

29. Congenital Abnormalities

Congenital posterior fossa abnormalities are frequently visualized on cervical spine MRI, often with accompanying neural axis and osseous findings. Sagittal images best evaluate the position of the cerebellar tonsils, which may normally lie as far as 5 mm below the foramen magnum. Extension of the tonsils below this level is seen in type 1 Chiari malformations, which are commonly asymptomatic. A symptomatic case—with the lack of space surrounding the medulla at the level of the foramen magnum leading to CSF obstruction—is demonstrated in Figure 29.1 A. Cerebellar herniation must be assessed with attention to tonsillar location relative to the anterior and posterior arches (arrows) of C1, as well as the foramen magnum. The tonsils frequently appear pointed in a type 1 malformation in contrast to their normal globular appearance. Clinical symptoms may also be due to the presence of hydromyelia, illustrated in addition in Figure 29.1 A.

Hydromyelia occurs secondary to dilatation of the central canal of the spinal cord and thus appears on MRI as an area of CSF SI within an enlarged cord. Cord wall thinning (posteriorly in Fig. 29.1 A) may occur in severe cases. Hydromyelia may occur at any location within the cord but most commonly within the low cervical/upper thoracic region when associated with Chiari malformations. With severe hydromyelia, the walls of the cord are markedly thinned and compressed against the adjacent dura. Intraspinal arachnoid cysts are a differential consideration but are quite rare. Hydromyelia involving the medulla is termed syringobulbia and is often symptomatic. The preservation of the fourth ventricle and the normal appearing midbrain colliculi help distinguish the Chiari 1 malformation of Figure 29.1 A from the type 2 malformation illustrated in Figure 29.2 B. In the latter, the fourth ventricle is usually slit-like, and the tectum is fused resulting in a beak-like appearance. In addition, in this instance, a cerebellar peg is present, the anterior-posterior dimension of the pons is foreshortened, and the insertion of the tentorium is low—all typical features of type 2 Chiari. Associated osseous and spinal abnormalities include

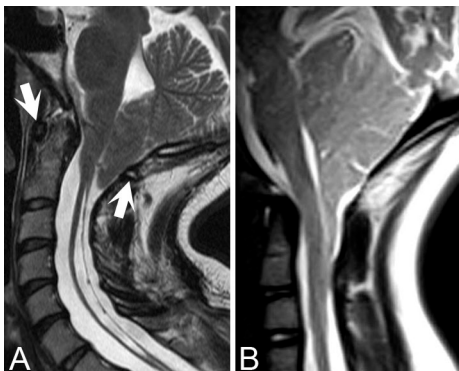


Fig. 29.1

anomalies of the posterior arch of C1 and lumbar spinal dysraphism (commonly a myelomeningocele), the latter seen in nearly all Chiari 2 patients. Additional cerebellar and brainstem findings as described in Chapter 23 may be present along with hydromyelia. The most significant osseous congenital abnormalities of the cervical spine include malformations of the craniocervical junction. Basilar invagination refers to the presence of the tip of the odontoid 5 mm or more above Chamberlain's line (drawn from the posterior margin of the hard palate to the posterior lip of the foramen magnum; see Chapter 33). Platybasia—marked by an angle between the clivus and the floor of the anterior cranial fossa greater than the normal 125-140 degrees—often accompanies this condition. Primary invagination is also associated with occipitalization (fusion of the atlas and occiput), whereas secondary lesions occur in osteoporosis, Paget's disease, achondroplasia, fibrous dysplasia, and osteogenesis imperfecta. Os odontoideum is defined by the presence of an ovoid ossicle distinct from the body of C2 and may be difficult to distinguish from a traumatic odontoid fracture, although the former may also arise secondary to trauma. Hemi- and fused vertebrae are also commonly encountered congenital disorders. The most common fusion anomaly is simply the fusion of two adjacent vertebral bodies, seen not infrequently in the cervical spine. Typically, the two fused vertebral bodies have a small AP dimension, with the intervening disk space often only partially visualized. Figure 29.2 A, B demonstrates multiple anomalies including right segmented hemivertebrae (C5, C6), fusions of the posterior elements (C1 to C2) and a wedge-shaped vertebral body (C7). As evident from this case, coronal imaging is essential (in addition to the sagittal and axial planes) for the proper evaluation of congenital abnormalities of the spine. The presence of hemivertebrae leads to a short, focal curve, often with severe scoliosis. Klippel-Feil syndrome is characterized by the congenital fusion of two or more cervical vertebrae. A short neck with limited mobility and a low posterior hairline may also be present. Three

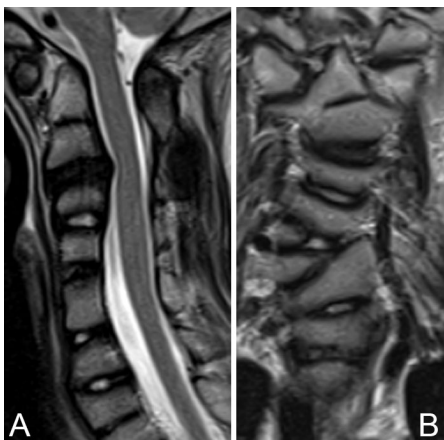


Fig. 29.2

types are defined (by Samartzis in 2006), which include a single congenitally fused cervical segment (type I), multiple noncontiguous, congenitally fused segments (type II), and multiple contiguous, congenitally fused cervical segments (type III). Figure 29.3 A demonstrates hypoplastic, fused C2-C4 vertebral bodies lacking intervening disk spaces. In this particular case the cervical spine was found to be fused from C1 to C6, and the case thus classified as Klippel-Feil type III. Diastematomyelia—a symmetric or asymmetric longitudinal splitting of the spinal cord—is not uncommonly seen in Klippel-Feil (as in Fig. 29.3 B). In Figure 29.3 B, the hemi cords appear tethered together by a fibrous band and are enveloped by a single dural tube (type 2). Type 1 diastematomyelia includes clefting of the meninges such that each hemicord is enclosed within its own dural sac typically with an intervening cartilaginous or bony spur between them. Both types of diastematomyelia are associated with multiple congenital and spinal abnormalities and also with an overlying cutaneous patch of hair (or other cutaneous stigmata).



Fig. 29.3