Although clinicians often worry about missing occult brain tumors in children with headaches, most pediatricians fortunately rarely, if ever, encounter such a child; however, all pediatricians routinely encounter children with three common pediatric neurosurgical disorders: (1) macrocrania, (2) the oddly shaped head, and (3) craniospinal developmental malformations (craniospinal dysraphism). These three entities prompt far more referrals to pediatric neurosurgeons and are emphasized in this article.

MACROCRANIA

Macrocrania is among the most common reasons for new patient referrals to a pediatric neurosurgical practice. These children either have initial head circumference measurements that exceed the 95th percentile or, more ominously, demonstrate a head circumference growth pattern that is accelerating, that is, significantly crossing percentiles. There are two major causes of macrocrania: (1) hydrocephalus and (2) benign extra-axial fluid collections of infancy (BEFI). Less common causes include diseases associated with macrocrania but without hydrocephalus (i.e., neurofibromatosis, achondroplasia, and Canavan's and Alexander's disease), tumors without associated hydrocephalus, intracranial cysts, pseudotumor cerebri, and posttraumatic subdural hematomas. There is also a familial form of macrocrania. This article focuses on hydrocephalus and BEFI, as those are by far the most frequently encountered causes of macrocrania.
Hydrocephalus

Humans produce cerebrospinal fluid (CSF) at a rate of 0.3 mL/min, regardless of age. The CSF flows from the lateral ventricles to the third ventricle via the foramen of Monro, from the third ventricle to the fourth ventricle through the aqueduct of Sylvius, and out of the fourth ventricle either through the laterally placed foramina of Luschka or through the midline foramen of Magendie. Once out of the ventricular system, the CSF flows into the spinal and cerebral subarachnoid spaces and ultimately becomes absorbed into the venous system. The chief site for CSF absorption into the venous system is along the superior sagittal sinus via the arachnoid granulations.

Under ordinary circumstances, CSF is absorbed at the same rate as it is produced. Hydrocephalus represents an imbalance between the rate of production and the rate of resorption. Overproduction of CSF (usually caused by choroid plexus tumors) is very rare. Much more frequently, CSF absorption is impaired. Impairment of CSF absorption can be caused by a mechanical obstruction within the ventricular system (i.e., blockage of flow from one ventricle to another) or obstruction distally at the arachnoid granulations, where CSF is absorbed into the bloodstream. The former is known as noncommunicating hydrocephalus, and the latter is referred to as communicating hydrocephalus.

Noncommunicating hydrocephalus can result from obstruction anywhere within the ventricular system but usually occurs at the foramen of Monro, the aqueduct of Sylvius, or the fourth ventricle and its outlet channels. Causes of noncommunicating hydrocephalus include tumors; cysts; inflammatory scarring; intraventricular hemorrhage; and, rarely, genetic disorders. Ventricular obstruction at the foramen of Monro can be caused by colloid cysts, subependymal giant cell astrocytomas (in tuberous sclerosis), choroid plexus papillomas and carcinomas, central neurocytomas, and infrequently, intraventricular meningiomas. Obstruction at the aqueduct of Sylvius by a mass lesion can be caused by gliomas of the midbrain tectum, pineal region masses, and posterior third ventricular tumors. The fourth ventricle can be obstructed by fourth ventricular tumors, including medulloblastomas, ependymomas, cerebellar astrocytomas, and brainstem gliomas. Intraventricular or arachnoid cysts can obstruct CSF flow and are typically located around the third ventricle (i.e., suprasellar) or fourth ventricle. Inflammatory responses that lead to scarring can be induced by intraventricular hemorrhage or infection. These stimuli initiate an ependymal reaction leading to obstruction of the CSF pathway. Finally, an X-linked disorder associated with aqueductal stenosis has been identified. This syndrome includes mental deficiency and spasticity and can be associated with other central nervous system anomalies, including agenesis of the corpus callosum, fusion of the thalami, and atrophy of the pons and corticospinal tracts.

Causes of communicating hydrocephalus include meningitis, trauma, and intraventricular hemorrhage. Each of these processes is thought to cause an inflammatory reaction that impedes the flow of CSF from the subarachnoid space to the venous system. In some cases of communicating hydrocephalus, no identifiable predisposing factor is present.

Clinical Presentation

The signs and symptoms of hydrocephalus depend greatly on the age of the child. The clinical course for children less than 2 years of age is relatively benign because infants can expand the size of their cranial vault, thereby minimizing the elevation in intracranial pressure. Head growth is abnormally fast,
and the head circumference crosses percentiles. The anterior fontanelle is full, the sutures are split, and the scalp veins may be prominent. Poor feeding or vomiting may occur. Developmental milestones may be delayed, and loss of developmental milestones may be present. Increased tone or hyperreflexia may be apparent, particularly in the legs. Late signs include lethargy, sixth nerve palsy, and limitation of upgaze with a chronic downward deviation of the eyes, or "sunsetting."

In older children, the cranium is less able to expand sufficiently to offset the mounting volume of CSF, and the signs and symptoms are, therefore, more typically those of elevated intracranial pressure. Headaches, especially those that awaken the child from sleep or are present immediately on awakening, are strongly suggestive of raised intracranial pressure. Other symptoms include nausea, vomiting, lethargy, difficulty with school performance, loss of developmental milestones, and behavioral change. On examination, papilledema, sixth nerve palsy, or sunsetting of the eyes are particularly telling signs.

Diagnostic Studies

Hydrocephalus can be confirmed radiographically by cranial sonography (if the anterior fontanelle is still open), computed tomographic (CT) scans, or magnetic resonance (MR) imaging. The latter two options, especially MR imaging, provide the best visualization of the brain, which helps to determine whether an underlying structural cause for the hydrocephalus exists. A special technique, cardiac gated cine-flow MR imaging, can dynamically measure the flow of CSF through the cerebral aqueduct during the cardiac cycle and is helpful in diagnosing aqueductal stenosis.

Treatment

The decision to treat children with signs and symptoms of raised intracranial pressure is straightforward. A much more difficult task is deciding whether to treat asymptomatic children with normal development but who have mild or moderate ventricular enlargement. The outcome for these patients, with or without treatment, is largely unknown. Some authors advocate using the thickness of the cortical mantle as a guide to whether the hydrocephalus should be treated. Data from Young and colleagues suggest that children with a cortical mantle thickness of 2.8 cm or more after treatment with a shunt have normal intelligence quotients (IQs), whereas those with a postshunt cortical mantle thickness of less than 2.0 cm are developmentally delayed. Based on their data, these authors suggested that children with untreated cortical mantles of at least 2.8 cm are unlikely to require shunting. Children with cortical mantles of less than 2.8 cm reached this thickness if shunted by 5 months of age, suggesting that a decision to treat a child based on the thickness of the cortical mantle should be made before 5 months of age; however, this recommendation is not universally accepted. Others have suggested that children less than 3 years of age with moderate ventriculomegaly be shunted because the outcome without treatment is not known with certainty. Unfortunately, currently no objective data are that clearly define which children are likely to benefit from treatment.

Medical treatment options for hydrocephalus are extremely limited. Agents that reduce CSF production, such as acetazolamide or furosemide, can be used but are associated with electrolyte and pH imbalances and rarely provide a long-term solution. Available surgical options are dictated by the type of hydrocepha-
lus (noncommunicating versus communicating) and the child's age. Ventricular shunting has been the mainstay of surgical treatment since the 1950s and is the only available option for children with communicating hydrocephalus and for infants with noncommunicating hydrocephalus. For older children with noncommunicating hydrocephalus, neuroendoscopy offers an alternative treatment option.

For patients with communicating hydrocephalus, the proximal end of the shunt system is inserted into a CSF reservoir, most commonly the lateral ventricle or, more rarely, the lumbar subarachnoid space. The proximal catheter is connected to a valve that serves to regulate flow. A wide variety of valves is available, and a full discussion about the mechanics of each type of valve is beyond the scope of this article. The valve is connected to a distal catheter that is passed subcutaneously to a distal site and allows CSF to be reabsorbed into the bloodstream. The peritoneum is currently the favored site for placement of the distal catheter because of the ease of insertion with minimal risk. For children older than 5 years, the pleural space is another alternative site; however, younger children have difficulty absorbing CSF from the pleura and may develop symptomatic pleural effusions. Other sites for the distal catheter include the venous system (atrium or superior vena cava) and the gallbladder.

