Pediatric Facial Plastic and Reconstructive Surgery

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Chapter 2: Congenital Anomalies of the Nose and Nasopharynx

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Congenital anomalies of the nose are related to the embryological development of the face during the first 12 weeks of fetal development. To better understand the clinical findings, pathology, and differential diagnosis, and to make a plan for the evaluation and treatment of these anomalies, we must review the embryological development of the nose and face during those critical first 12 weeks of development.

Embryology

The development of the face begins at about 3 to 4 weeks of age. At this stage the embryo is composed of a brain covered with a membrane; the anterior neuropore is still present and the optic vesicles are placed on the lateral aspect. At this point the neural fold develops as a longitudinal infolding along the dorsal aspect of the embryo. The margins of this infolding form the neural crest, which continues to expand into the neural groove and undergoes closure, creating the neural tube. During the same time the neural-crest cells, which are "pluripotential cells," migrate laterally and are important in the development of the face as they are the tissue of origin of the various germ layers that create the facial structures. As these cells migrate laterally they pass posteriorly to the developing eye and anteriorly over the frontonasal process. At the same time, the fetal head is flexing as the neural-crest cells reach their predetermined location to create the first and second branchial arch mesenchyme. Now the development of the matrix of neural tubes and cellular differentiation occurs, which leads to facial process development by the end of the 4th week.

As thyis step is completed, the head consists of a large bulging mass at the cephalic end, which contains the forebrain. Two small thickenings just above the stomadeum depression in the epithelium form the nasal placodes. The stomadeum is created by the separation of the first branchial arch into the maxillary and mandibular portions. In a short period of time this depression connects with the primitive foregut, forming the oral plate, which ruptures to form the oral cavity. At the same time the nasal placodes are developing as horseshoe-shaped bulges, forming the medial and lateral nasal processes. As the medial nasal processes approach each other int he midline, the embryo's face begins to have recognizable human features. During this time the changes that occur with amazing rapidity and precision are critical in the formation of many of the congenital anomalies of the head and neck.

The processes that are developing are really ridges of ectoderm overlying proliferating, differentiating, and migratory neural-crest cells. Once the overlying facial features are formed, there is not a dramatic change in appearance, but the smoothing out of the tissues by the tremendous differential growth of the mesenchymal tissue results in enlargement of the fetus, as well as in changes in facial configuration.

During the 6th week there is rapid growth of the medial nasal processes and the maxillary processes toward each other, with fusion to form the upper lips, philtrum, philtral ridges, and columella. The lateral nasal processes become less prominent, but a deep groove is formed between them and the maxillary process that extends from the stomadeum to the medial canthus of the eye. This depression, the nasal maxillary groove, will later be closed by overlying epithelium and will form the nasolacrimal duct.

During the next 2 weeks, the pits of the nasal process burrow deeper and above the stomadeum to form a thin sheet called the nasobuccal membrane, which will rupture to create a nasal cavity with a primitive choana. During this time, the two sides are separated by the forming nasal septum with its anterior septal cartilage and the columella, as a result of the mesodermal flow from neural crest cells into the frontonasal process. Other neural crest cells enter the craniobasal region to form a cartilage model that will ossify and become the posterior septum, ethmoid complex, cribriform plate, and sphenoid bone. The ethmoid complex is derived from lateral and medial mesenchymal masses that surround the oflactory sacs. The olfactory nerves penetrate the area between the lateral and medial masses, and later are surrounded by cartilage that will ossify to become the cribriform plate. The remainder of the posterior septum comes from mesenchymal tissue forming the vomer. The septum is then fused in the midline with the palatal processes starting anteriorly and continuing posteriorly to form the hard palate and separating the oral and nasal cavity. The nasobuccal membrane will have ruptured, allowing communication with the oropharynx via the permanent posterior choana.

If during this 12-week, rapidly moving, complex event there are genetic or external influences that effect the normal movement of cells and cell masses, there will be a permanent effect on the fetus. Later steps in the developmental sequence will be further altered resulting in the congenital anomalies we will be discussing.

