Chapter 21: Craniofacial Surgery for Congenital and Acquired Deformities

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Modern craniofacial surgery has evolved from the work of many pioneers in the field, most notably Dr Paul Tessier and Professor Hugo Obwegeser. The term *craniofacial* is descriptive of this branch of surgery, which deals with anomalies of the bones and soft tissues of the cranium and the face. In the purest sense, craniofacial surgery refers to those procedures that combine an intracranial and extracranial approach to correct facial deformities. The techniques that have been developed in craniofacial surgery are directly applicable to the expanding field of skull base surgery, particularly in the treatment of tumors of the anterior cranial fossa and infratemporal fossa. In the broadest sense, a wide range of treatment options is available depending on the age of the patient and the deformity. Patients with Treacher Collins syndrome may undergo bone grafting to malar and orbital areas to improve bony deficiencies caused by facial anomalies. At a later age, they may undergo orthognathic correction of maxillomandibular disharmonies. Similarly, patients with otomandibular dysostosis may require ramus reconstruction with costochondral grafts between the ages of 6 and 7, additional orthognathic corrections when in their teens, and soft-tissue augmentation later on. Patients with oral facial clefting require closure of the cleft and reconstruction of the underlying bony deficiencies at a very early age, followed by subsequent corrections of orbital hypertelorism between the ages of 2 and 5, and finally, orthognathic correction of maxillary and mandibular deformities in their teens. Craniofacial surgery is useful for correcting not only congenital anomalies, but also acquired deformities caused by trauma or tumors of the cranial and facial bones.

Evaluation of patients with craniofacial deformities requires a team approach. The team usually consists of the craniofacial surgeon, neurosurgeon, oral and maxillofacial surgeon, speech pathologist, audiologist, pedodontist, orthodontist, prosthodontist, social worker, geneticist, pediatrician, and ophthalmologist. Other consultants may be added to the team as needed. Patients who are referred to a craniofacial clinic are seen by all members of the team at the same visit. At a subsequent team conference each consultant makes recommendations, which are integrated into a comprehensive report by the team leader and presented to the patient's family and treating physician as a long-term treatment plan.

The entire field of craniofacial surgery is beyond the scope of this chapter. The purpose of this chapter is to discuss the pathogenesis and evaluation of patients with congenital anomalies of the craniofacial skeleton and to provide an overview of the treatment of these congenital anomalies. This requires a combined intracranial and extracranial approach to the upper facial skeleton.

Pathogenesis

Patients requiring craniofacial surgery usually present with multiple anomalies associated with syndromes, but these anomalies may occur as isolated events. Craniofacial anomalies can be grouped into three different categories. The first is a malformation, which is defined as a morphologic defect of one or more organs resulting from intrinsically abnormal development. The second is that of deformation, which is an abnormal form or position of a part of the body caused by nondisruptive mechanical forces. The third category

is that of disruption, which is a defect of an organ or region of the body resulting from a breakdown of or interference with originally normal development. Classifying the craniofacial anomaly into one of these categories is helpful for planning long-term treatment and genetic counseling for the family. The various anomalies can be considered in regard to their different modes of presentation.

Craniosynostosis and craniofacial synostosis

Craniosynostosis is a premature fusion of a cranial suture. Craniofacial synostosis is an extension of this condition in which not only do the sutures of the cranium fuse prematurely, but also those of the skull base and face. Craniosynostosis may be either simple or multiple. Simple craniosynostosis refers to premature fusion of one suture of the calvarium. Multiple craniosynostosis involves more than one suture, such as both coronal sutures or coronal sutures and a lambdoid suture. Fusion of a cranial suture present at birth is referred to as primary craniosynostosis, which is a defect that has occurred in utero. In contrast, secondary craniosynostosis involves postnasal premature suture fusion, which can be caused by a ventriculoperitoneal shunt or sickle cell anemia.