A relatively new and exciting surgical technique that can be offered as an alternative to patients with obstructive hydrocephalus is endoscopic third ventriculostomy. This procedure is performed with the aid of a small endoscope that is passed into the ventricular system and, under direct vision, is used to create a small hole (or fenestration) between the third ventricle and subarachnoid spaces, bypassing the ventricular obstruction. Endoscopic third ventriculostomy is effective in 60% to 80% of patients. Patient selection is crucial; only patients with noncommunicating hydrocephalus are reasonable candidates for the procedure. In addition, most series report a higher success rate in children over 2 years of age, although this limitation is not recognized by all. Younger children may respond less readily to endoscopic third ventriculostomy because they have relatively underdeveloped arachnoid granulations with less efficient CSF absorption; third ventriculostomy in these patients may, therefore, simply convert a noncommunicating to a communicating hydrocephalus.

The success rate for endoscopic third ventriculostomy is approximately 80% in children older than 2 years with noncommunicating hydrocephalus but decreases to 60% to 70% for children younger than 2 years. The long-term failure rate for third ventriculostomy is very low. Third ventriculostomy therefore offers the chance of a cure for patients older than 2 years with noncommunicating hydrocephalus. For younger children, we perform a shunt initially and offer endoscopic treatment later, at the time of a shunt malfunction.

Complications

Shunts can treat, but cannot cure, hydrocephalus, and they are associated with several complications, including, most commonly, shunt malfunction. The incidence of shunt malfunction in several large series is 30% to 40% within 1 year and 50% within 5 or 6 years following insertion. The signs and symptoms of shunt malfunction are similar to those of untreated hydrocephalus and can mimic a flulike illness with headache, nausea, and vomiting. It is critical to listen carefully to the parents because they often can tell from experience when the shunt has malfunctioned. Although a child may rarely deteriorate rapidly (over minutes or hours) to lethargy, coma, and impending herniation, symptoms more commonly develop over several days, weeks, or even months.
Symptoms may be steadily progressive or may occur intermittently. Examination may disclose papilledema even in the absence of any clinical signs or symptoms. Pumping the shunt gives little useful information and may actually suck debris or choroid plexus into the shunt; the authors, therefore, do not recommend routinely pumping the shunt to make a diagnosis.

Ancillary diagnostic studies confirm the diagnosis. A CT scan usually (but not always) shows ventricular enlargement; changes in ventricular size may be subtle, and a comparison should always be made with prior baseline scans if available. Unfortunately, some patients have no discernible change in ventricular size, even with clear-cut shunt malfunction. Radionuclide shunt flow studies are often abnormal in cases with obvious shunt malfunction but unfortunately are of little benefit in questionable cases. A shunt tap can be performed by inserting a small gauge needle into the shunt reservoir and may show poor proximal flow from the ventricular catheter, an elevated opening pressure, or consistent improvement in symptoms after removal of CSF.

Unfortunately, none of these ancillary tests is infallible, and one must remain highly suspicious of shunt malfunction in the presence of a strong clinical history. When pediatric neurosurgeons were polled, the clinical history was most frequently identified as the most important determinant in diagnosing shunt failure, followed by change in ventricular size on CT scans and elevated pressure on a shunt tap.

The most common reason for malfunction is occlusion of the proximal catheter by debris consisting of choroid plexus, glial or ependymal tissue, or clotted blood. Occlusion of the distal catheter is the next most common cause. Valves are an infrequent cause of shunt malfunction. Other rare causes of shunt malfunction include fracture of the catheter system with or without disconnection, improper placement of either the proximal or distal end, and migration of the distal end from the appropriate location. In some cases, the exact source of shunt malfunction cannot be determined. Shunt malfunction is treated by either revising the failed portion of the shunt or replacing the entire system.

Shunt infection rates vary from 3% to 29%, with an average of 5% to 15% per shunt operation. Seventy percent occur within 2 months, and 80% within 6 months, of surgery. Age affects shunt infection rates; infants less than 6 months old face a 15% risk for infection, whereas the rate for children older than 6 months is only 5%. Half of the patients with shunt infection present with shunt malfunction. Other symptoms include fever, meningismus, abdominal pain, tenderness, and erythema along the shunt tract. The diagnosis is established by aspirating fluid from the shunt system for culture, cell count, glucose, and protein. Characteristically, the CSF exhibits a leukocytosis with a left shift, a low glucose, and a high protein. CSF obtained via lumbar puncture is not adequate to diagnose a shunt infection because lumbar CSF cultures are positive in only 50% of proven shunt infections (conversely, a shunt tap is not adequate to exclude a diagnosis of bacterial meningitis).

Shunt infections are most frequently caused by Staphylococcus epidermidis, followed by Staphylococcus aureus, gram-negative rods (especially in neonates), and Propionibacterium acnes. Shunt infections are treated with intravenous (IV) antibiotics. Although some advocate adding intraventricular antibiotics, the utility of this remains unclear. The infected shunt usually must be removed. A temporary external drain is usually inserted to provide temporary CSF drainage and also serves as an ongoing source of CSF cultures during antibiotic treatment. A new shunt is inserted only after the infection has been adequately eradicated, as determined by several negative CSF cultures. This regimen was universally
successful in treating shunt infections in a study by James. Alternatively, the infected shunt can be treated in situ, subsequently removed at the end of treatment, and replaced immediately with a new shunt; however, this method is less successful, eradicating the infection in 90% of cases. Antibiotic treatment without replacing the shunt is successful in only 30%; however, *Haemophilus influenzae* and pneumococci are unique in that they may often be treated successfully with antibiotics alone.

Other complications of shunts include visceral injury during insertion and erosion into the viscera by the distal catheter over time. Ventriculostomy shunts can cause superior vena cava obstruction, pulmonary embolism, cor pulmonale, endocarditis, and arrhythmias. Shunt overdrainage can cause symptoms from low pressure (headaches brought on by sitting or standing and relieved by recumbency) or subdural hematomas from ventricular collapse that is too rapid. A complication unique to lumboperitoneal shunts is the development of a Chiari I malformation. The preferential drainage of the spinal CSF disturbs the balance between the cranial and spinal CSF spaces and displaces the cerebellar tonsils downward through the foramen magnum and into the cervical spinal canal. For this reason, lumbar shunts are used less frequently than are ventricular shunts.

Potential complications of endoscopic third ventriculostomy include vascular injury to the basilar artery or its branches (extremely rare, but with devastating consequences). Other potential complications include transient or permanent hormonal loss (most commonly diabetes insipidus) incurred by injuring the hypothalamus or pituitary stalk, and short-term memory loss from damage to surrounding structures. Fortunately, complications are rare.

### Benign Extra-Axial Fluid Collections of Infancy

Benign extra-axial collections of infancy are the most common cause of macrocrania in infants seen in the office setting. Head circumference measurements usually are less than the 95th percentile at birth, but by 3 or 4 months of age begin to accelerate and cross percentiles. The accelerated growth continues until about 12 to 18 months of age, at which point the curve begins to parallel, but usually does not significantly deviate back toward the normal curve (unpublished observations, 1997) (Fig. 1). There is a preponderance of males in most series. BEFI usually resolves without treatment by 2 years of age. The cause is unknown. Young children have few or no arachnoid villae, and instead absorb CSF through small parasagittal dural channels. BEFI may result from a transient imbalance or delayed development of these parasagittal dural channels and may, therefore, be a mild and self-limiting form of communicating hydrocephalus.

