Skull deformities
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Biologic variation allows the unique craniofacial character and the many asymmetries found in each individual. The human face has more basic, divergent craniofacial patterns than most other species [1]. By altering its own environment, the human species may have been able to reduce its own natural selection pressures and homogeneity and thus increase its phenotypic variability [2]. This article focuses on recognizing where this spectrum of normal morphologic variation in shape and size ends and the pathology or dysmorphology of the skull deformity begins. Pediatric skull deformity is the most common reason for referral to a pediatric neurosurgeon, and pediatricians and neurosurgeons frequently collaborate in diagnosis, treatment, and follow-up. This article discusses the evaluation and management of a range of skull deformities encountered by the pediatrician. Particular attention is paid to distinguishing normal variants from true pathologic conditions and to determining when a child should be referred for neurosurgical evaluation.

Development of the neurocranium

Embryology and the normal neurocranium

Understanding the development of a skull deformity requires an understanding of normal cranium development and morphology. Growth of the neurocranium is largely determined by growth of the brain. The brain grows rapidly, reaching 90% of adult head size by age 1 year and 95% by age 6 years, with growth essentially ceasing at 7 years. By the end of the second year, the bones have interlocked at the sutures, and further growth occurs by accretion and absorption [3]. Cranial
anomalies thus develop most commonly within the prenatal period and in the following period of skull growth. It is this period of development that must be examined for further understanding the genetic and environmental causes of craniosynostosis and the resulting abnormal skull morphology.

The neurocranium is embryologically divided into the vault (calvarium), formed from membranous bone, and the basicranium, formed in cartilage. The initial neurocranium development depends on the formation of the brain and its surrounding membranes including the dural membrane. Brain absence has been shown to result in acalvaria [4]. The initial surrounding membrane is derived of mesoderm and neural crest ectomesenchyme and is subdivided into an inner endomeninx and outer ectomeninx. The outer ectomeninx further subdivides into an outer osteogenic layer in which the ossification centers form skull bones and an inner layer of dura mater [5]. The inner endomeninx becomes the outer arachnoid layer and inner pia mater. The ossification centers form frontal, parietal, squamous occipital, and squamous temporal bones, whereas the intervening areas form fibrous sutures and fontanelles. The sutures generally fuse at the end of the second year of life. The diamond-shaped anterior fontanelle, which is the largest fontanelle (approximately 4 × 2.5 cm), normally closes at approximately 2.5 years of life or sooner. The triangular-shaped posterior fontanelle generally closes at 2 to 3 months of life. The sphenoid (anterolateral fontanelle) and mastoid fontanelles (posterolateral fontanelle) are irregular and small and also generally close by 2 to 3 months of age [3,5].

Defects occur in the completion of ossification, resulting in disorders such as cranium bifidum, and in premature closure of the calvarial sutures. Premature suture closure, or craniosynostosis, its resulting skull deformities, and means of

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**Fig. 1.** (A) Major cranial sutures and bones. (B) Normal patterns of skull bone growth perpendicular to sutures. (From Dufresne C, Carson B, Zinreich S. Complex craniofacial problems. New York: Churchill Livingstone; 1992. p. 160; with permission.)
distinguishing these deformities from positional or deformational skull deformities are considered in this article.

**Causes of prematurely fused sutures**

Genetic and environmental factors suspected of causing craniosynostosis have been heavily investigated. In contrast to other craniofacial dysmorphology such as oroclefting many craniosynostoses have been found to result from single-gene mutations; environmental, teratogenic causes are more rare [5]. Of the genetic mutations found to be responsible for craniosynostosis, mutations in the fibroblast growth factor receptors (FGFR 1, -2, -3) and the *TWIST* and *MSX2* genes are most commonly cited [5]. The rare environmental factors that have been linked etiologically to craniosynostoses include rickets (caused by vitamin D deficiency, resistance to vitamin D, chronic renal failure, or hypophosphatemia) and hyperthyroidism [5]. The teratogens that have been implicated in the causation of craniosynostosis include phenytoin, retinoids, valproate, aminopterin/methotrexate, fluconazole, and cyclophosphamide [5]. Most recently, local intrauterine factors related to constraint of the fetus have also been linked to cases of craniosynostosis and are deemed risk factors predisposing to craniosynostosis. These factors include abnormal positioning in utero, overcrowding caused by multiple pregnancy, oligohydramnios, and increased duration of the first stage of labor, all with resulting increased fetal head constraint [5].

**The effects of prematurely fused sutures on skull growth**

Several theories have been proposed to explain the development of the resulting skull deformity once a fused suture is present. In the nineteenth century,
Virchow explained that fused sutures act as barriers to the brain’s normal growth, forcing the brain to expand in abnormal directions and thus creating a visible skull deformity. He observed that, acting as the underlying and expanding matrix of the skull, the brain and its development and growth directly and strongly affect skull growth and shape. He noted that when a suture is synostosed, the rapid force of brain growth is restrained and altered because the brain cannot expand in the direction of the synostosed suture. The brain and the skull shapes are thus forced in dimensions perpendicular to the fused suture as a consequence of a compensatory growth along the adjacent open sutures. This theory is still commonly accepted today [5].

Normal variations in cranial morphology versus dysmorphology

The normal human cranial shape demonstrates diversity related to both genetic and environmental influences and ranges from the extremes of the long, narrow dolicocephalic head with a cranial width-to-length index of less than 0.75, to the mesocephalic head with a cranial width-to-length index between 0.75 and 0.80,
and to the wide, short, globular brachycephalic head with a cranial width-to-length index of greater than 0.80 (Fig. 3) [5]. It superimposed upon this spectrum of normal cranial morphology that dysmorphology may occur.