Craniosynostosis may be either isolated or syndromic. In the isolated form, the abnormality is usually more limited in extent and severity than when it occurs as part of a syndrome. In isolated unilateral coronal craniosynostosis, for example, there is restriction of skull growth on the affected side and compensatory overgrowth along the ipsilateral lambdoid suture. The overall result is a retrusion of the forehead on the affected side and a bulging of the occiput on the same side. The deformities found in multiple craniosynostosis are much more severe. Fusion of bilateral coronal and lambdoid sutures results in the deformity called *turricephaly* (tower skull). Not only is the skull shaped like a tower, but also there is a significant risk of increased intracranial pressure and marked restriction on brain growth.

Many theories have been proposed about what causes primary craniosynostosis. Moss believed that the abnormality centered in the skull base. He proposed that skull base deformity, and in particular that of the sphenoid, resulted in a premature fusion of calvarial sutures due to the transmission of abnormal forces from the skull base through the dura to the sutures themselves. Virchow believed that the calvarial sutures themselves fused prematurely and that the skull base deformities were secondary to the calvarial deformity. Park and Powers suggested that the abnormality was centered in the mesenchymal cells in the embryo, which affected both the calvarial and basilar sutures. Another hypothesis is that of fetal head constraint. Graham and colleagues noted that in some cases of craniosynostosis, the mothers reported premature descent of the fetus into the pelvis.

There are a number of conditions that can cause secondary craniosynostosis, including low CSF pressure, caused by a ventricular peritoneal shunt, thalassemia major, sickle cell anemia, and hyperthyroidism. It is important to classify the craniosynostosis because isolated nonsyndromal fusion has the best surgical outcome. Patients who have one suture fused and do not have a syndrome usually require only one surgical procedure to correct the craniofacial deformity. However, patients with multiple sutural fusions may require multiple surgical procedures. This distinction is important because the patient's family needs to be prepared for the possibility of multiple major procedures.

In contrast, craniofacial synostosis involves abnormalities of the sutures of the cranium, the face, and the cartilaginous growth centers (or synchondrosis) of the skull base. Patients with Apert syndrome manifest atypical facies, consisting of forehead retrusion with a wide skull, midface hypoplasia and retrusion, varying degrees of orbital hypertelorism, and exorbitism. Because both coronal sutures are fused, the forehead is retropositioned and the patients have a short, wide skull (brachycephaly). The fusion of facial sutures, particularly of the skull base synchondroses, restricts downward and forward growth of the skull base and of the maxillae. Because of this restriction and the resultant lack of spatial movement of the midface, growth occurs primarily with deposition and resorption of bone in the midface. This produces the classical retromaxillism and micromaxillism found in Apert syndrome.

Orbital hypertelorism

Orbital hypertelorism is defined as an increase in the actual interorbital distance. This can be measured by a PA cephalogram or axial CT scan as the distance between the two anterior lacrimal crests (dacryon). The normal range of measurements for this distance is from 18 mm in the newborn to 25 mm in the adult. Orbital hypertelorism is classified as mild (30-34 mm), moderate (35-39 mm), or severe (greater than 39 mm). The term *orbital hypertelorism* describes a physical finding and does not denote any specific syndrome; rather, it is found in many syndromes. Orbital hypertelorism may also be found in craniofacial anomalies that are not a part of a syndrome. These anomalies usually involve midline defects of the craniofacial skeleton such as the various nasofrontal, nasoethmoid, and nasoorbital encephaloceles in which there is a defect in the anterior cranial base with the leptomeninges and brain extending into the nasofrontal and ethmoid complex. Hypertelorism in these cases is a result of a failure of midline fusion and is not associated with a specific syndrome. Hypertelorism associated with a syndrome is usually accompanied by marked widening of the ethmoid sinus complex.

Orbital hypertelorism may also be the result of a disruption, most frequently caused by amnionic bands. In these cases, although the midline structures have the potential to fuse normally and there is no syndrome present, the hypertelorism is produced by a distracting force such as amnionic banding. The craniofacial clefting seen in these patients does not correspond to a failure along an embryologic line of fusion, but rather is caused by a distracting extrinsic force.