### Clinical Presentation and Diagnostic Studies

Most children with BEFI are otherwise asymptomatic and present with accelerated head growth. The anterior fontanelle is usually flat or minimally full, and no sutureal diastasis is present. Developmental milestones are usually normal, although some (7%) have mild transient delays in major motor skills (unpublished observations, 1997). The diagnosis is confirmed with cranial sonography, CT, or MR imaging, with CT being the most logical choice because it shows the prominent extra-axial spaces and can identify other significant changes in the brain tissue better than can sonography. The characteristic radio-
graphic feature of BEFI is prominent bilaterally symmetric CSF containing spaces located between the brain and skull, most prominent over the frontal regions (Fig. 2). The ventricles are usually normal but may be mildly enlarged in as many as 45% of infants.\textsuperscript{52, 62, 69, 77, 90-92}

The extra-axial collections of BEFI must be distinguished from the subdural fluid collections or hematomas of child abuse. Whereas the extra-axial fluid in BEFI has the signal characteristics of CSF, seems to be in the subarachnoid space, and is associated with widened cerebral sulci,\textsuperscript{36, 52, 62, 77, 93} the subdural hematomas that follow child abuse are of mixed density or are slightly more hyperdense than CSF and compress the adjacent sulci or produce mass effect on the ventricles or cerebral hemispheres. MR imaging is extremely useful in questionable cases because it accentuates the differences in intensity between CSF and blood products. Thus, whenever any doubt about the radiographic features exists, MR imaging should be performed.\textsuperscript{4}

Treatment

Treatment is unnecessary in essentially all cases because CSF absorption is corrected when adequate CSF absorption capacity develops, usually by 2 years of age; however, infants with BEFI should be watched closely during the first 18 to 24 months with frequent head circumference measurements and developmental assessments. Ventricular shunting is reserved for exceptional patients whose head growth does not slow down by 18 to 24 months or for those children with more significant developmental delays potentially attributable to the BEFI.

CRANIOSYNOSTOSIS AND THE ABNORMALLY SHAPED HEAD

The cranial sutures normally allow growth of the calvarium during the first several years of life. Craniosynostosis represents an abnormal fusion of one or more of these cranial sutures\textsuperscript{3} and results in restricted bone growth perpendicular to the fused suture. This, together with compensated growth at other cranial sutures, results in a misshapen head.\textsuperscript{17} The incidence of craniosynostosis is approximately 4 per 10,000 live births.\textsuperscript{75} Sagittal synostosis is most common, followed by unilateral coronal, bilateral coronal, metopic, and lambdoid synostosis. Involvement of multiple sutures is less common and more often reflects a genetic syndrome. Single-suture synostosis is most often simply a cosmetic disorder having no demonstrable effect on brain growth or development; however, multisutural involvement, particularly when involving three or more sutures, may restrict calvarial growth to such a degree that intracranial hypertension and developmental delays may result.\textsuperscript{87}

In the final analysis, almost all misshapen heads can be simply divided into four broad categories with a corresponding involvement of a particular suture: (1) the short head (brachycephaly, usually caused by bilateral coronal synostosis); (2) the elongated head (dolichocephaly or scaphocephaly, usually caused by sagittal synostosis); (3) the triangular-shaped head, which comes to a point anteriorly (trigonocephaly, usually caused by metopic synostosis), and the asymmetric head, in which one side is different from the other (plagiocephaly). Although plagiocephaly generally involves both the frontal and occipital regions to some degree, most patients have either primarily frontal asymmetry (frontal plagiocephaly, usually caused by unilateral coronal synostosis) or primarily
Figure 1. Head circumference growth chart for a cohort of infants with BEFI; A, boys, B, girls. The curve crosses above the norm at about 3 to 4 months, and continues to accelerate away from the curve until about 12 months, at which time the curve parallels, but does not deviate back toward, the normal curve. (Data from Partington, Dias, Li, Winston (in press).)

Illustration continued on opposite page
Figure 1 (Continued).
occipital asymmetry (occipital plagiocephaly, most often caused by positional molding and very rarely caused by lambdoid synostosis). If one keeps this schema in mind, it is usually quite easy to categorize misshapen heads and pinpoint the problem. Each of these conditions is discussed in turn. In addition, microcephaly as it relates to craniosynostosis is discussed because it also prompts occasional referrals to a pediatric neurosurgeon for evaluation.

The Short Head—Brachycephaly

Brachycephaly is of two types: (1) frontal and (2) occipital, of which occipital brachycephaly is by far the more common. Most cases of occipital brachycephaly are caused by positional molding caused by supine positioning during the first months of life. This is discussed later, together with occipital plagiocephaly.

Clinical Presentation

Frontal brachycephaly is caused by bilateral coronal synostosis. On examination (Fig. 3), the forehead is flattened bilaterally (more so than the occiput), and the anterior fontanelle is more anteriorly positioned. Both coronal sutures are palpably (and, in some cases, visibly) ridged, and are more anteriorly positioned, whereas the lateral portion of the coronal suture normally lies a little more than a finger breadth anterior to the tragus of the ear. The lateral margin of the fused coronal suture lies further forward just behind the lateral rim of the orbit, producing a small anterior fossa. The superior orbital rims, particularly along their lateral aspects, are bilaterally flattened (retruded) and deviated slightly superiorly. Fusion of other sutures, particularly in the genetic syndromes, may produce additional deformities. Finally, if associated midface hypoplasia is pres-
ent, the inferior orbits and maxilla are severely flattened (Fig. 3), the orbits are shallow, and proptosis of the globes is present bilaterally. The nose may be short, stubby, and upturned slightly, producing a “parrot’s beak” (particularly in Apert’s syndrome).

Bilateral coronal synostosis often occurs in the setting of one or another genetic syndrome (Table 1). Some common associated anomalies that suggest a genetic syndrome include syndactyly (which, if severe suggests Apert’s syndrome and, if partial or less severe—especially limited to portions of the second and third digits—suggests Saethre-Chotzen, Pfeiffer’s, or Jackson-Weiss syndrome), broad thumbs or great toes (suggesting Pfeiffer’s syndrome or, if involving only the toes, less commonly Jackson-Weiss or Saethre-Chotzen syndrome), ptosis (suggesting Saethre-Chotzen syndrome), and hypertelorism with nasal clefting (suggesting craniofrontonasal dysplasia). Some of the more common genetic disorders and their associated anomalies are listed in Table 1; an exhaustive review of these syndromes is found in reference 16.

Diagnostic Studies

Although bilateral coronal synostosis is usually obvious clinically, several radiographic features confirm the diagnosis. Because as many as 50% of children with bilateral coronal suture synostosis have involvement of other sutures as well, all sutures should be carefully evaluated. The cardinal features of bilateral coronal synostosis on plain radiographs (Fig. 4) are marked brachycephaly;
<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Mode of Inheritance</th>
<th>Craniosynostosis</th>
<th>Clinical Characteristics</th>
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<tbody>
<tr>
<td>Crouzon</td>
<td>AD</td>
<td>Coronal</td>
<td>Facial asymmetry, low-set frontal hairline, ptosis, variable brachydactyly and cutaneous syndactyly especially fingers 2 and 3, normal thumbs, occasional broad great toes and feet minimally involving digits 2-4, mental deficiency (40%)</td>
</tr>
<tr>
<td>Apert</td>
<td>AD</td>
<td>Coronal</td>
<td>Midface hypoplasia, shallow orbits, proptosis, strabismus, radiohumeral synostosis, joint contractures, arachnodactyly, femoral bowing</td>
</tr>
<tr>
<td>Saethre-Chotzen</td>
<td>AD</td>
<td>Coronal</td>
<td>Coronal * Brachycephaly, proptosis, midface hypoplasia, dysplastic ears, clefting of nasal lip, various abnormalities of hands and feet</td>
</tr>
<tr>
<td>Pfeiffer</td>
<td>AD</td>
<td>Coronal</td>
<td>Midface hypoplasia, downslanting palpebral fissures, variable brachydactyly and cutaneous syndactyly of hands and feet</td>
</tr>
<tr>
<td>Jackson-Weiss</td>
<td>AD</td>
<td>Coronal</td>
<td>Broad first metatarsals and fused tarsal bones, occasionally broad great toes, normal thumbs. May have Pfeiffer-like, Saethre-Chotzen-like, or Crouzon-like variants</td>
</tr>
<tr>
<td>Carpenter</td>
<td>AR</td>
<td>Coronal</td>
<td>Antley-Bixler</td>
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<tr>
<td>Craniofrontonasal dysplasia</td>
<td>X-L</td>
<td>Coronal</td>
<td>Craniofrontonasal dysplasia</td>
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AD, autosomal dominant; AR, autosomal recessive; X-L, X-linked.

sclerosis or obliteration of the coronal sutures; anterior bowing of both coronal sutures, with a shallow anterior cranial fossa and shallow orbits on the lateral film; and bilateral harlequin eye deformities on the anteroposterior film. On CT scans, the coronal sutures are difficult or impossible to visualize, the sphenoid wings are anteriorly displaced, and the orbits and anterior cranial fossa are shallow. In addition, the temporal fossa is deep and wide because of compensatory growth along the squamosal suture.