Nasal Dermoid (Nasal Dermoid Sinus Cyst)

The appearance of a subcutaneous epithelial cyst in the area of the nose may represent a simple epidermal inclusion cyst or a nasal dermoid sinus cyst (NDSC). The former is simply a cyst with a fibrous wall, lined with stratified squamous epithelium, and confined to the subdermal or subcutaneous layers of the skin. An NDSC may have a similar appearance, but it may or may not have an opening to the skin or have an intracranial connection to the dura. On microscopic exam it will have a fibrous cyst wall lined with squamous cell epithelium as well as other skin appendages including sebaceous glands, hair follicles, and sweat glands.

Embryology

Nasal dermoid sinus cysts represent an embryological defect similar to the defect that forms a nasal glioma or anterior encephalocele. The externalnose forms initially as a cartilaginous foundation that is continuous posteriorly with the ethmoid labyrinth. External to this cartilaginous foundation the nasal and frontal bones form via intramembranous ossification during the 2nd to 3rd month of life. Between the frontal bones and nasal bones there is a space filled with a firm mebrane called the fonticulus frontalis. Between the nasal bones and the more posterior cartilage is the prenasal space. At the cranial end of the prenasal space there is a herniation of dura through the foramen caecum, which passes through the

prenasal space and is continuous with the periosteal lining of the nasal bones. As development proceeds, the foramen caecum and fonticulus frontalis are obliterated by bony growth.

Various theories concerning the embryopathogenesis of NDSCs have been proposed. Cysts basically fall into two categories: those that result from a cranial developmental defect and those that result from a cutaneous developmental defect. Pratt feels that there is failure of the skin to separate from the dura mater, which results in ectoderm being carried into the prenasal space for varying distances, including through the foramen caecum and intracranially to form a dermoid cyst or a persistent nasal dermal sinus. A tract may have cystic dilatation anywhere along its course or may undergo fibrous obliteration at any point along the tract. If there is failure of or incomplete retraction and obliteration of the neuroectodermal elements and the foramen caecum does not close, glial tissue may extend along the pathway, producing a glioma or encephalocele.

Diagnosis

Nasal dermoid sinus cysts are usually noter at birth or shortly after. They may range from a small pit in the skin with a few hair extruding somewhere between the glabella and the base of the columella, to midline cysts with or without a skin connection. If there is a skin pit it will intermittently drain small amounts of cheesy debris or cloudy exudate. Occasionally they do not drain and the diagnosis is not made until adulthood. If there is a cyst without a connection to the skin allowing drainage, it will gradually increase in size, remaining unattached to the skin, but thinning the skin and making it difficult to dissect a plane between the two. If there is a sinus tract, it will usually extend cephalic to the skin pit in a subcutaneous plane to the level of the nasal bones. At this point the sinus tract or cyst will create a depression splaying out the upper lateral cartilages and quadrilateral cartilage of the septum, causing the nose to be widened. The tract will usually pass beneath the nasal bones and into the prenasal space, although rarely the tract may stay superficial to the nasal bones and go between the nasal bones and frontal bone to enter the fonticulus nasofrontalis and extend through the foramen caecum. In either path the sinus tract may widen to form another cyst under the nasal bones and then extend superiorly into the anterior cranial fossa via the foramen caecum, the crista galli, or the cribriform plate, and sometimes as far posterior as the basisphenoid.

Intracranial extension is variable and in various reports has ranged from 0% to 85%. Intracranial extension is more frequent if a skin fistula tract is present, but may also be present with cysts and no skin pit.

Untreated NDSCs may become infected and require antibiotic therapy. Sometimes they may be mistaken for an infected epidermal inclusion cyst and be incised and drained. If there is intracranial extension, Paller et al reported a 22% incidence of infection, which included superficial infections, recurrent meningitis, osteomyelitis of the frontal and nasal bones, and frontal lobe abscesses. These complications may press for early surgical therapy.

The majority of NDSCs occur spontaneously, although there have been rare reports of a familial incidence. The reported incidence for midline nasal masses is 1/20.000 to 1/40.000 live births, which include NDSCs, nasal gliomas, and encephaloceles. There is a slight male preponderance in most series.

The differential diagnosis includes epidermal inclusion cysts, which are rare before puberty and are usually attached to the skin. Hemangiomas have a bluish color, are compressible, and are not attached to the skin or underlying tissues. The most critical lesions to differentiate are tumors of neurogenic origin, which will be nasal gliomas or encephaloceles.