Facial clefting

Cleft lip and palate are the most commonly seen forms of facial clefting. However, other clefts may arise in the craniofacial skeleton. These clefts may result from either a failure of embryonic tissue migration or of its failure to fuse. The ensuing facial cleft is the result of a malformation process. Facial clefting may also be the result of a deformation process, for example, the Robin sequence in which patients manifest micrognathia, glossoptosis, and palatal clefting. In this sequence, the clefting is theorized to be a result of the tongue lying between the palatal shelves, preventing fusion. Superior displacement of the tongue is a result of the retrodisplacement of the mandible caused by extrinsic compression. The Robin sequence is seen in association with many syndromes, including mandibulofacial dysostosis, Stickler syndrome, del (6q) syndrome, fetal alcohol syndrome, amniotic band sequence, and CHARGE association. Tessier has created a classification system for clefts involving the

facial and the cranial skeleton. This classification relates to malformations that occur along embryologic lines of fusion. Some facial clefts cannot be placed within this classification scheme nor can they be explained embryologically. These abnormalities are the result of disruptions.

A classic example of facial clefting is seen in the Treacher Collins syndrome. This syndrome is autosomal-dominant, with the patient exhibiting malar hypoplasia, antimongoloid slant of the eyes, decreased posterior height as a result of a short mandibular ramus, severe antegonial notching, and an open dental bite. In the most pronounced form of Treacher Collins syndrome, the facial skeleton exhibits clefts 6, 7, and 8 from the Tessier system. Clefts in these areas may extend through the inferior and lateral orbit, as well as the arch of the zygoma, producing the severe malar deficiency seen in this syndrome.

Disorders of branchial arches

Oculo-auriculo-vertebral spectrum

The oculo-auriculo-vertebral spectrum encompasses not only a wide variety of craniofacial anomalies but also a number of specific conditions that are frequently confused. These include hemifacial microsomia, otomandibular dysostosis, Goldenhar's syndrome, and first arch syndrome. Because of the wide variety of the degree and severity of these complexes, they are all referred to by the overall term *oculo-auriculo-vertebral spectrum.* In the mildest of forms, the patient may have merely a unilateral microtia; in very severe forms, there may be a total absence of the mandibular ramus and the ear on the affected side, ipsilateral anophthalmia, and epibulbar dermoids. The frequency of occurrence of the spectrum ranges from 1:3.500 to 1:26.550 live births. The male to female ratio is 3:2. Two hypotheses have been proposed to explain this spectrum of deformities. Poswillo induced hematoma formation in the region of the ear of animal embryos. This resulted in destruction of the tissues of the ear and mandible and produced a condition that was very similar to that seen in the human expression of the spectrum. Other theories suggest that there may be changes in the neural crest cells that result in a lack of development in the first arch area. Patients with this spectrum have at least microtia or another ear deformity. Over 50% of these patients have facial asymmetry, and when bilateral involvement is present, the deformity is always worse on one side than the other.

Epibulbar dermoids and colobomas are common findings as well. In more severe cases, patients may manifest central nervous system disorders, including Arnold-Chiari malformation, hypocephalus, or agenesis of the corpus callosum. Additionally, pulmonary, renal, and cardiac anomalies have been reported.

The Goldenhar complex includes the fusion of cervical vertebrae in approximately 20% to 30% of the cases. Cleft lip and palate have also been reported in these patients.

Mandibulofacial dysostosis

Mandibulofacial dysostosis, also known as *Treacher Collins* or *Franceschetti-Zwahlen-Klein* syndrome, is an autosomal dominant disorder with a variability of expression. The upper facial findings have been described in the section on orbital clefts. First branchial arch

anomalies are also associated with the syndrome. In addition to hypoplasia of the zygomas and other orbital and upper facial findings, there is a decrease in the height of the mandibular ramus and the gonial angle is more obtuse than normal. Significant antegonial notching may also be present and is usually correlated with a significant degree of mandibular deficiency. Cleft palate may be found in approximately 35% of these patients.

The combination of upper facial clefting with malar and orbital abnormalities and mandibular deficiency makes mandibular facial dysostosis a relatively easy syndrome to recognize.

