniofacial programs were developed. Parents became aware of their children's deformities and were referred to centers where appropriate diagnoses could be made.

The second process was the recognition of the direct relationship between SIDS and prone-sleeping positions in 1992. This awareness led to the *Back to Sleep* program, which encouraged mothers to maintain their babies in a supine or side-lying position and to avoid the prone position except when the babies were under constant observation and completely awake.⁵ The success of the abandonment of prone lying has primarily been attributed to media attention. Happily, the incidence of SIDS has decreased 40%.⁶ An unexpected consequence of this program, however, was a dramatic increase in the number of children referred and treated for BPM.^{10,18}

Personal Approach to the Management of BPM

Early recognition of BPM or occipital plagiocephaly leads to the most effective management of the condition. Unfortunately, the condition can rarely be diagnosed during the newborn period. In a well-designed Dutch Study of 380 well newborns, 23 (6%) had significant degrees of asymmetry only 9 of which were asymmetrical at 7 weeks of age. During that time, 75 other babies developed significant asymmetry. Consequently, the overall percentage of babies with significant BPM was 22% (84 of 380). The flattening had occurred during the interim at the time of maximum skull plasticity. All babies receiving well baby care are seen at about 7 to 8 weeks of life for their initial inoculations. At that time, the examiner should view the baby from the top of the head. If flattening is noticed at that time and neck-stretching exercises are instituted promptly, it is unlikely that other forms of treatment will ever be necessary.

Cranial remodeling helmets or bands effectively manage BPM.^{1,2,8,13} However, these devices are expensive. The use of the band is analogous to orthodontia in that the bands require frequent adjustment to maximize their benefit. Which children require the use of the bands? After a thorough review of the literature on cranial asymmetry to determine the late negative outcomes associated with this condition,²⁰ I found only four cases of adults who required surgery for this condition. Two of these four adults underwent lysis of the SCM muscle for torticollis. There were no reports of orthodontic or difficulties with the temporomandibular joint, no association with strabismus, and no functional deficits as a result of expectant treatment for this condition.²⁰

Anecdotally, after observing large numbers of normal individuals in social situations such as theaters and sporting events, I have identified only two adults who would have wished to have been treated for this condition during infancy. One was a scientist with a large head and severe plagiocephaly who had considerable trouble finding a football helmet that fit.

Informally, I have also polled a large number of individuals who should be familiar with BPM if the condition were widespread. Barbers, hairdressers, and dentists have all denied being aware of the condition. The only professionals who consistently recognized the problem were opticians. They had a difficult time fitting eye glasses for such patients and occasionally needed to fit a pair with one wing shorter than another.

I participate in many educational events to make primary care physicians and nurse practitioners aware of this problem so that it can be diagnosed during early infancy. If the neck-stretching exercises can be commenced before the age of 6 months, it is unlikely that any other form of treatment will be necessary. Most of these patients are now cared for by their primary care physicians, but I have been active in the assessment of their need for cranial remodeling bands to help patients obtain payment from their insurance plans. I have testified before the new devices committee of the Arizona Health Care Cost Containment System, which is the state of Arizona's Medicaid health maintenance organization. I have recommended that all patients be assessed as early as possible after their first inoculation and that they begin the exercises. If the condition has not improved considerably after 2 months, the patient should be considered for cranial remodeling therapy. This strategy has led to improved care and decreased demand for cranial remodeling bands.

Several studies have shown that mechanical treatment of BPM is effective.¹⁷ One study compared mechanical treatment to orthotic treatment. Both were effective, but the time required to achieve a good result was three times longer in the patients treated mechanically compared to the banded patients.¹⁵ Early diagnosis and mechanical treatment of the torticollis are the primary ways that BPM should be managed. The use of cranial remodeling bands should be reserved for patients referred late or for those unresponsive to neck-stretching exercises. The official position of the American Academy of Pediatrics relative to surgery for this condition is cautious. They do

not rule out the possibility that surgery may be necessary in some cases referred late or resistant to treatment, but it should be limited to the most severe cases.¹⁸

Conclusion

Cranial asymmetry and occipital flattening are common, and their incidence has grown since supine-lying strategies were adapted to prevent SIDS. The Rotating Stool Test can help diagnose positional molding and points to the importance of mechanical treatment such as neck-stretching exercises in the management of this common condition.

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