**Cranial dysmorphology classifications**

Craniofacial dysmorphology has been divided into the three categories of malformations, deformations, and disruptions, depending on the earliest or primary defect in morphogenesis. This division has been created to elucidate further whether all of the anomalies can be traced to one single event or time in morphogenesis and, thus, to aid physicians in understanding the cause and in counseling families about recurrence risk, treatment, and prevention recommended for each anomaly.

Malformations occur during the first 8 weeks of pregnancy during embryogenesis of a specific organ, result from genetic, environmental causes or a combination of the two, and are associated with higher recurrence rates than the environmentally related disruptions and deformations. Disruptions result from destructive processes that occur after organogenesis to a morphogenetically normal tissue or organ (eg, amniotic banding resulting in a cleft lip). Disruptions are thought to be primarily environmental in origin and thus have low recurrence rates [5]. Deformations occur after embryogenesis and result from nondisruptive mechanical forces causing abnormal formations or distortions of morphogenetically normal body parts (eg, positional plagiocephaly). Deformations may be caused by direct local uterine or postnatal positional factors, may resolve with an altered environment, and are associated with low recurrence rates (Table 1).

Depending on their severity and developmental timing, these malformations, deformations, and disruptions can result in consistent patterns of multiple anomalies, which are further classified as either syndromes or sequences. A sequence is a pattern of multiple abnormalities resulting from a single anomaly or mechanical factor, such as an amniotic band sequence or torticollis-plagiocephaly sequence. In contrast, a syndrome is a pattern of anomalies that are pathoge-

<table>
<thead>
<tr>
<th>Features</th>
<th>Malformations</th>
<th>Deformations</th>
<th>Disruptions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Time of occurrence</td>
<td>Embryonic</td>
<td>Fetal</td>
<td>Embryonic</td>
</tr>
<tr>
<td>Level of disturbance</td>
<td>Organ</td>
<td>Region</td>
<td>Area</td>
</tr>
<tr>
<td>Perinatal mortality</td>
<td>+</td>
<td>–</td>
<td>+</td>
</tr>
<tr>
<td>Phenotypic variability</td>
<td>Moderate</td>
<td>Mild</td>
<td>Extreme</td>
</tr>
<tr>
<td>Spontaneous correction</td>
<td>–</td>
<td>+</td>
<td>–</td>
</tr>
<tr>
<td>Postural correction</td>
<td>–</td>
<td>+</td>
<td>–</td>
</tr>
<tr>
<td>Surgical correction</td>
<td>+</td>
<td>±</td>
<td>+</td>
</tr>
<tr>
<td>Relative recurrence rate</td>
<td>High</td>
<td>Low</td>
<td>Extremely low</td>
</tr>
<tr>
<td>Approximate frequency in newborns</td>
<td>2%–3%</td>
<td>1%–2%</td>
<td>1%–2%</td>
</tr>
</tbody>
</table>

*From Mooney PM, Siegel MI. Understanding craniofacial anomalies. New York: Wiley-Liss; 2002; with permission.*
netically related but not known to represent a single sequence, such as Apert’s syndrome [5]. The discussion of syndromes is relevant when reviewing craniosynostosis, because cases of craniosynostosis are classified as syndromic or nonsyndromic. These differentiations are important, because the treatment, associated findings, and prognosis differ between syndromic and nonsyndromic craniosynostosis. Nonsyndromic cases are generally single-suture synostoses involving the metopic, sagittal, or coronal sutures. Syndromic cases include Apert’s syndrome, Crouzon’s syndrome, and Pfeiffer’s syndrome, as discussed later.

**Craniosynostosis classification schemes**

The classifications for craniosynostoses have evolved over time as the understanding of the causes of various craniosynostoses has grown. The first classification scheme was proposed by Virchow in 1851 and is a descriptive, morphologic nomenclature based on head shape and size (Table 2) [5]. These terms are still widely used by physicians and surgeons for describing an anomaly and in planning the recommended surgical intervention. This scheme, however, is solely descriptive and does not make important distinctions that affect prognosis and recurrence, such as distinguishing between identical-appearing deformational or syndromic-related anomalies. A second classification scheme soon evolved as a result of observations of recurrent patterns of associated anomalies and repetitively observed recurrences among families suggesting a genetic cause. These anomalies also occurred with variations in the phenotypic distinctions among the disorders, making the purely descriptive classification scheme less clear. This second scheme attempted to reconcile the descriptive terminology described by Virchow with the newly emerging genetic understanding of the anomalies, although the specific genetics were not yet understood (Table 3) [5]. More

<table>
<thead>
<tr>
<th>Term</th>
<th>Meaning</th>
<th>Suture Involved</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dolichocephaly</td>
<td>Long head</td>
<td>Sagittal suture</td>
</tr>
<tr>
<td>Scaphocephaly</td>
<td>Keel-shaped head</td>
<td>Sagittal suture</td>
</tr>
<tr>
<td>Acrocephaly</td>
<td>Pointed head</td>
<td>Coronal suture, Coronal/lambdoid, or all sutures</td>
</tr>
<tr>
<td>Brachycephaly</td>
<td>Short head</td>
<td>Coronal suture</td>
</tr>
<tr>
<td>Oxycephaly</td>
<td>Tower-shaped head</td>
<td>Coronal/lambdoid or all sutures</td>
</tr>
<tr>
<td>Turricephaly</td>
<td>Tower-shaped head</td>
<td>Coronal suture</td>
</tr>
<tr>
<td>Trigonocephaly</td>
<td>Triangular-shaped head</td>
<td>Metopic suture</td>
</tr>
<tr>
<td>Plagiocephaly</td>
<td>Asymmetric head</td>
<td>Unilateral lambdoid or positional</td>
</tr>
<tr>
<td>Kleeblattschadel</td>
<td>Cloverleaf skull</td>
<td>Multiple but not all sutures</td>
</tr>
<tr>
<td>Craniofacial dysostosis</td>
<td>Midface deficiency</td>
<td>Craniosynostosis with involvement of cranial base sutures</td>
</tr>
</tbody>
</table>

recently, as the actual genes responsible for the best-defined craniosynostosis syndromes have been identified, a third scheme of classification based on molecular genetics has developed. The three systems are complementary, because no single system yet describes completely the molecular pathogenesis, phenotype, prognosis, and recurrence risk.