Evaluation

Patients with congenital craniofacial deformities should be evaluated by a multidisciplinary team, each member of which should be highly experienced. Because of the multisystem problems found in these patients, experts from a number of disciplines are necessary to evaluate and formulate an overall treatment plan. In the initial evaluation of patients with craniofacial deformities, it is important to systematically list all of the known anomalies. Additionally, a detailed family history is important to determine if any relatives have manifested any similar deformities. An attempt should be made to classify the collective group of physical findings into a known syndrome. Should a syndrome be identified, the patient can be evaluated for other associated anomalies that may be unrecognized at the time of the initial presentation. If the patient's condition cannot be classified into a known syndrome, the anomaly may be a sporadic one that is not genetically related. This is important information for patients and their families.

Radiographic evaluation of these patients should include skull films to evaluate sutures of the cranium for any evidence of craniosynostosis. X-ray studies may be needed to determine bone age and to evaluate for syndactyly or abnormalities of bone metabolism. A three-dimensional CT scan (Fig. 21-1) may be obtained to evaluate the overall form of the cranium and the face and to examine the skull base in relation to the basal and cranial sutures. If there is any fear of underlying central nervous system anomalies, magnetic resonance imaging (Fig. 21-2) is helpful for ruling out such deformities as corpus callosum agenesis, holoprosencephaly, or Arnold-Chiari malformation. In older children who are cooperative, lateral and PA cephalometric radiographs are taken routinely to evaluate the position of the facial bones in relation to each other and to the cranial base. Annual cephalometric examinations are important to evaluate continued facial growth and development, and to aid in the planning of orthognathic surgery and orthodontics. Dental casts are an important way of evaluating any existing malocclusion in older patients. The casts can be analyzed, and appropriate orthodontic treatment can be initiated. When patients require orthognathic surgery, such as LeFort III + 1 osteotomy for a patient with Crouzon syndrome, the combination of cephalometric planning analysis with cast surgery allows the team to predict the final outcome of surgery.

Another way of evaluating the craniofacial skeleton and of following facial growth and development is via anthropometry. This technique uses specific measurements of the facial skeleton, done externally with calipers. For example, bitemporal width measures the distance in centimeters between the two temporal fossa, and bizygomatic width measures the distance between the lateral aspect of the two malar prominences. Two measurements can be compared

with each other in a ratio, and these ratios can provide information about the relationship of one structure to another. For example, if all measurements of the craniofacial skeleton except the bitemporal width are within normal limits, that abnormal finding can be compared with the other normal measurements. If the ratio of bitemporal to bizygomatic width is below the normal value, the craniofacial skeleton is too narrow in the bitemporal area, as occurs in trigonocephaly. This information can be used clinically to evaluate patients for surgery and to follow their facial growth and development. It can also be used for clinical research because it helps to establish normative values for the various anthropometric measurements.

Treatment

There is an extensive range of treatment modalities available for use in patients with craniofacial deformities. These include such methods as closure of soft tissue clefts and subsequent bone grafting; augmentation of deficient areas using bone grafts or soft tissue flaps; various orthognathic and craniofacial osteotomies; and mandibular reconstructions. The entire spectrum of treatment modalities is beyond the scope of this chapter, so this discussion will be limited to those that involve the combined intracranial and extracranial approaches that are useful in dealing with the upper craniofacial skeleton.

Surgical approaches

The coronal approach offers the best exposure for surgery of the craniofacial skeleton. This involves a bicoronal incision with extensive dissection anteriorly above the pericranium. At a point approximately 2 cm above the supraorbital ridges, the pericranium is incised and subpericranial dissection is then carried down into the orbits. This preserves the supraorbital and supratrochlear nerves. The dissection medially can then be carried down over the nasal bones and into the medial orbits, preserving the lacrimal sac. If necessary, the medial canthal ligaments can be detached, repositioned, and reattached toward the end of the procedure. Laterally, the dissection is carried down over the temporalis muscle through the superficial layer of the deep temporal fascia. The dissection proceeds inferiorly in this layer through the superficial temporal fat pad to the arch of the zygoma. Dissecting in this fashion allows the zygomatic arch to be approached from its superior aspect and deep to the frontal branch of the facial nerve. At this point, the periosteum of the zygomatic arch is incised superiorly, and a subperiosteal dissection of the arch is accomplished with the frontal branch of CN VII being retracted laterally and protected by the superficial layer of the deep temporal fascia and the periosteum of the zygoma. The dissection along the zygoma is then extended superiorly to connect with the dissection of the frontal bone and proceeds circumferentially around the orbits, preserving the infraorbital nerves and detaching the lateral canthal ligaments. This approach allows total exposure of the cranium, the orbits, the zygomas, and the nasoethmoid complexes for osteotomies to be performed. In some cases the orbital floor may not be readily visualized through the coronal approach, so a transconjunctival-lateral canthotomy or subciliary incision may be necessary for further exposure. This may be particularly important for the correction of orbital hypertelorism when osteotomies are made through the floor of the orbit and through the face of the inferior orbital rim at the level of the infraorbital foramen.