Preoperative radiological evaluation is for the purpose of distinguishing between NDSCs, nasal gliomas, and encephaloceles. Today both magnetic resonance imaging (MRI) and computed tomography (CT) are used for this purpose. MRI is the most useful for encephaloceles, identifying brain tissue extending through defects in the floor of the anterior cranial fossa. Using T1- and T-2 weighted images cerebrospinal fluid (CSF) may be distinguished from fat. Barkovich et al found that both the CT and MRI were useful in diagnosing intracranial extension of a nasal dermoid sinus tract, but both also had certain pitfalls. One pitfall was normal widening of the septal cartilage superiorly being interpreted as expansion from a sinus tract in the septum. Fatty change in the frontal and nasal bones may be mistaken for fatty tumors such as the intracranial extension of a nasal dermoid. Finally, the fatty change in the crista galli or perpendicular plate of the ethmoid may be mistaken for intracranial extension. By looking at both sagittal and coronal views one can distinguish marrow from intracranial extension. Both CT and MRI may mislead the surgeon, and this is the reason we prefer to approach an NDSC from the nasal side first to prevent an unnecessary craniotomy.

Treatment

The successful treatment of NDSCs is complete surgical excision of all epidermal elements. Since most lesions are diagnosed before 1 year of age, indication for and timing of surgery becomes important. Treatment is indicated for all lesions except for the rare circumstance where the patient has an asymptomatic nasal pit that does not drain, there is no evidence of intraseptal or intracranial cyst, and the patient does not want surgery. Some authors have advocated waiting until the child is 2 to 5 years of age. However, there is the risk of repeated infections of the cyst, which will make dissection more difficult, or life-threatening meningitis if there is an intracranial extension.

My approach has been to evaluate the patient with a CT scan when the diagnosis is first made. If the CT shows a defect in the crista galli or anterior cranial fossa, and MRI is obtained looking for evidence of an intracranial extension. Neurosurgical consultation is also obtained on all patients, in case at the time of surgery there is an unsuspected intracranial extension. If there is no history of infection and the child is less than 1 year of age, I will follow them with plans to excise the lesion at about 1 year of age. If there is a cyst present on the nasal dorsum or in the columella, I watch it for enlargement. If it starts to enlarge, thinning the skin over it, or there is an infection, I will go agead with surgery at any age.

Our approach is to start by excising the extracranial portion of the NDSC first. If the lesion can be completely excised from this approach, no further surgery is necessary. If the tract extends intracranially, the proximal end is tagged with a hemoclip and the neurosurgeons will remove the intracranial portion via a small bifrontal craniotomy. This may be done at the same sitting, or if the initial procedure has been prolonged it may be delayed for 1 to 4 weeks in very young children.

The key to successful removal is adequate exposure of the nasal dorsum and the ability to continue deep to the nasal bones. Many authors advocate a midline dorsal incision from the glabella to the inferior portion of the cyst or inferior enough to include an elliptical excision of the skin pit, if present. Although this approach gives excellent exposure, it leaves a visible scar directly on the nasal dorsum. I have preferred to use a horizontal incision at the glabella and then to extend lateral limbs inferiorly on each side of the nasal dorsum like the lower limb of an external ethmoidectomy incision. This makes an inverted-U flap based inferiorly, with each limb being in a relaxed skin tension line. The skin pit is excised with a small elliptical excision oriented either vertically or horizontally. This incision gives excellent exposure with an excellent cosmetic scar. If there is a cyst in the tip of the nose or columella, I use a standard external rhinoplasty incision across the midcolumella with lateral incisions along the caudal margin of the lower lateral cartilage. This gives excellent exposure with minimal external scars, but if a sinus tract extends deep and posterior to the nasal bones, dissection may be difficult at the distal extent of the tract. Some authors advocate this approach in all cases because of the excellent cosmesis. Again, a separate incision must be made if there is a skin pit.