Table 3
Commonly used clinical genetic classifications

<table>
<thead>
<tr>
<th>Diagnostic Category</th>
<th>Name of Disorder</th>
<th>Cause</th>
</tr>
</thead>
<tbody>
<tr>
<td>Isolated craniosynostosis</td>
<td>Morphologically described</td>
<td>Unknown, uterine constraint, or FGFR3 mutation</td>
</tr>
<tr>
<td>Syndromic Craniosynostosis</td>
<td>Antler-Bixler syndrome</td>
<td>Unknown</td>
</tr>
<tr>
<td></td>
<td>Apert’s syndrome</td>
<td>Usually one of two mutations in FGFR2</td>
</tr>
<tr>
<td></td>
<td>Baere-Stevenson syndrome</td>
<td>Mutation in GFRG2 or FGFR3</td>
</tr>
<tr>
<td></td>
<td>Bailler-Gerold syndrome</td>
<td>Mutation in TWIST heterogenous</td>
</tr>
<tr>
<td></td>
<td>Carpenter’s syndrome</td>
<td>Unknown</td>
</tr>
<tr>
<td></td>
<td>Craniofrontonasal dysplasia</td>
<td>Unknown gene at Xp22</td>
</tr>
<tr>
<td></td>
<td>Crouzon’s syndrome</td>
<td>Numerous different mutations at FGFR2</td>
</tr>
<tr>
<td></td>
<td>Crouzonomesodermoskeletal syndrome</td>
<td>Mutation in FGFR3</td>
</tr>
<tr>
<td></td>
<td>Jackson-Weiss syndrome</td>
<td>Mutation in FGFR2</td>
</tr>
<tr>
<td></td>
<td>Muenke’s syndrome</td>
<td>Mutation in FGFR3</td>
</tr>
<tr>
<td></td>
<td>Pfeiffer’s syndrome</td>
<td>Mutation in FGFR1 or numerous mutations in FGFR2</td>
</tr>
<tr>
<td></td>
<td>Saethre-Chotzen syndrome</td>
<td>Mutation in TWIST</td>
</tr>
<tr>
<td></td>
<td>Shprintzen-Goldberg syndrome</td>
<td>Mutation in FBN1</td>
</tr>
</tbody>
</table>


The pediatrician’s role in identifying cranial vault anomalies

Approximately 2% of infants are born with some extrinsically caused deformity; the remainder of deformities result from an intrinsic abnormality [6]. A rough approximation of craniosynostosis incidence is 0.014 to 0.048 cases per 1000 births or 1 in 2100 to 3000 newborns [5,7]. A pediatrician is the first physician to examine a child and offers the first opportunity for intervention. Thus, pediatrician has a responsibility to be aware of normal variations in cranial shape and size, the clinical findings characteristic of deformational and craniosynostotic skull shapes, and the differences between the two (Table 4).

If a skull deformity is present, the physical examination and clinical history are the most helpful and revealing pieces of information in the child’s evaluation. The pediatrician should be attentive to questioning regarding developmental milestones, irritability, headaches, emesis, and visual complaints. On the exami-
<table>
<thead>
<tr>
<th>Type</th>
<th>Epidemiology</th>
<th>Skull Deformity</th>
<th>Clinical Presentation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sagittal</td>
<td>Most common CSO affecting a single suture, 80% male</td>
<td>Dolicocephaly or scaphocephaly (boat-shaped)</td>
<td>Frontal bossing, prominent occiput, palpable keel ridge, OFC normal and reduced biparietal diameter</td>
</tr>
<tr>
<td>Coronal</td>
<td>18% of CSO, more common in females; Associated with Apert’s syndrome (with syndactyly) and Crouzon’s disease which includes abnormal sphenoid, orbital, and facial bones (hypoplasia of the midface)</td>
<td>Unilateral: plagiocephaly; Bilateral: brachycephaly, acrocephaly</td>
<td>Unilateral: flattened forehead on affected side, flat cheeks, nose deviation on normal side, higher supraorbital margin leads to harlequin sign on radiograph and outward rotation of orbit may result in amblyopia Bilateral: broad, flattened forehead. In Apert’s syndrome accompanied by syndactyly and in Crouzon’s disease by hypoplasia of the midface and progressive proptosis</td>
</tr>
<tr>
<td>Lambdoid</td>
<td>10%–20% of CSO, M:F ratio 4:1</td>
<td>Lambdoid/occipital plagiocephaly; right side affected in 70% of cases.</td>
<td>Unilateral: flattening of occiput, indentation along synostotic suture, bulging of ipsilateral forehead leading to rhomboid skull, ipsilateral ear is anterior and inferior Bilateral: brachycephaly with bilateral anteriorly and inferiorly displaced ears Pointed forehead and midline ridge, hypotelorism,</td>
</tr>
<tr>
<td>Metopic</td>
<td>Association with 19p chromosome abnormality</td>
<td>Trigoncephaly</td>
<td>Tower skull with undeveloped sinuses and shallow orbits, and elevated intracranial pressure</td>
</tr>
<tr>
<td>Multiple</td>
<td></td>
<td>Oxycephaly</td>
<td></td>
</tr>
</tbody>
</table>
nation, special attention should be paid to the alertness of the patient, the head circumference, the pupils and fundoscopic evaluation for papilledema, the fontanelles, and the shape and characteristics of the deformity. The pediatrician should schedule frequent examinations, which would include photographing the child and following the head circumference and developmental milestones. The pediatrician should refer appropriate cases to a neurosurgeon or craniofacial team. Although these diagnoses may be rare, their clinical significance requires diagnosis and referral for further evaluation and treatment recommendations.