**Treatment**

Bilateral coronal synostosis is ideally repaired at 5 or 6 months of age and involves a bilateral fronto-orbital reconstruction. Both the forehead and the
orbital roofs bilaterally are removed, recontoured, and advanced to a more anterior position.

The Long Head—Dolichocephały

Dolichocephaly (also commonly called scaphocephaly) describes a head that is both too long in the anteroposterior dimension and usually abnormally narrow biparietally. Although some cases occur in premature infants who are laid with their heads turned to the side in the neonatal intensive care unit (termed NICUcephaly), most cases are caused by isolated sagittal synostosis. Sagittal synostosis affects boys three times more frequently than girls. Associated intracranial abnormalities are lacking, clinical evidence of elevated intracranial pressure is extremely rare, and intelligence is normal. Isolated sagittal synostosis is familial in only 2% of cases, although sagittal synostosis can also be associated with fusion of other sutures as part of a genetic syndrome.

Clinical Presentation

The dolichocephalic head shape (Fig. 5A) in sagittal synostosis is almost always apparent at birth and progresses further within a few weeks. The head is both long and narrow. There may be frontal bossing, occipital prominence, or both. The mid-portion of the head may be sunken like a saddle, with prominent frontal and occipital regions, producing a peanut shape to the head. The sagittal suture is almost always prominently ridged and is also quite long. A simple test of sagittal suture length is to measure the distance between the anterior and posterior fontanelles; normally, this distance is roughly equal to the distance between the nasion (at the root of the nose) and the anterior fontanelle, but in infants with sagittal synostosis the distance between the two fontanelles is much greater.

Diagnostic Studies

Plain radiographs usually confirm the diagnosis. The head is elongated on the lateral projection, and frontal or occipital prominence may be present (Fig. 5B). The mid-portion of the sagittal suture may contain an indentation corresponding to the saddle deformity seen clinically. Most often, prominent perisutural sclerosis is present along either side of the sagittal suture on the anteroposterior view; heaped up sclerotic edges may produce sutural “beaking.” Sclerosis may be focal, involving only a portion of the suture, and may be better defined on tangential views. Facial bones are normal; any abnormality should suggest associated coronal synostosis.

Treatment

Treatment is usually undertaken at approximately 2 to 4 months to capitalize on the rapid brain growth during the first year. Sagittal synostosis is typically corrected by first removing a 4-cm to 6-cm-wide, midline strip of bone between the anterior and posterior fontanelles, as well as a small keyhole portion of the occipital bone behind the posterior fontanelle. Beyond this, several modifications have been advocated, which result in both immediate and long-term improvement compared with a narrow midline strip craniectomy. Whatever the technique, all procedures work best when performed early. Longer delays (beyond approximately 8 mo) result in poorer operative results and, particularly
Figure 5. Dolichocephaly owing to sagittal synostosis. A, View from above in an infant with sagittal synostosis shows an elongated head shape. B, Radiographic features of dolichocephaly. The elongated head is evident on the lateral radiograph (arrows show the normal coronal sutures). (From Fernbach SK, Naidich TP: Radiological evaluation of craniosynostosis. In Cohen MM: Craniosynostosis: Diagnosis, Evaluation and Management. Raven Press, New York, 1986, p 194, copyright of Michael Cohen, MD; with permission.)
when treatment is delayed beyond 12 to 18 months, require a more extensive
(and potentially more morbid) cranial reconstruction. It is therefore imperative
that referrals be made early; the diagnosis is usually obvious at birth and does
not improve with time.

The Triangular Shaped Head—Trigonocephaly

Clinical Presentation

Trigonocephaly caused by metopic synostosis accounts for approximately
10% of cases of synostosis. Two forms of metopic synostosis probably exist.
The first and more common form is characterized by an isolated palpable
(and occasionally visible) midline bony ridge in the forehead but without any
significant change in head shape or hypotelorism. In most cases, nothing further
need be done for these individuals; occasionally, the midline ridge needs to be
burred down, but reconstructions are not necessary. The second form is associ-
ated with secondary changes in skull shape producing trigonocephaly. The
forehead is triangularly shaped and resembles the keel of a boat. Bitemporal
narrowing compared with the midparietal width gives a teardrop shape to the
head when viewed from above. Hypotelorism and retrusion of the lateral orbits
are often present as well and further exaggerate the teardrop shape of the head.
A palpable and often visible ridge is present along part or all of the metopic
suture. Some cases may result from a midline embryonic field defect, which
results in metopic synostosis and disorders of midline brain development, such
as holoprosencephaly or septo-optic dysplasia and disordered midline facial
development producing a median cleft lip. Associated intracranial abnormali-
ties should be sought via CT or MR scans in all children with trigonocephaly
because they may predict future developmental delays.

Diagnostic Studies

Plain radiographs and CT scans (Fig. 6) in patients with trigonocephaly
demonstrate the prominent midline keel, obliteration of the metopic suture or
metopic perisutural sclerosis, bitemporal narrowing and narrow anterior fossa,
lateral orbital retrusion, and hypotelorism.

Treatment

The timing of surgical repair for metopic synostosis generally follows that
for coronal synostosis, with repairs ideally recommended around 5 or 6 months
of age. As in coronal synostosis, a bilateral procedure is recommended, advanc-
ing the lateral portions of the orbits forward, removing the midline keel, and
remodeling the frontal bone to correct the triangular shape of the forehead.
Orbital hypotelorism is treated, if necessary, by moving the two orbital roofs
laterally and plating a midline strut of bone between them. Rarely, the entire
orbits (both superiorly and inferiorly) need to be repositioned; this is more
commonly required in older children.

The Asymmetric Head—Plagiocephaly

Clinical Presentation

Children with plagiocephaly almost always have one of two conditions. The
less common of the two is frontal plagiocephaly, caused by unilateral coronal
synostosis (Fig. 7A). These children have flattening of the affected side of the forehead and flattening, lateral canthal retrusion, and slight superior displacement of the affected brow (giving a "raised eyebrow" appearance). The contralateral forehead is often slightly prominent compared with the affected side. In addition, several secondary clinical findings shore up the diagnosis. The occiput on the affected side is also flattened, giving a trapezoidal shape to the head. The root of the nose deviates toward, and the nasal tip away from the affected side, canting the nose. The midface is often mildly asymmetric as well (because of changes in the skull base rather than to midface hypoplasia), and a malalignment of the orbits may be present in the vertical plane (vertical dystopia). Finally, the coronal suture on the affected side often has a prominent ridge and is anteriorly displaced compared with the opposite side.

A far more common cause of plagiocephaly, and one that currently accounts for the majority of outpatient referrals to the pediatric neurosurgeon, is occipital plagiocephaly (also known as occipital flattening or positional molding) (Fig. 8A). Occipital plagiocephaly (OP) is caused by the child persistently lying supine with the head turned toward the affected side; at least 10% are associated with torticollis.\textsuperscript{19, 23} The incidence of this disorder has risen dramatically following the implementation of the American Academy of Pediatrics recommendations urging parents to sleep their infants in the supine position to reduce the risk for SIDS.\textsuperscript{1} Craniofacial clinics have witnessed as much as a 400% increase in referrals over the past several years, and many of these infants have been erroneously diagnosed with lambdoid synostosis.