The skin is injected with 1% Xylocaine and 1:100.000 epinephrine for hemostasis. First, the elliptical excision is made around the skin pit through the dermis, taking care not to cut into the sinus tract. The skin incision is then made, freeing the nasal skin from the sinus tract and/or cyst to its most caudal extent on the nose. Many times the dermis has been thinned by pressure from the cyst and extreme care must be taken to not "button-hole" the skin. I have found the microscope to be an invaluable aid in this delicate dissection and then following the tract deep into the nose. Once the skin is elevated, the sinus tract may be dissected from its bed on the nasal dorsum. Middle ear elevators like a Rosen or McCabe dissector are especially useful to free the tract. The upper lateral cartilage and septal cartilage will usually be flattened ande concave from the pressure of the sinus tract. As one continues cephalically, the tract will usually go between the nasal bones and the cartilage, although occasionally it will extend superiorly over the nasal bones and go between the nasal bones and the frontal bone. With magnification it is easy to follow and distinguish a sinus tract from other tissue. It may be necessary to remove a small amount of the caudal end of the nasal bones or do a medial osteotomy for exposure as the tract extends posteriorly toward the foramen caecum or the crista galli. At this point the tract may end in a blind pouch with just a fibrous tract extending superiorly toward the anterior cranial fossa. With the middle ear elevators, I remove as much of this fibrous tract as possible and then carefully examine the speciment to make sure I can see the blind end of the sinus tract. If there is any question, a frozen section of the fibrous tract is done to make sure there are no epithelial elements left in the most distal portion of the tract. Sometimes this tract may end as a dilated cyst in the center of the septum. Care must be taken to remove all of the sinus tract lining.

If the sinus tract extends superiorly through the floor of the anterior cranial fossa, the dissection is carried to the bony margin. The distal end of the sinus tract is closed with one or two hemoclips, and Gelfoam or Gelfilm is used to fill the defect for an inferior marker when the intracranial portion is resected. Consultation is then made with the neurosurgical team and a decision is made whether to proceed with the intracranial portion at the same setting or delay for 1 to 4 weeks. In our hands, either way has been equally successful and the decision is usually made on the basis of the length of the anesthesia and the patient's age.

The incisions are closed in two layers using either 6-0 nylon or 6-0 fast absorbing catgut in the skin. Normally a drain is not used, but a light tape and pressure dressing is used for 2 to 3 days over the skin, which has been elevated.

If a neurosurgical approach is necessary, a bifrontal craniotomy is used. An extradural dissection is used down to the defect, which is usually in the area of the crista galli. As the dissection is carried out and the bony defect found, the sinus tract may be identified and lifted out, finding the hemoclips that will identify the inferior margin. The sinus tract may end at the dura where it may be peeled off, but more frequently it will have a cyst extending into the falx and a small portion of dura must be excised for complete removal. The dura can usually be closed primarily.

We use perioperative antibiotics unless there is an intracranial extension with infection present. In that case antibiotics would be used for 7 to 10 days.

Postoperative complications are usually limited, but postoperative seromas or infection may occur. With an intracranial resection we have had one temporary CSF leak, which closed spontaneously after 2 weeks. There is concern over the cosmetic appearance of the nose, but so far the postoperative results have been satisfactory.

Nasal Gliomas and Encephaloceles

Gliomas and encephaloceles are rare lesions that are made up of glial tissue and arise from similar embryological defects. The incidence is difficult to determine from the literature since they are so uncommon and obviously all cases are not reported. It has been reported that encephaloceles occur in the USA and Europe in 1 in 35.000 live births, but are more common in southern Asia with an incidence of 1 in 6.000 live births. My personal observation is that nasal gliomas and encephaloceles are about equal in occurrence, but both are much less common than NDSCs. Our definition of encephaloceles for the purpose of this chapter includes all types of intracranial herniations that are connected to the CSF system.

Embryology

These lesions are thought to arise from an embryological defect: faulty closure of the foramen caecum during the 3rd and 4th week of fetal life as the anterior neuropore is closing. It is thought that there is a failure of the appropriate migration of the neural crest cells, which allows a bony defect to develop along the floor of the anterior cranial fossa.

One theory is similar to the theory for the development of NDSCs. The dura that is in contact with the epithelium fails to retract through the foramen caecum and so instead of having an epithelial tract, one no whas a dural tract with or without actual brain tissue present. In the case of the nasal glioma, there is retardation of the neural crest cell migration, which allows brain tissue to be trapped extracranially, and located intranasally (30%), extranasally (60%), or a combination of the two (10%). About 15% maintain a fibrous connection to the dura, which normally passes through the cribriform plate area.