*Distinguishing true craniosynostosis from birth molding and positional or deformational lambdoid plagiocephaly*

Differentiating between birth molding, positional plagiocephaly without synostosis, and true synostosis is one of the first distinctions to be made and properly understood. In making this distinction, the history and physical examination provide the most valuable information. Normal birth molding results in findings that resolve within hours to weeks of birth without intervention and is clinically different in timing of presentation and quality from positional or deformational lambdoid plagiocephaly. Characteristically, passage of the infant through the birth canal results in pressure in the anteroposterior dimensions and less in the side-to-side dimension. The calvarium thus grows longer in the vertical dimension as the parietal bones rise away from the skull base and the frontal and occipital bones slip under the anterior and posterior borders of the parietal bones, respectively. This overriding quality is often palpable and visible at birth but resolves, unlike synostosis cases, which are present at birth but progress, or deformational cases, which are generally not present at birth but develop during infancy.

Deformational plagiocephaly results from local pressure on a specific region of the skull, typically in one occipital region resulting in occipital/lambdoid plagiocephaly or the so-called “lazy lambdoid.” The clinical finding of lambdoid plagiocephaly is becoming more frequently seen by pediatric neurosurgeons and in many practices is now the most common reason for referral to a pediatric neurosurgeon. Nonsynostotic plagiocephaly has been reported as occurring in up to 48% of healthy neonates [8]. This high incidence and the clinical similarities to actual lambdoid synostosis have fueled a renewed and widespread interest in the topic of lambdoid plagiocephaly and its treatment. The point of referral to a pediatric neurosurgeon in these cases is making the distinction between a true synostosis and a deformational or positional plagiocephaly without synostosis. At the New York University School of Medicine, it has been observed that cases of true lambdoid craniosynostosis are exceedingly rare and that almost all cases of lambdoid plagiocephaly are deformational and rarely require surgical intervention. This finding has also been supported by results of a study at Boston Children’s Hospital that found that true lambdoid synostosis accounts for only 2.3% of all synostosis cases [8]. This differentiation is best made by acquiring a complete clinical history and physical examination and rarely requires further studies (Table 5).
Risk factors for deformational plagiocephaly that can be identified in the patient's history include intrauterine crowding and positioning, decreased mobility in patients with developmental delay, mental retardation, prematurity or chronic illness, and cases of intentional positioning or abnormal posturing such as torticollis and congenital disorders of the cervical spine [5]. For example, many postulate that the recent increase in the previously rare diagnosis of lambdoid plagiocephaly might be attributed to a recommendation of the 1992 American Academy of Pediatrics “Back to Sleep” campaign [9]. The campaign advocated that newborns be placed in a supine sleeping position, sometimes with a foam wedge to tilt the head to one side to decrease aspiration and the risk of sudden infant death syndrome. Parents of patients with deformational plagiocephaly classically report that their child nearly always lies in one position with the head turned to one side. This history in addition to the physical examination nearly always allows one to distinguish deformational plagiocephaly from true craniosynostosis.

There are several important points to review regarding the physical examination in suspected synostosis. In cases of true synostosis, a palpable bony prominence or ridge is often appreciated over the suspected suture, and gentle firm pressure on either side of the suture by the thumbs fails to cause relative movement of the bones on either side of the suture. In contrast, in cases of positional plagiocephaly, there is no sutural ridging, and movement of the bones may be appreciated. The overall head shape is also different. In cases of deformational plagiocephaly, a parallelogram shift in the cranial shape occurs, resulting in a rhomboid-shaped skull characterized by a flattening of the occiput with an associated bulging of the ipsilateral forehead with the ipsilateral ear sheared anteriorly and inferiorly to the contralateral ear. The contralateral orbit and forehead may also be flattened. These findings are unique to positional plagiocephaly.

<table>
<thead>
<tr>
<th>Finding</th>
<th>Posterior Plagiocephaly Without Synostosis</th>
<th>Unilambdoid Synostosis</th>
<th>Unicoronal Synostosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ipsilateral ear position Flattening</td>
<td>Anterior Ipsilateral occipitoparietal</td>
<td>Posterior-inferior Ipsilateral occipitoparietal</td>
<td>Anterior-superior Ipsilateral frontal with orbital rim elevation</td>
</tr>
<tr>
<td>Vertex view Suture</td>
<td>Parallelogram</td>
<td>Trapezoid</td>
<td>Trapezoid</td>
</tr>
<tr>
<td>Bossing</td>
<td>Ipsilateral frontal, contralateral occipital</td>
<td>Contralateral parietal, ipsilateral mastoid</td>
<td>Contralateral frontal</td>
</tr>
<tr>
<td>Skull base (3D-CT)</td>
<td>No tilt, no midline deviation</td>
<td>Tilt, posterior fossa deviation off midline</td>
<td>Anterior fossa deviation off midline</td>
</tr>
</tbody>
</table>

Table 5
Pertinent features and differential diagnosis of plagiocephaly

unlike the trapezoid-shaped skull with an ipsilateral mastoid bulge, posterior displacement of the ear, and contralateral occipital bossing that develop in true lambdoid synostosis (Figs. 4, 5). In rare, difficult cases when the examination and history are not diagnostic, a skull radiograph is generally the best first diagnostic study to confirm positional plagiocephaly and to demonstrate the patency of the suture. In cases of synostosis, the skull radiograph may show a sclerotic margin.