Occipital plagiocephaly is best appreciated by looking down onto the top
of the child’s head. The clinical features are characteristic; ipsilateral occipital flattening and ipsilateral frontal bossing give a parallelogram shape to the head, as if the entire side of the head had been pushed anteriorly with respect to the other side. In contrast, both coronal and lambdoid synostosis produce both frontal and occipital flattening on the same (affected) side, yielding a trapezoidally shaped head. This distinction is critical to avoid misdiagnosis. The severity of occipital flattening is nearly always greater than the degree of frontal asymmetry.

Additional clinical features of OP are often present and include anterior (and sometimes inferior) deviation of the ipsilateral ear (best assessed by placing the fingers in both external auditory canals and looking down from above), and prominence of the ipsilateral malar eminence. The vertex may also slope up toward the affected side when viewed from the front, and some authors have suggested an inverse relationship between the degree of occipital flattening and

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Figure 7. Frontal plagiocephaly owing to unilateral coronal synostosis. A, Clinical features in an infant with left coronal synostosis. The forehead is flattened, the lateral brow elevated and retruded on the ipsilateral side. The contralateral forehead is prominent, and the nasal root is tilted toward the affected side. There is also mild flattening of the occiput on the affected side, giving a trapezoidal head shape. Radiographic features of unilateral coronal synostosis. B, Lateral view shows sclerosis and anterior deviation of the ipsilateral coronal suture with respect to the contralateral suture (crossed arrow), and elevation of the ipsilateral orbit (arrows). C, Frontal view shows a harlequin eye orbital deformity on the left, ipsilateral deviation of both the metopic suture (arrows) and the root of the nasal septum (crossed arrow). (Fig. B from Fernbach SK, Naidich TP: Radiological evaluation of craniosynostosis. In Cohen MM: Craniosynostosis: Diagnosis, Evaluation and Management. New York, Raven Press, 1986, p 197, copyright of Michael Cohen, MD; with permission.)

Illustration continued on opposite page
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Figure 7 (Continued).
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Figure 8. *See legend on opposite page*
Figure 8. Occipital plagiocephaly owing to positional molding. A, Infant with parallelogram-shaped head, showing right occipital flattening, anterior deviation of the right ear, and mild prominence of both the right forehead and malar eminence. (From Ripley CE, Pomatto J, Beals SP, et al: Treatment of positional plagiocephaly with dynamic orthotic cranioplasty. J Craniofacial Surg 5:150, 1994; with permission.) B, Water's view demonstrates perisutural sclerosis along the course of the right lambdoid suture (arrows). C, CT scan with bone windows in another patient demonstrates a parallelogram head shape and perisutural sclerosis along the left lambdoid suture (arrows). Note also the prominent subarachnoid spaces (BEFI). (A & B from Cohen M: Craniosynostosis: Diagnosis, Evaluation, and Management. New York, Raven Press, 1986, copyright of Michael Cohen; with permission.)

the degree of vertex sloping. A bald spot is often present on the affected side of the occiput where the child has lain on the head. The head may seem very round and wide when viewed from the front, and the ipsilateral orbit may appear slightly larger. The lambdoid suture may be ridged but is more commonly indented. Finally, as discussed earlier, some infants may have bilateral flattening that produces an occipital type of brachycephaly in which the calvarium gradually slopes back to a more posteriorly displaced vertex with a flattened occiput; although bilateral, usually one side is flatter than the other.

Unlike infants with craniosynostosis in whom the deformity is usually obvious at birth, three fourths of infants with OP have normal head shapes at birth and develop OP slowly over the first 3 or 4 months of life. Nearly 80% of affected infants lay supine with the head turned toward the side of the flattening during the first months of life. Eighty percent of affected infants are boys, and 80% of cases involve right-sided flattening. The right-sided predominance likely represents a marked predilection for normal infants to lie with their heads turned toward the right in a study of 30 infants with OP, the authors found a perfect concordance between the positional preference of the child and the side of the occipital flattening. The male preponderance may be explained by the common co-occurrence of BEFI (see the previous discussion of macrocrania) among infants with OP. BEFI also arises more frequently in boys than in girls (by a ratio of 2:1), and among boys with BEFI, a greater
proportion (48% versus 21%) also have OP (Partington, Dias, Li, and Winston, unpublished observations, 1997). These authors and others\textsuperscript{54} have proposed that the relative increase in intracranial water content may allow greater cranial deformation in these infants, although some have suggested the converse—that OP, by interfering with CSF absorption, may predispose infants to develop benign extra-axial collections.\textsuperscript{54}

Diagnostic Studies

In the child with frontal plagiocephaly, several radiographic features of unilateral coronal synostosis are almost always present (see Fig. 7B). The affected suture is indistinct, and perisutural sclerosis is common. The suture is deviated anteriorly compared with the affected side, so that both sutures are readily seen in different positions on true lateral radiography. Perhaps most importantly, the sphenoid wing of the skull base is elevated on the affected side, giving the characteristic unilateral harlequin eye deformity on the anteroposterior radiograph (see Fig. 7B). Axial CT scans show the indistinct suture, perisutural sclerosis, elevated sphenoid wing, flattened frontal bone and shallow anterior fossa on the affected side, and the relative bulging of both the contralateral forehead and the ipsilateral temporal fossa; coronal views show the harlequin eye deformity and deviation of the nasal bone.\textsuperscript{29}

The radiographic features of OP may be confusing. An indistinct portion of the lambdoid suture or perisutural sclerosis on plain radiographs and CT scans (Fig. 8B and C) may suggest lambdoid synostosis; however, the parallelogram, rather than trapezoidal, head shape should suggest a deformational problem rather than synostosis.\textsuperscript{34} Moreover, microscopic examination of the excised sutures has shown no histologic evidence of true synostosis in most infants.\textsuperscript{54} We proposed that infants with OP initially deform their skulls in a plastic way and, if their positioning is changed sufficiently early, the changes in skull shape are reversible; however, with time, secondary histologic changes in the sutures and basicranium (somewhat like a reactive “bone callus”) may produce radiographic changes that simulate synostosis.\textsuperscript{19}

Treatment

Surgery for unilateral coronal synostosis, as with the bilateral form, is ideally performed at approximately 5 or 6 months of age. The surgical management of unilateral coronal synostosis has changed over the past two decades. Previous treatments using a unilateral approach (advancing only the affected side of the forehead and the lateral portion of the affected orbit—a lateral canthal advancement) left some residual craniofacial deformities, such as contralateral frontal prominence and nasal deviation, which many authors now agree are better treated by a more extensive bifrontal craniotomy and bilateral orbitofrontal reconstruction similar to that proposed for bilateral coronal synostosis.\textsuperscript{37,75}

Three treatment options are available for infants with OP. At the outset, the goals of treatment must be kept in mind. First and most importantly, OP seems to be simply a cosmetic problem; there is no evidence that OP produces raised intracranial pressure, interferes with brain growth or development, or causes developmental delays (although delayed infants who spend a disproportionate amount of time in one position may be predisposed to develop OP). Therefore, all treatments must be recommended with the goal of improving cosmesis. Secondly, because OP is largely an occipital deformity, all but the most severe deformities are at least partially covered by hair as the child grows.
The first and most common treatment is to change the infant's sleeping position. A towel or blanket roll is placed (or affixed with Velcro or adhesive tape) under the affected side of the sleeping infant to encourage the infant to sleep on the contralateral side of the head. During the day (and only under close adult supervision), the infant can even be turned into a side-lying position with a commercially available foam side-lyer (this is not recommended at night because side lying may increase the risk for SIDS). These simple techniques are effective in most cases, and most parents are pleased with the result. A recent morphometric analysis of infants with mild and moderate occipital deformities showed a 50% improvement in measured asymmetries with positional changes alone.