Osteotomies

After complete exposure of the craniofacial skeleton, the predetermined osteotomies are performed as required for correction of the deformity. In patients with craniofacial synostosis, the forehead and upper orbits are in a retruded position. Surgery in these patients is performed initially at 3 to 6 months of age to allow for suture release and normal brain growth. The most common osteotomy performed for this is the fronto-orbital osteotomy (Fig. 21-3). This consists of bifrontal craniotomy and upper orbital osteotomies involving the anterior cranial fossa, upper half of the orbits, the root of the nose, and the temporal fossa to include the greater wing of the sphenoid. This then permits the forehead and the upper orbital complex to be mobilized and brought forward into a more normal position. After the bone is placed into its new position, fixation is accomplished with either wire or miniplate osteosynthesis.

Similarly, orbital hypertelorism is corrected through a combined intracranial and extracranial approach (Fig. 21-4). After a bifrontal craniotomy is done, periorbital osteotomies are made involving the four quadrants of the orbits. The bone cuts traverse the anterior cranial fossa, lateral orbital wall, and through the orbital floor into the inferior orbital fissure and medially through the medial orbital wall posterior to the posterior lacrimal crest. They then extend laterally into the inferior orbital fissure. Sagittal osteotomies are made of the lateral orbital rim. Additional osteotomies are made through the frontal bone, the nasal process of the maxilla medially, and through the infraorbital rim at the level of the infraorbital foramen. If the cause of the deformity is an encephalocele, this needs to be resected before orbital mobilization is performed. If, however, there is a widened ethmoid block, then following the orbital osteotomies, paramedian resections are done of the nasal bone and nasal process of maxilla. These are followed by a complete ethmoidectomy to remove the widened intraorbital ethmoid block. By preserving a midline strut of nasal septum and cribriform plate, olfactory function is not compromised. After the removal of aberrant intraorbital tissue, the orbits are translocated medially and secured in place with wires, bone grafts, and miniplates.

Monoblock facial advancement may be performed in children 4 to 6 years of age with severe midfacial and forehead retrusion, such as that seen in Crouzon and Apert syndrome (Fig. 21-5). In this procedure, a bifrontal craniotomy is performed, followed by upper orbital osteotomies involving the upper half of the orbits, root of the nose, and into the temporal fossa. This is followed by a total midface mobilization achieved by disengaging the midface from the orbits through the posterior body of the maxilla into the pterygopalatine fossa and pterygomaxillary suture. The nasal septum is also sectioned, and the midface is then fractured with the disimpacting forceps and mobilized anteriorly. After midface fixation with bone grafts and miniplates, the upper orbital complex is then replaced and secured with plates and bone grafts. The forehead is remodeled and repositioned into its new forward position.

Future

Much research is being done on the molecular biology of the various syndromes of the head and the neck. For example, disorders of mucopolysaccharide metabolism are well known to be the cause of the facial features seen in Hurler syndrome. Inborn errors of bone metabolism may cause the facial deformities seen in craniotabular dysostosis. If the specific gene locus for the metabolic error could be identified, that gene could be altered, enabling

metabolism to proceed in a normal fashion, and hopefully, lessening the ultimate outcome of the deformities caused by the metabolic disorder. Additionally, advances in fetal surgery may soon permit its practical use in humans. If craniofacial anomalies are detected in utero, it may be possible some day to correct some anomalies, such as craniosynostosis or facial clefting before birth.