On confirmation of deformational plagiocephaly, it is necessary to diagnose the cause of the skull deformity and to recommend appropriate treatment to be followed closely by a period of frequent serial evaluations. For cases of infants with poor mobility, repositioning for mild cases or molding-helmet therapy in more severe cases is appropriate. Repositioning has been shown to be effective in improving the deformity in approximately 85% of mild cases. For severe cases or for mild cases that do not improve with repositioning, helmet therapy may be advised [10]. The authors have found that all helmets are equally efficacious and safe and that, despite commercial claims, no one product is superior. One such helmet is the DOC Band (Cranial Technologies Inc., Tempe, Arizona), the first helmet for which approval by the Food and Drug Administration was sought; it is a plastic helmet with foam lining that weighs 6 oz and can be used on infants ranging from 3 to 24 months of age with plagiocephaly, scaphocephaly, or brachycephaly that does not result from true synostosis.

The aim of any helmet is to place mild pressure on prominent areas of the skull while allowing room for flattened areas. The helmet is worn 23 hours per day,
Unilateral lambdoid synostosis

Positional molding

Ipsilateral ear displaced posteriorly (variable)
Ipsilateral occipitomastoid bossing
Pastoral bossing
Flattening

Contralateral occipital bossing
Flattening
and the children are followed closely for weekly or biweekly adjustments of the helmet, documentation of improvement, and examination of the scalp for areas of irritation. If no improvement ensues, further studies or surgery may be contemplated for severe deformities. Mild to moderate deformity resulting from synostosis or deformation rarely warrant surgical correction, because hair growth later covers the defect satisfactorily. At the New York University School of Medicine, satisfactory improvement is typically seen with repositioning and helmet therapy, and surgery for lambdoid plagiocephaly is not indicated. Further, it is important to counsel parents that no evidence supports operating on a single-suture stenosis or plagiocephaly to prevent intellectual impairment or developmental delay, and that an intervention is directed at cosmetic improvement [8].

For children with torticollis, treatment of the torticollis is recommended with or without the addition of helmet therapy depending on the severity of the deformity. Torticollis and nonsynostotic occipital plagiocephaly reportedly occur in 1 in 300 live births [8]. Torticollis is defined as a form of dystonia resulting in a failure to control the head position [3]. It may be congenital or of acquired muscular origin caused by the limited room in the uterus during gestation, muscle spasm, hematoma within the muscle, a cerebellar tumor, or atlantoaxial rotatory subluxation but consistently results in a tightened, shortened sternocleidomastoid muscle on the affected side causing an affected infant to tilt the head to the tight side and turn away [3]. Torticollis is often not detected at birth but is observed during infancy as the neck begins to lengthen and the head tilting becomes more pronounced, with a preferred resting position and possible resulting plagiocephaly. Treatment for torticollis includes early involvement with a physical therapist for neck strengthening and stretching, head-turning, and repositioning exercises. The child may benefit also from resting in the prone position, avoiding positioning such as in walkers or jumpers where stress is placed on the neck, and propping the neck in straight alignment. If no improvement ensues within 1.5 or 2 years, further treatment may be recommended, including options such as local botulinum toxin injection, endoscopic tenotomy or bipolar Z-plasty of the sternocleidomastoid, selective rhiotozotomy, or spinal accessory nerve section followed by placement in collar [3,11]. The authors have found that surgical intervention is needed in only rare cases, and more than 95% of cases of torticollis are successfully treated with physical therapy.

True craniosynostosis: nonsyndromic and syndromic craniosynostosis

Of all patients treated for craniosynostosis disorders, approximately one sixth are affected by syndromic craniofacial malformations [12]. Syndromic cases of
craniosynostosis represent a unique group of patients with associated morphologic and functional abnormalities not seen in simple nonsyndromic patients who may experience isolated craniosynostosis. The separate abnormalities experienced by syndromic patients evolve at different times during the patients’ lives and require treatment by physicians trained in different specialties. Of neurosurgical interest is the increased frequency of hydrocephalus, elevated intracranial pressure (ICP), and Chiari malformations that are uncommon in nonsyndromic craniosynostosis [12]. It is the belief at the New York University School of Medicine that it is imperative that these patients be treated and followed by a multidisciplinary craniofacial team including a pediatric neurosurgeon, plastic surgeon, ophthalmologist, neurologist, otolaryngologist, orthodontist, psychologist, geneticist, and social worker to make the proper diagnosis and to attend properly to all the developing issues that these children encounter [12]. The most common nonsyndromic craniosynostoses include metopic, sagittal, and coronal craniosynostosis. The most common syndromes including craniosynostosis are Apert’s syndrome, Crouzon’s syndrome, and Pfeiffer’s syndrome.

**Metopic synostosis: trigonocephaly**

The metopic suture is the first suture to close and normally does so just after birth. Premature closure of the metopic suture results in trigonocephaly, a characteristic pointed forehead with a midline metopic ridge deformity from the
bossing of the central frontal bone (Figs. 6, 7). If the premature fusion also includes the frontonasal suture, hypotelorism results. More severe cases also display severe constriction of the forehead with resulting symmetric expansion of the skull posterior to the coronal sutures. This diagnosis is usually easily made by the experienced physician and confirmed with radiograph or CT scan. In rare cases, the underlying brain is malformed with both arrhinecephaly and holoprosencephaly observed. These rare cases with associated frontal lobe malformations are associated with mental retardation. Treatment of metopic suture synostosis depends on the severity of the case. Mild cases with only visible or palpable suture often do not require surgical intervention, and the ridge often becomes less noticeable as the child grows. Previously, some neurosurgeons have advocated burring down the keel or removing the keel from the coronal suture to the frontonasal suture; most, including those at the New York University School of Medicine, favor a bilateral cranial vault remodeling procedure.