Encephaloceles are divided into two types, sincipital and basal. In the sincipital encephaloceles there is a failure of the foramen caecum to close just anterior to the crista

galli, allowing the encephalocele to pass between the ethmoid and frontal bone. It will then present externally in one of three locations. The nasofrontal type will present between the frontal and nasal bone in the area of the glabella. As it projects forward, it will cause a downward displacement of the nasal bones and a lateral displacement of the orbital wall. In the nasoethmoidal type the encephalocele will present between the nasal bones and cartilages of the nose. This displaces the nasal bones and frontal process of the maxilla superiorly. In the naso-orbital type, the defect is in the medial orbital wall, between the lacrimal bone and the frontal process of the maxilla. In this type the frontal bone, nasal bone, and nasal cartilages are normal and the mass presents between the lower eyelid and nose.

Basal encephaloceles form a defect in the anterior cranial fossa from the anterior border of the cribriform plate to the supraorbital tissue or posterior clinoid fissure. They will herniate into the nose, nasopharynx, or orbit. They are divided into four types, depending on the location of the defect. Transethmoidal lesions herniate through the cribriform plate and the encephalocele will present intranasally. Sphenoethmoidal lesions have a bony defect in the posterior cribriform plate and will present intranasally or in the posterior ethmoid cells. Transsphenoidal defects are usually large and extend from the cribriform plate to the posterior clinoid with the mass presenting in the nasopharynx. Sphenoorbital encephaloceles extend through the superior orbital fissure and present in the posterior portion of the orbit.

Diagnosis

Nasal gliomas are really areas of heterotopic brain tissue without a fluid-filled space that connects with the ventricles or subarachnoid spaces of the brain, but may have a fibrous or glial stalk attachment to the dura. Microscopically, the tissue contains foci of glial cells and fibrous tissue. They are surrounded by a pseudocapsule of fibrous tissue. These lesions are firm, rubbery, and noncompressible, and rarely have mitosis in the cells. As a result they are not neoplastic lesions and expand slowly in proportion with the growth of the child. Most present in or around the glabella of the nose, but only rarely in the midline. The skin overlying the lesion may be erythematous, telangiectatic, or violaceous, but will be mobile over the underlying mass. Those that present intranasally may cause nasal obstruction, septal deviation, or deformity of the surrounding bony structures, or may be large enough to prolapse from the nose. The masses do not enlarge with crying or jugular vein compression (Furstenberg's sign).

There are rare reports of other head and neck locations for heterotopic brain tissue separate from the subarachnoid space, presenting in a separate compartment of the brain, the nasopharynx, soft palate, pterygomaxillary space, and/or maxillary sinus. These lesions are identical microscopically to nasal gliomas and may even include ependymal cells and immature choroid plexus. Since they lack a connection to the dura, one would assume that they result from a defect during the closure of the anterior neuropore, leaving neural tissue separated from the brain, or possibly from misdirection of the migrating pluripotential neuroectodermal cells. As with nasal gliomas, these lesions grow in proportion to the child.

Encephaloceles do not seem to have a genetic predilection, but are often associated with other congenital abnormalities (30% to 40%). Sincipital encephaloceles most often present in the midline at the glabella or to one side of the nose. The size is variable from small elevations to masses as large as the child's head. The overlying skin may be

hyperpigmented, bluish, thin, smooth, wrinkled, or, in rare cases, with exposed brain and CSF leak. The masses are firm, somewhat compressible, rarely have transmitted pulsations, and may transluminate.

Basal encephaloceles are less common than sincipital encephaloceles and usually present in the nose or nasopharynx. The sphenoorbital type will present with proptosis. The other three types, transethmoidal, sphenoethmoidal, and transsphenoidal, will present with unilateral or bilateral nasal obstruction, feeding problems, and airway obstruction, which may be mistaken for a choanal atresia. On examination, there is a polypoid mucosal covered mass pedicled high in the nasal vault or nasopharynx that may pulsate or expand with venous compression (positive Furstenberg's sign). There may be unilateral rhinorrhea. Many smaller encephaloceles may go undetected and the patient will present with recurrent meningitis or spontaneous CSF rhinorrhea.