The second treatment option is to use a cranial orthosis, or "molding helmet," which restricts calvarial growth at prominent areas while allowing continued growth at the points of flattening. Another orthotic device, the Dynamic Orthotic Cranioplasty, or DOC (Southwestern Orthotics, Phoenix, AZ), takes this one step further by dynamically molding prominent areas while allowing growth in flattened areas. Anthropometric measurements pretreatment and post-treatment with DOC demonstrate approximately a 40% improvement in cranial asymmetry. The third and currently the least common treatment is surgery, which was based on an older concept that OP was caused by lambdoid synostosis. This led to several surgical procedures designed to correct OP by removing the lambdoid suture (strip craniectomy) or reconstructing the occiput (unilaterally or even bilaterally). Surgery has become a much less common treatment for OP (although it is still effective for the rare true lambdoid synostosis) and is now generally used in only severe or recalcitrant cases. One procedure, lambdoid strip craniectomy, was evaluated using a CT morphometric analysis and improved calvarial asymmetries by only one third, less than the results obtained with DOC or simple positional changes. Based on the information available, the authors cannot recommend surgery except for severe cosmetic deformities that have failed more conservative treatments.

Microcephaly

Microcephaly (defined as a head circumference of less than the fifth percentile for age) may be caused by genetic predisposition or makeup (familial microcephaly or constitutional microcephaly), disordered brain development, prenatal or postnatal brain insults, or craniosynostosis. Craniosynostosis as a cause of microcephaly is extraordinarily rare. Most such cases involve fusion of multiple cranial sutures, and the deformity in head shape is obvious. The most severe deformity, kleebletschädel, is characterized by a trilobed head with gross, sometimes even monstrous, deformities. Others have either marked brachycephaly, turricephaly (tower skull), or oxycephaly (pointed skull).

An extraordinarily rare subgroup presents with microcephaly, relatively normal head shapes and facies, clinical evidence of intracranial hypertension, and radiographic evidence of delayed fusion of all cranial sutures. Among 665 patients with synostosis in one series, only four (presenting between 28 and 117 months) had delayed pansynostosis. One of the four had dolichocephaly, one had a family history of craniosynostosis, and another had a family history of hypophosphatasia (a condition known to predispose to craniosynostosis). If one excludes these three patients, then only 1 of 665 patients with craniosynostosis (less than 0.2%) presented de novo with isolated microcephaly, normal head shape and facies, and no family history. Therefore, in the absence of an abnor-
mal head shape or clear-cut signs of intracranial hypertension (e.g., extreme irritability, persistent vomiting, or papilledema), craniosynostosis is extremely unlikely to be the cause of isolated microcephaly.

**CRANIOSPINAL DYSRAPHISM**

Most contemporary texts divide dysraphic malformations into those that are open (e.g., anencephaly, myelomeningocele, and meningocele) and those that are occult (e.g., spinal lipoma, diastematomyelia, dermal sinus tracts, myelocystocele, thickened filum terminale, and caudal agenesis). Although superficially attractive, this classification scheme is confusing in that many of the “occult” malformations have clinically apparent cutaneous markers (e.g., hemangiomas, sinuses, appendages, or tufts of hair), which, for the astute clinician, serve as markers for the underlying spinal cord anomaly. The authors have instead recently begun to emphasize a classification based on reputed embryogenetic mechanisms (Table 2).

**Myelomeningocele**

Myelomeningocele is the most common dysraphic malformation compatible with life and occurs with a frequency of 1 in 1200 to 1400 live births. Myelomeningocele represents a localized failure of primary neurulation—a portion of the neural tube has failed to properly close, and the neural tissue (or placode) therefore remains attached to the skin (Fig. 9) and is, by definition, exposed on the back of the infant (i.e., there is no such thing as a closed myelomeningocele). The authors briefly review some of the salient features of myelomeningoceles; the interested reader is referred to several recent reviews for a more complete discussion.

It is important to give accurate and reliable information when counseling prospective parents who are carrying a fetus with a myelomeningocele. Most children with isolated myelomeningoceles (i.e., those without associated major anomalies of other organs) survive to adulthood, and the life expectancy is nearly normal. Eighty percent have normal intelligence, although of these, 60% have some learning disability (verbal scores are better than performance scores, and math and problem solving are particularly difficult). A peculiar form of

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**Table 2. EMBRYOGENETIC CLASSIFICATION OF COMMON SPINAL DEVELOPMENTAL MALFORMATIONS**

<table>
<thead>
<tr>
<th>Malformation</th>
<th>Reputed Embryogenetic Mechanism</th>
</tr>
</thead>
<tbody>
<tr>
<td>Myelomeningocele</td>
<td>Segmental failure of primary neurulation</td>
</tr>
<tr>
<td>Lipomyelomeningocele</td>
<td>Premature dysjunction (separation of neuro- and cutaneous ectoderm)</td>
</tr>
<tr>
<td>Dermal sinus</td>
<td>Incomplete dysjunction (separation of neuro- and cutaneous ectoderm)</td>
</tr>
<tr>
<td>Split cord malformations</td>
<td>Failure of midline axial integration during gastrulation</td>
</tr>
<tr>
<td>Thickened filum terminale</td>
<td>Disordered development of caudal cell mass</td>
</tr>
<tr>
<td>Myelocystocele</td>
<td>Unknown disorder of caudal cell mass</td>
</tr>
<tr>
<td>Sacral agenesis</td>
<td>Failure of development of caudal cell mass</td>
</tr>
</tbody>
</table>
attention deficit disorder without hyperactivity has been recognized in these children and may respond to methylphenidate (Ritalin). Hydrocephalus is present in 85% of children but bears little relationship to intelligence.\textsuperscript{40} Sixty percent of preadolescents are community ambulators (although this number decreases during adolescence), and about 80% are "socially continent" (meaning they are dry, although many perform clean intermittent catheterization).\textsuperscript{56}

Myelomeningocele is a static disease, and any deterioration in children with myelomeningocele should prompt a search for an underlying cause. Unfortunately, most children with myelomeningocele experience one or more episodes of clinical deterioration sometime during their lives. By far, the most common cause of deterioration is a shunt malfunction, which can present in a bewildering number of ways. Nowhere else in pediatric neurosurgery is clinical judgment so important (and misjudgment so treacherous) as in the evaluation of a child with myelomeningocele and suspected shunt malfunction. In addition to the usual triad of headache, nausea, and vomiting, these children may present with neck or back pain (especially at the myelomeningocele closure site); seizures (either new in onset or a change in a preexisting pattern of seizures); significant changes in behavior or school performance; swallowing or other evidence of hindbrain dysfunction; changes in upper or lower extremity strength, coordination, balance, or tone; changes in urinary or bowel habits; and scoliosis or other orthopedic deformities. Finally, as discussed earlier, children with shunt malfunction may have papilledema without any symptoms.\textsuperscript{20} In short, shunt malfunction in this population can be the root cause for any deterioration, and the clinician should always check the shunt before entertaining any other treatment options. If any doubt exists about shunt function, the shunt should be explored operatively before undertaking any other procedures.

Other causes of deterioration in children with myelomeningoceles include spinal cord tethering (occurring in approximately one third of children with myelomeningoceles) and hindbrain and spinal cord dysfunction caused by the Chiari malformation or syringomyelia (occurring in 15-20% of children with myelomeningoceles). Spinal cord tethering is identified radiographically by an
abnormally low position of the caudal spinal cord and produces symptoms in some children because the placode is bound down in the scar of the myelomeningocele closure and cannot move with the child's growth and movements. The Chiari malformation (Fig. 10) refers to a constellation of brain malformations, part of which is a descent of the cerebellar tonsils and caudal brainstem through the foramen magnum at the skull base and into the rostral spinal canal, which produces symptoms by compressing the brainstem and rostral spinal cord. Syringomyelia (Fig. 10) refers to a cystic collection of spinal fluid within the center of the spinal cord, which produces symptoms by stretching and compressing the adjacent spinal cord tissue.