Sagittal synostosis: scaphocephaly

Sagittal synostosis is the most common craniosynostosis, with a birth prevalence of 1 in 5000 [13]. Sagittal synostosis is identified by the characteristic dolichocephaly or scaphocephaly (i.e., a boat-shaped skull with a markedly reduced biparietal diameter but increasing head circumference) that crosses growth percentile lines during the first few months of life. The head is elongated with frontal bossing and an occipital knob or bullet protuberance (Figs. 8–11).
Frequently, a palpable ridge can be appreciated along the longitudinal midline sagittal suture between the anterior fontanelle and junction of the lambdoid sutures. Sagittal synostosis is more common in males (80%) [13]. As in metopic synostosis, focal neurologic deficits and elevated ICP are rare. Mental retardation is even less common [6].
Surgical treatment by a neurosurgeon with any of the many variations of the classic linear strip craniectomy, including endoscopic strip craniectomy, excising the sagittal suture from anterior to the coronal suture to posterior to the lambdoid suture, is generally recommended within the first 3 to 6 months of life [14]. It is necessary that the excised strip be at least 3 cm in width, because infants have tremendous osteoblastic potential and reossification across the channel and recurrence may occur within weeks if the craniectomy is not sufficiently wide [6]. If diagnosis or recurrence occur after 6 months, a more formal cranial vault
remodeling procedure is necessary, which generally requires a team consisting of a pediatric neurosurgeon and a craniofacial surgeon for optimal results.

**Coronal synostosis: plagiocephaly, brachycephaly**

Coronal synostosis is typically unilateral and accounts for approximately 18% of craniosynostosis. It is more common in females [3,15]. It is also the most common synostosis to be associated with a syndrome, typically when it is bilateral. Sixty-one percent of all coronal cases are sporadic [15]. Unilateral coronal synostosis results in plagiocephaly with the characteristic concave flattening of the forehead and of the elevated supraorbital margin on the affected side and bossing of the frontal bone on the opposite side. This distortion results in a characteristic finding on coronal radiographs called the harlequin sign (Figs. 12–14). The deformity often includes the supraorbital ridge and frontal bone and thus affects the orbital position, resulting in outward deviation of the orbit on the abnormal side and amblyopia. This orbital asymmetry is often the most disturbing aesthetic deformity [6]. If untreated, the tip of the nose may also deviate toward the unaffected side, with the root deviating to the ipsilateral side, and the cheeks may become flattened. Unilateral coronal synostosis is infrequently associated with increased ICP or a congenital malformation. Bilateral coronal synostosis results in brachycephaly with a broad, flattened forehead and

![Fig. 12. Unilateral cranial synostosis resulting in plagiocephaly. (From Dufresne C, Carson B, Zinreich S. Complex craniofacial problems. New York: Churchill Livingstone; 1992. p. 173; with permission.)](image-url)
wide, foreshortened cranium (Figs. 15, 16). The orbital depth is frequently decreased, resulting in exorbitism. It is generally seen in craniofacial dysmorphism in syndromic craniosynostosis with multisuture synostosis.

Previously, coronal synostosis was treated with simple linear craniectomy, with poor results. Presently, the treatment of choice is cranial vault remodeling and fronto-orbital advancement. Surgical correction is usually best accomplished by a craniofacial team including a neurosurgeon and plastic surgeon [15]. It is also necessary that a craniofacial group evaluate patients suspected of having coronal synostosis, because coronal synostosis is the most commonly involved suture found in syndromic craniosynostosis.

Multisuture synostosis: oxycephaly

A short and narrow head shape characterizes multisuture synostosis. The most commonly affected sutures are the sagittal and coronal sutures. In some cases, all of the sutures except the metopic suture are affected, resulting in a condition described as Kleeblattschadel or cloverleaf skull with a characteristic frontal telescoping of the skull referred to as craniotelecephaly. ICP is generally elevated, and the frequency of mental retardation is high. Multisuture synostosis with elevated ICP should be treated on diagnosis to avoid further neurologic damage [15,16]. Surgery should be performed with a craniofacial team including a
neurosurgeon and a plastic surgeon and typically consists of formal cranial vault remodeling and fronto-orbital advancement.

**Crouzon’s syndrome**

Crouzon’s syndrome is an autosomal dominant syndrome with an incidence of 1 in 25,000 births. Mutations in the FGFR2 gene have been found in more than half of studied affected subjects, and the specific gene for the syndrome has been mapped to chromosome 10q26 region [15]. Crouzon’s syndrome is characterized by a broad variability in the severity of craniosynostosis with the coronal sutures nearly always involved (95% of cases), mostly in association with the sagittal and
lambdoid sutures (75% of cases) or only the sagittal suture (20% of cases) [15]. Brain malformations are infrequent, and mental retardation is rare, distinguishing this syndrome from other syndromes with similar craniofacial features. The craniosynostosis is associated with a marked shortening in the depth of the anterior cranial fossa with orbital roof hypoplasia and advancement of the posterior wall of the orbits, all which result in the characteristic and severe ocular proptosis characteristic of the syndrome with its associated secondary exposure keratitis and conjunctivitis. Additional prominent features are midface retrusion, conductive hearing loss in half of the patients, mandibular overjet with an anterior overbite, and a small nasopharynx requiring mouth breathing (Fig. 17).