The diagnosis is best confirmed and the defect outlined with CT and MRI scans. Usually both will be used as the CT scan best identifies the area and extent of bony defects. The MRI best demonstrates the soft tissue components of these lesions. Herniated brain tissue is well identified adn with T1 and T2 weighting, CSF can be identified. With these two modalities a differential diagnosis of NDSCs, nasal gliomas, heterotropic brain tissue, and encephaloceles may usually be made without the use of angiograms.

Treatment

As with NDSCs, the treatment of nasal gliomas and encephaloceles is surgical. The surgical approach for nasal gliomas is similar to that of an NDSC. If after evaluation with MRI and/or CT scan the lesion appears to have no intracranial extension, the lesion is approached by the appropriate external incision for adequate exposure. If the glioma is intranasal, a lateral rhinotomy approach is usually used. The use of the microscope and microscopic instruments is similar to the procedure described for the nasal dermoid sinus cyst. It is important that all of the gliomatous tissue be removed or it will continue to grow with the child. If obvious or occult CSF fluid is encountered, one must assume that the diagnosis was wrong and that one is dealing with an encephalocele. In this case an intracranial approach will be necessary unless a local septal, mucosal, or turbinate flap can be created to seal the leak if it is recognized during the surgical procedure.

Hengerer and Oas report that five nasal gliomas had intracranial procedures as their primary approach, with four being normal, and in one patient a cribriform plate and dural defect was found. They recommended the following indications for intracranial exploration as a primary procedure: (a) a history of meningitis, (b) a compressible intranasal mass, (c) a positive Furstenberg sign, (d) radiographic evidence of an osseous defect or a soft tissue mass in the cribriform plate, and (e) CSF rhinorrhea.

Encephaloceles will tend to enlarge with time, which will increase the cosmetic deformity and potentially the amount of brain tissue and vital structures that may be involved. There is also the risk of meningitis when an encephalocele is untreated. For these reasons, early surgical intervention should be carried out as soon as the patient's condition permits it. If hydrocephalus is present, it should be first corrected with a shunt procedure. Nasofrontal encephaloceles have short, small necks, and may be amenable to an extracranial approach.

The nasoethmoidal and nasoorbital types should be approached intracranially, and the encephalocele separated from the brain with a watertight dural closure. The bony defect may be repaired with a bone graft or alloplastic material like tantalum mesh. The external portion may be removed at a later date or at the same time. If there are skeletal abnormalities in children under 3 years, much of the deformity will correct itself with normal growth. Excessive skin should be resected and the incision closed in a cosmetic fashion. Older children may need either primary or secondary correction of the associated hypertelorism. Sargent et al have described a one-stage reconstruction for these patients.

Intranasal basal encephaloceles should not be approached transnasally as a primary procedure. There will be a known CSF leak and there is the risk of meningitis working through a contaminated field. It is also difficult to obtain a tight dural closure. The primary approach should be intracranial with division of the encephalocele and repair of the dural defect. Osseous repair is similar to the repair for sincipital defects. If there is a residual encephalocele in the nose it can be removed later via a lateral rhinotomy. In transsphenoidal encephaloceles, there may be vital structures such as the pituitary hypothalamus or third ventricle prolapsed into the defect. Exposure is difficult and one must try to reduce the encephalocele rather than transect the stalk. There is a high risk to the operative procedure, and surgical intervention must be carefully weighed in view of the risk-benefit ratio.

Other Anomalies of the Nose

With all of the embryological fusion planes present, it is surprising that there are not more congenital anomalies of the nose. The most common defect associated with defects in the fusion planes of the face are the cleft lip and palate described extensively elsewhere in this book.

Congenital anomalies of the nose may include complete agenesis of the nose and midline clefting of the nose to various degrees. Lateral nasofacial clefts result from inappropriate fusion of the nasal maxillary groove in their severest form. Less severe forms may occur as small defects in the alar margin to actual deficiency or absence of the ala.

Other conditions that have been reported include proboscis lateralis and bilateral proboscis. These are reduplication defects with either an accessory portion of the nose laterally or some degree of reduplication of part or all of the nose. These may be associated with other cranial anomalies and hypertelorism.