Tethered cord and Chiari malformations are present radiographically in nearly every child with myelomeningocele, and syringomyelia is present in 20% to 40%.56, 57, 98 The mere presence of any of these malformations does not, by

Figure 10. Sagittal MR scan of a child with myelomeningocele and Chiari II malformation. The cerebellar vermis and caudal brainstem are descended through the foramen magnum into the cervical canal (large arrows). A smaller cervical and larger thoracic syringomyelia cavities are also present (small arrows). (From Dias MS, McLone DG: Spinal dysraphism. In Weinstein SL: The Pediatric Spine: Principles and Practice. New York, Raven Press, 1994, p 361; with permission.)
itself, suggest the need for treatment. Rather, treatment is reserved for children with evidence of clinical deterioration. Spinal cord tethering may produce back or leg pain, deterioration in lower extremity function, changes in urinary or bowel function, and scoliosis or other orthopedic deformities. The Chiari malformation may produce headache or neck pain, lower cranial nerve dysfunction (including swallowing difficulties, stridor or vocal cord palsy, recurrent aspiration pneumonia, or apnea), upper extremity deterioration, ataxia or gait disturbance, and scoliosis. Syringomyelia may produce segmental motor weakness, lower extremity hyperreflexia or spasticity, loss of sensation (classically described as a dissociated loss of thermal and pain sensation), and worsening gait; bowel or bladder dysfunction is rare. A complete discussion of these entities is beyond the scope of this article, and the interested reader is referred to recent monographs for a more complete discussion.

Occult Spinal Dysraphism

Although recognizing myelomeningoceles, more problematic is identifying children with occult craniospinal dysraphism, including spinal lipomas, split cord malformations (diastematomyelia or diplomyelia), cranial or spinal dermal sinus malformations and dermoid tumors, neurenteric cysts, myelocystoceles, thickened and fat infiltrated filum terminale, and caudal agenesis. Although some of these malformations are truly occult, most have some sort of associated cutaneous marker that, if appreciated, direct attention to the underlying spinal cord lesion. Recognizing these cutaneous markers as signs of underlying spinal cord malformations is critical because any of the associated dysraphic malformations can cause spinal cord injury from tethering and lead to progressive and sometimes sudden neurological deterioration, and, although they usually can be stabilized by spinal cord untethering, neurologic, urologic, and orthopedic problems are often irreversible when they occur. For these reasons, most pediatric neurosurgeons recommend treatment of the spinal cord malformation prophylactically in infancy, before the onset of symptoms.

These occult spinal cord malformations bind, or tether, the spinal cord in an abnormally low position (spinal cord tethering is generally defined radiographically as a conus medullaris, that lies caudal to the mid-body of the second lumbar vertebra). Tethering prohibits the conus from rising with continued somatic growth and in most patients eventually results in progressive spinal cord dysfunction as a result of stretching, ischemia, and decreased mitochondrial oxidative metabolism. Tethering from any cause may produce back, leg, or groin pain; progressive lower extremity sensorimotor dysfunction (weakness, contractures, reflex changes, and sensory abnormalities or loss); urinary dysfunction from a neurogenic bladder; bowel incontinence or constipation; lower extremity orthopedic deformities, such as pes cavus, hammer toes, and equinoval varus deformities; and progressive scoliosis.

Spinal Lipomas (Lipomyelomeningocele)

Clinical Presentation. Spinal lipomas are the most common of the occult dysraphic malformations. An associated subcutaneous fatty mass (Fig. 11A) is present in 70% of patients. Other cutaneous markers of spinal lipomas include midline lumbosacral hemangiomas, dimples, skin tags, or appendages. Spinal lipomas are thought to arise through premature separation (dysjunction) of the
neural tube from the cutaneous ectoderm (skin); this allows mesenchymal cells access to the inside of the neural tube, where they are induced to form fat.

**Diagnostic Studies.** An MR image shows the lipoma as a hyperintense lesion on T1-weighted images (Fig. 11B). Preoperative workup should also include a formal assessment of lower extremity muscle function (a manual muscle test, or MMT), a urologic evaluation including urodynamic studies, and an orthopedic assessment of lower extremity deformities.

**Treatment.** Surgery involves debulking the lipoma, removing its attachment with the spinal cord, and untethering the spinal cord. A cosmetic excision of the subcutaneous lipoma without addressing the underlying spinal cord anomaly is not adequate. The subcutaneous component of the lipoma always extends through a dorsal defect in the spine (often, but not always, between dysplastic

Figure 11. See legend on opposite page
**Figure 11.** Spinal lipoma. A, A subcutaneous fatty mass underlies the lumbosacral region. Note the hemangioma in the overlying skin. B, Sagittal T-1 weighted MR scan in another infant shows a hyperintense fatty mass attached to the dorsal aspect of the spinal cord. C, At surgery, the subcutaneous lipoma extends into the dorsal aspect of the spinal cord (arrows).

laminae), and ends within the intramedullary portion of the spinal cord (Fig. 11C). Rarely, the lipoma may be purely intradural, having no subcutaneous component, but still tethers the spinal cord.

**Dermal Sinus Tracts**

Dermal sinus tracts are thought to represent incomplete dysjunction (that is, separation of the neural tube from cutaneous ectoderm) during neurulation. As a result, a tract of cutaneous tissue remains attached to the nervous system. The ostium of the lumbosacral dermal sinus tract overlies the lumbosacral spine, and the stalk penetrates the underlying spinal canal to end on the dorsum of the spinal cord just above the tip of the conus medullaris. In addition to tethering the spinal cord, dermal sinus tracts can cause symptoms and signs through at least three other mechanisms. First, they can serve as a portal of entry for bacteria leading to recurrent infections (bacterial meningitis or spinal abscess). Second, dermoid tumors can develop within the spinal cord or canal and compress the spinal cord. Third, desquamation of epithelial cells and debris from the dermoid tumor can produce an intense inflammatory response, resulting in aseptic meningitis.53

**Clinical Presentation.** Dermal sinus tracts almost always have some associated cutaneous marker, usually the tract itself, which produces a lumbosacral dimple. Dermal sinus tracts should be differentiated from the benign coccygeal dimple, which is present in approximately 4% of normal infants.53 As its name implies, the benign coccygeal dimple is located at or just above the tip of the coccyx within the gluteal cleft. The tip of the coccyx is readily palpated deep to
the dimple. In contrast, the dermal sinus tract is located higher, in the "flat" of the lumbosacral area, well rostral to the coccyx and almost always cranial to the end of the gluteal cleft (Fig. 12). Dermal sinus tracts may be irregular, surrounded by heaped up areas of skin or small dermal masses (Fig. 12), or associated with cutaneous hemangiomas, skin tags, or tufts of hair within the ostium. Abnormal or asymmetric forking of the gluteal cleft is common; the dermal sinus tract may be located at the end of one fork. Finally, dermal sinus tracts may be associated with focal neurologic deficits, a neurogenic bladder, or orthopedic deformities. Benign coccygeal dimples are never associated with any of these findings.53

Cranial dermal sinus malformations are most commonly located either anteriorly (frontonasal sinus tracts) or in the occiput (occipital sinus tracts); rarely, they may be parietal in location. Frontonasal dermal sinus tracts typically begin along the dorsum of the nose, anywhere between the tip and the nasion (Fig. 13), and travel between the skin and nasal cartilage to end at or near the anterior skull base. Although they often appear innocuous and may end harmlessly in the extracranial space, some sinuses extend intracranially through a tiny defect in the anterior skull base near the foramen cecum and serve as a portal of entry for intracranial infection (meningitis or brain abscess) or as a source of intracranial dermoid tumors.61, 68, 96 Similarly, the occipital dermal sinus tract can also have intracranial extension into the supratentorial compartment, the posterior fossa, or both with similar complications.12, 95

Diagnostic Studies. An MR image of the lumbar spine shows the spinal cord to be tethered. Dermoid tumors, if present, may be seen within the thecal sac or spinal cord parenchyma but may be isointense to CSF or cord and, therefore, difficult to visualize. Unfortunately, dermal sinus tracts are not always

Figure 12. Dermal sinus tract. Buttocks of a child with a dermal sinus (arrowhead) and surrounding subcutaneous dermoid tumor (small arrows).
Figure 13. Fronto-nasal dermal sinus tract. An infant with a small dimple at the nasion (arrowhead).

visible on MR imaging, and the absence of a visible tract does not exclude a connection. If the lesion seems clinically suspicious, especially if the conus is abnormally low, the malformation should be explored regardless of whether a tract is visible on MR imaging.