Apert’s syndrome

Apert’s syndrome is also an autosomal dominant syndrome with features similar to Crouzon’s syndrome that results from a mutation in the FGFR2 gene and has an incidence of 1 in 55,000 births [17,18]. It is similarly characterized by craniosynostosis, most commonly involving bilaterally fused coronal sutures, but mental retardation and underlying brain malformations, including corpus callosum agenesis, cortical atrophy, or hydrocephalus, are more common than in Crouzon’s syndrome [15,18]. The syndrome is most easily distinguished from
Crouzon’s syndrome by the soft tissue and bony syndactyly involving the second, third, and fourth fingers and all toes, which are not seen in Crouzon’s syndrome. Other characteristics that may aid in the differentiation are the less severe hypertelorism and proptosis, occasional cleft palate (which is less common in Crouzon’s syndrome), a beaked nose and flat facies, and down-slanting palpebral fissures. These faciocranial abnormalities are present at birth, unlike those in

Fig. 16. Bicoronal synostosis surgery. (From Texas Pediatric Surgical Associates, Houston, Texas; with permission.)

Fig. 17. Crouzon’s syndrome with bilateral coronal synostosis, midface hypoplasia, exorbitism, and beaked nose. (From Dufresne C, Carson B, Zinreich S. Complex craniofacial problems. New York: Churchill Livingstone; 1992. p. 118; with permission.)
Crouzon’s syndrome, which are generally mild at birth and develop over infancy (Fig. 18).

**Pfeiffer’s syndrome**

Pfeiffer’s syndrome is clinically related to but genetically distinct from Apert’s and Crouzon’s syndrome. It is an autosomal dominant syndrome related to the $FGFR1$ and $FGFR2$ genetic mutations with an incidence of 1 in 200,000 live births [15]. It is characterized by craniosynostosis of varied severity, often with a cloverleaf deformity and mental retardation, varying degrees of syndactyly, broad great toes and thumbs, hypertelorism, a beaked nose, maxillary hypoplasia, mandibular prognathism, and upper airway anomalies and obstruction. The facial deformities are less severe than those of Apert’s syndrome, and the limb anomalies separate this entity from Crouzon’s syndrome (Fig. 19).

**Macrocephaly**

Macrocephaly, or, more specifically, macrocrania refers to a head circumference greater than the ninety-eighth percentile. It is one of the most common reasons for referral to a pediatric neurosurgeon. Macrocrania is most commonly a result of familial macrocrania and represents a normal variation in head size but
may also be a sign of an underlying abnormality requiring further investigation. The keys in making this distinction are the trend in head circumference growth and physical examination. In familial macrocrania, the head circumference may spike at 4 to 6 months of age to above the ninety-eighth percentile but then maintains its curve on the head circumference growth chart over time. Pathologic macrocephaly continues to increase. Patients with familial macrocrania do not exhibit neurologic signs and symptoms. Thus, the first steps in investigating macrocephaly must include a family history of head size, measurements of the parents’ heads, and frequent documentation of a child’s head circumference and neurologic status. Signs that warrant further investigation into the cause of macrocrania include irritability, developmental delay, focal neurologic deficits, a bulging fontanelle, split sutures, papilledema on fundoscopy, or a child’s head size that is out of proportion to the parents’ size and continues to grow beyond the ninety-eighth percentile at a rate above the normal growth curve for that percentile.

**Microcephaly**

Microcephaly is defined as a head circumference more than 2 SD below the mean for gestational age and sex. Microcephaly may reflect a normal familial variation or result from underlying pathologic processes such as a brain malformation or craniosynostosis. It is important to identify microcephaly that results from craniosynostosis, because these patients may benefit from surgical treatment with improved cerebral development, unlike patients with microcephaly resulting from other causes, such as brain malformations.

Fig. 19. Pfeiffer’s syndrome with broad thumbs. (From Dufresne C, Carson B, Zinreich S. Complex craniofacial problems. New York: Churchill Livingstone; 1992. p. 120; with permission.)
Neuroradiologic imaging

In most cases of craniosynostosis, the physical examination and clinical history provide the diagnosis. It is unusual that radiographs or CT are necessary for diagnosis. The goals of imaging in the treatment of craniofacial anomalies are now to demonstrate the relationship of the overlying and underlying tissues and to plan the surgical intervention. The advent of CT has revolutionized this process. CT provides greater detail of the bony anatomy and demonstrates soft tissue anatomy with a three-dimensional reconstruction. MRI and ultrasonography may be used in the future, but the diagnosis will probably continue to rely on patient examination and history.

Timing of surgical intervention

Because brain growth is so rapid and dynamic, timing of surgical intervention is thought to be crucial to both aesthetic and functional success and continues to be a subject of constant debate [6,19–21]. At the New York University School of Medicine, sagittal synostosis is corrected by simple linear craniectomy before 6 months of age. All other synostosis cases and sagittal synostosis diagnosed after 6 months are treated at the time of diagnosis if there is elevated ICP or near 9 months of age when there is no issue of elevated ICP. The craniofacial team has found that this timing allows for the best results because of the quality of bone at this age [6,19]. The cranial vault is still malleable but also is able to withstand the necessary manipulation and instrumentation. “Surgically, the earlier the procedure, often the better. At an earlier age, bone is more malleable and thus easier to remodel; risk of infection is minimized because of lack of development of the paranasal sinuses; healing capacity is greatest; grafting is optimized as membranous bone, the ideal replacement for membranous bone, is plentiful in the infant skull; and operating times and blood loss are decreased. Further, because the brain is most plastic in the infant, any damage occurring during surgery has less likelihood of being dysfunctional” [6].

With age, skull deformation increases in severity and permanency, increasing the number of procedures necessary, the extent of the surgery, and the risk of complications. “The beauty of treating very young, mildly deformed individuals is the simplicity of treatment and the large role that the natural process of brain growth, not the craniofacial surgeon, has in recontouring the skull to normalcy” [6]. Timing of surgery is an active topic of discussion among craniofacial surgeons and neurosurgeons; some consider even less than 6 weeks of age as the ideal time for surgery, whereas others report that surgery before 3 months of age has been associated with a higher rate of recurrence. The authors’ experience has shown that this early timing of surgery helps prevent permanent skull and brain deformity and has the best cosmetic result. A later intervention may be acceptable in cases when other life-threatening or permanently disabling deformities must be corrected first. “Generally speaking, it is best to treat the
craniofacial deformity as early as possible, but in some cases craniofacial surgery may be delayed by more urgent surgical procedures” [6].

**Reasons to treat**

Surgical correction of craniosynostosis may present a formidable challenge to the neurosurgeon and plastic surgeon and risk to the patient. It is therefore essential to understand the goals of surgical intervention and the prognosis and risks incurred by not intervening. These goals and risks are cosmetic, neurologic, and psychologic, and thus treatments are aimed at improving appearance and function. Theoretically, a stenosed calvarium decreases the intracranial volume, thereby constraining the brain, meninges, optic nerves, and other cranial nerves and raising the ICP. Conditions associated with a markedly elevated ICP increase the frequency of mental retardation [15,16]. Studies have demonstrated that ICP increases in proportion to the number and types of sutures involved. Using epidural sensors, Renier [16] demonstrated an elevated ICP (> 15 mm Hg) in 83% of patients with Crouzon’s symptom, in 58% of oxycephalic patients, in 53% of patients with Apert’s syndrome, in 28% of brachycephalic patients, in 14% of plagiocephalic patients, in 8% of trigonocephalic patients, and in 7% of scaphocephalic patients. Renier [6,16] noted that the elevated ICP is further aggravated by increased age and that early surgical intervention is corrective until 6 years of age, when cerebral atrophy begins to compensate. The relationship between craniofacial deformity, elevated ICP, and mental retardation remains controversial, and it may be that the neurologic impairment or hydrocephalus, if present, are not results of the craniosynostosis but rather of an embryologic insult that resulted in both the synostosis and neurologic disease. It is now generally accepted that patients with isolated craniosynostosis should not be at risk for neurologic impairment unless the craniostenosis is severe. Thus, in cases of an isolated suture synostosis, such as metopic or sagittal suture synostosis, the reason for intervention may be primarily cosmetic and psychologic.

A second, less quantifiable, but equally important reason for intervention is the possible psychosocial developmental sequelae of leaving craniofacial patients untreated. Dufresne [6] writes, “The potential emotional handicap associated with growing up with a very abnormally shaped head can be as devastating as an actual physical defect.” Many studies have demonstrated the “decline of positive peer relations and nurturant interactions with adults and the greater chance for deficiency in parent-child relationships for such deformed individuals” [6]. Functional and cosmetic results were studied by Pertshuk and Whitaker [22,23] and reported for two groups of patients, those younger than 18 months of age and those older than 6 years of age. Different qualities of functional results defined as postoperative neurologic or psychosocial improvements were observed in the patients. In the younger group of patients, 41.2% experienced a pronounced burst in development and affect, accelerating through milestones that they had only slowly achieved preoperatively. Older patients did not demonstrate
this developmental burst—the brain was thought to have compensated for the elevated pressure with decreased growth—but almost 100% of this older group did demonstrate a social burst characterized by increased self-esteem and extroversion. The procedure is thus not only cosmetic but is also potentially a neurologically and psychologically beneficial intervention.

**Multidisciplinary craniofacial centers**

The authors believe that pediatric patients with skull deformities, specifically patients with syndromic craniosynostosis, because they are so rare, experience the highest level of care and success in treatment when cared for by a multidisciplinary craniofacial team including a pediatric neurosurgeon, plastic surgeon, pediatric orthodontist, geneticist, otolaryngologist, ophthalmologist, psychologist, and social worker [6]. Craniofacial disease requires a continuing superspecialized evaluation from each member of the team, and this evaluation can be most effectively and consistently offered to a family over a longitudinal course of treatment when the team of professionals is consolidated. In this setting, the physicians are capable of formulating an appropriate treatment plan with consolidated surgeries when possible, performing the planned procedures, and establishing adequate follow-up for evaluating the results.

For example, at the New York University School of Medicine, there is a weekly craniofacial conference in which patients are presented, opinions are given by each member, and a consolidated plan is given to the family and put into action. These patients are then followed by the same group at regular intervals or when necessary and can be regularly evaluated for postoperative concerns and complications such as infection (4.4%–6.2% of craniofacial procedures), delayed development of elevated ICP, rare cerebrospinal fluid leak, seizure, persistent deformity requiring further intervention, upper airway obstruction, or ophthalmologic deficit including strabismus, amblyopia, or papilledema [6,24]. Each member of the team provides different but complementary insight into each case. The geneticist may help in the genetic diagnosis and present other rare associated conditions that must be evaluated and would be otherwise unknown to the surgical group. The preoperative evaluation, surgical intervention, and postoperative care encompass cosmetic and neurologic challenges requiring both a plastic surgeon and neurosurgeon. In cases of syndromic synostosis, there are commonly associated concerns requiring an orthodontist, otolaryngologist, and ophthalmologist. All these children and their families may benefit from the additional perspective and contributions of social workers and psychologists accustomed to their specific and unique needs.

**Summary**

To recognize and make the proper diagnosis of a craniofacial abnormality, it is helpful for the pediatrician to understand the normal embryology, morphologic
variations, and the characteristics, types, and timings of defects that can occur. It is important that pediatricians and other caregivers of infants and young children be able to recognize the common cranial abnormalities so that the families may be properly counseled and referred to an appropriate multidisciplinary craniofacial center. Because the most common causes of distortion or asymmetry are craniosynostosis or deformation, these abnormalities are the primary subjects of this article.

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