The radiographic workup of cranial dermal sinus tracts should include both cranial MR imaging and CT scans. The dermal sinus tract or associated dermoid tumors may best be visualized on MR scans with gadolinium enhanced sagittal and coronal views, but a small bony defect may be visible only on coronal CT scans with bone windows, even though an intracranial abnormality on MR imaging cannot be found.

Treatment. As with the lumbosacral dermal sinus tracts, all suspicious lesions should be explored even if radiographic studies are negative. If there is known intracranial extension, the entire tract is explored and removed, either in a combined extracranial and intracranial procedure or in two separate procedures. If no visible intracranial extension is present, the extracranial tract is usually excised locally and the tract followed toward the skull base; an intracranial exploration is performed if intracranial extension is found at the time of surgery.

Split Cord Malformations

Split cord malformations (SCM) are anomalies in which the spinal cord is split or clefted over a portion of its length. The terms diastematomyelia and diplomyelia were used in the past but have generated a great deal of confusion; therefore, these terms have been largely supplanted by the common term split cord malformation. Both malformations seem to result from a disorder of midline axial integration during gastrulation to produce a split neural tube.

Two types of SCMs are recognized. Type 1 lesions are double dural sac malformations in which both the spinal cord and dural sac are split, and there is an extradural bony or fibrocartilaginous spur interposed between the two thecal sacs (Fig. 14A). Type 2 malformations are single dural sac malformations in which the spinal cord, but not the thecal sac, is clefted, and therefore, both
Figure 14. Split cord malformations (SCM). A, CT myelogram in a child with a type I SCM shows two hemicords (arrowheads) each within its own dural sac, and separated by a bony septum (arrows). (From Pang D, Dias MS, Ahab-Barmada M: Split cord malformation: Part I: A unified theory of embryogenesis for double spinal cord malformations. Neurosurgery 31:452–480, 1992; with permission.) B, CT myelogram in a child with a type II SCM contains two hemicords (arrows) within a common dural sac. No intervening septum is apparent, but a septum was encountered at surgery. (Courtesy of D. Pang, MD.) C, Lumbar hypertrichosis (fawn's tail) in a child with a type I SCM.

hemicords are contained within a common dural sac (Fig. 14B). Although the earlier literature suggested that the type 2 malformations did not contain any tethering element, at surgery a fibrous band of tissue (analogous to the bony spur of the type 1 malformations) is usually found between the two hemicords. Therefore, both types of malformations contain a tethering element that results in neurologic deterioration.71

Clinical Presentation. Cutaneous stigmata are present in as many as 80% of patients with SCMs. Although the most common (20–55% of patients) and specific of these is a focal area of hypertrichosis (the “fawn’s tail”; Fig. 14C),
other cutaneous stigmata include cutaneous hemangiomas, dimples, lipomas, and bony abnormalities. Vertebral malformations are common and may be multiple, particularly with the type 1 lesions; hemivertebrae, sagittally clefted (butterfly) vertebrae, and missing or duplicated vertebrae have all been described. Rarely, a portion of the spinal column is completely split (the split notochord syndrome). Associated enteric malformations (e.g., diverticula, duplications, ectopic bowel, or neurenteric cysts) and renal anomalies (e.g., agenesis, duplication, horseshoe kidneys, or pelvic kidneys) may also be present.

**Diagnostic Studies.** Radiographic evaluation of SCMs includes MR imaging or a spinal CT scan with intrathecal contrast (a CT myelogram). Although the sensitivity of spinal MR imaging is improving, CT myelography is more sensitive at evaluating the details of these malformations for surgical planning. High-resolution CT myelography is particularly good for identifying thin intradural fibrous tethering bands in the type 2 malformations, which may not be visible on MR imaging. Approximately 15% of SCMs have tandem lesions, so that complete spinal MR imaging is necessary.

**Treatment.** Surgical treatment is usually recommended prophylactically, before these children develop signs and symptoms. At surgery, the tethering bony spurs (type 1) or fibrous bands (type 2) are resected and, in the type 1 malformations, the dural cuff between the two hemicords is excised and a single dural sac reconstituted (described as "converting pants to a dress"). Finally, the filum terminale is usually short and thickened and is sectioned as well.

**Thickened Filum Terminale**

The filum terminale is a nonfunctional strand of tissue that projects from the end of the conus to the bottom of the thecal sac. A thickened filum terminale is defined as one that is greater than 2 mm thick (either on MR scans or CT myelography). As many as 90% have fat within the filum terminale (the so-called "fatty filum"), and 86% are associated with a low-lying conus medullaris. Although as many as 6% of normal individuals have some fat within the filum, the association of a thickened, fat infiltrated filum and a low-lying conus suggests a tethering lesion. As many as 50% of patients have cutaneous markers, but many patients first present with sensorimotor findings or a neurogenic bladder.

Plain radiographs sometimes demonstrate a defect in one or more vertebral laminae (spina bifida occulta), and MR imaging (Fig. 15) demonstrates the low-lying spinal cord and a filum terminale, which is thickened and infiltrated with fat (producing a hyperintense strand of tissue connecting the conus and the distal thecal sac on T1-weighted images). Surgical treatment involves sectioning the filum terminale near its caudal end.

**Terminal Myelocystocele**

A myelocystocele is a rare caudal spinal cord malformation in which a dilation of the caudal end of the spinal cord is present, with a corresponding dilatation of the caudal thecal sac, giving a "double-bubble" appearance to the distal neuraxis on MR imaging. Usually an associated lipoma is also present. The size is variable, but some can reach a monstrous size (Fig. 16). Clinically, the lesions appear as closed, skin-covered, lumbosacral masses that resemble a spinal lipoma. Many of these have erroneously been termed "closed myelomeningoceles" (a misnomer because, as previously discussed, a myelomeningocele...
cannot, by definition, be closed). The embryogenesis is unknown but probably involves abnormal development of the caudal cell mass (the multipotent ball of cells that remains under the skin at the caudal end of the embryo after primary neurulation is completed and that gives rise to the caudal neural tube and filum terminale during secondary neurulation). Surgical treatment involves repairing the myelocystocele and untethering the spinal cord from the lipoma.

Caudal Agenesis

The syndrome of caudal agenesis is usually included in the spectrum of dysraphic malformations, although tethering or other neurosurgical lesions are less common in these patients. The coccyx and part or all of the sacrum is usually missing; rarely, the agenesis can extend rostrally to involve the lumbar or even lower thoracic segments. The syndrome is more common in the offspring of diabetic mothers (1% of the offspring born to diabetic mothers have sacral agenesis, and 16% of children with sacral agenesis are offspring of diabetic mothers); both hyperglycemia and ketones have been implicated in the embryopathy.

The child is born with varying degrees of paralysis and atrophy of the distal leg musculature producing an “inverted champagne bottle” appearance. The buttocks are flattened and atrophic. A neurogenic bladder is almost universal. Sensory function is curiously preserved.

Lumbosacral spine radiographs reveal absence of a variable portion of the
Figure 16. Myelocystocele. A, Infant with a huge skin-covered lumbosacral mass. The child also had bilateral equinovarus deformities and exstrophy. B, Sagittal T1-weighted MR image shows a huge double compartment cystic mass. The spinal cord ends in a terminal dilatation (arrows) and is surrounded by a larger dilatation of the thecal sac, giving the appearance of a sac within a sac. (Courtesy of Keith Aronyk, MD, Department of Neurosurgery, University of Alberta at Edmonton.)
caudal spinal column, most commonly including part or all of the sacrum; hemivertebrae or fused vertebrae may also be seen. All children with sacral agenesis should receive baseline lumbosacral MR imaging to exclude important spinal cord anomalies. The spinal cord in caudal agenesis ends bluntly (Fig. 17), confirming the absence of the distal conus medullaris. Although sacral agenesis is a static malformation which, in and of itself, need not be treated, tethering lesions, such as myelomeningoceles, split cord malformations, lipomas, dermoid tumors, fibrous bands, and thickened filum terminale, can all cause neurologic deterioration in these patients and should be treated if found. In addition, bony or dural stenosis of the spinal canal may cause progressive deterioration and may also require treatment.

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