legs, and loss of bladder control. Nearly all children with MMC have an associated Chiari malformation, "type II" (CM2, also called an Arnold-Chiari malformation). CM2 is characterized by displacement of the cerebellum and lower brainstem through the foramen magnum, usually causing obstructive hydrocephalus. Spina bifida does not have to be associated with an open defect. In cases of closed NTD, patients may present with leg spasticity, foot deformities, and bladder abnormalities, and the overlying skin may show nevi, lipomas, abnormal dimples, or hairy tufts.

Diagnosis/Differential

MRI is the method of choice for evaluation of NTDs. In the case of a child with an open caudal defect, the imaging should look for other associated nervous system abnormalities, such as CM2 and hydrocephalus. Closed NTDs are definitively diagnosed by MRI, but patients may present with symptoms later. Other disorders that can present with gait abnormalities and deformities include spastic diplegia, vitamin B12 deficiency, multiple sclerosis, and other conditions presenting with spastic paraparesis.

Treatment

Treatment is surgical repair of the MMC with subsequent shunting of the hydrocephalus. Bladder dysfunction may necessitate intermittent catheterization and treatment of urinary tract infections and genitourinary reflux. A recent clinical trial indicated that fetal surgery before 26 weeks gestation to repair the MMC defect resulted in better neurologic outcomes, possibly by preventing the spinal cord injury and CSF leakage that can occur during the third trimester with an open defect.

Prognosis

Infants with open NTDs such as MMC have a much more severe presentation and course than those born with closed defects. Fetal surgery at qualified centers may improve MMC outcome.

Syringomyelia

Definition/Epidemiology

Syringomyelia or syrinx is a cystic cavitation of the central portion of the spinal cord. The estimated prevalence of 8/100,000 is likely an underestimate.

Pathology

The central canal cysts are most commonly located in the cervical spine and consist of CSF-filled space lined by glial cells, in contrast to hydromyelia, where the dilated central canal is lined with ependymal cells. The syrinx can be septated and irregular, and it may develop in association with Chiari malformations (CM1 and CM2), trauma, tumor, or a tethered cord.

Clinical

The classic presentation is a dissociated sensory loss (pain and temperature loss with preservation of light touch and proprioception) in the neck, arms, or legs. A cervical lesion produces a cape-like dissociated sensory loss of the arms and shoulder, along with atrophy of the hands and arms with increased tone and hyperreflexia in the legs. Extension into the medulla (syringobulbia) may cause lower cranial neuropathies.

Diagnosis/Differential

Diagnosis is confirmed by MRI, which will also differentiate the cysts from neoplasms, infections, and other spinal cord lesions.

Treatment

If the syrinx is associated with CM1 or CM2, then posterior fossa decompression or shunting of the hydrocephalus may improve the symptoms. Direct evacuation or shunting of the syrinx itself is less frequently done and not of established benefit.

Prognosis

Syringomyelia can be slowly progressive but spontaneous resolution has been seen. Therefore, conservative treatment has been advocated, especially in children.

DEVELOPMENTAL DISORDERS

Autism Spectrum Disorder

Definition/Epidemiology

Autism spectrum disorder (ASD) is characterized by 1) impaired social communication and interactions and 2) restricted and repetitive behaviors. The prevalence of ASD is 1/88 and is four times higher in boys than girls.

Pathology

The marked social impairments that characterize ASD are not associated with specific pathology or physical findings.

Clinical Presentation

ASD presents in early childhood with lack of interest or inclination to relate to others. Young children with autism may be physically healthy with good motor skills, but they are hard to engage, do not reliably respond to their name being called, and are slow to develop social and communicative gestures such pointing and waving.

Diagnosis/Differential

The lack of a clear biologic marker or simple clinical test means that the diagnosis of ASD relies on careful evaluation of the child by experienced examiners. (See Table 115-2 for DSM-V criteria for ASD). ASD may be difficult to diagnosis or distinguish from other forms of mental retardation and psychiatric disorders. ASD should be distinguished from acquired encephalopathies (e.g., epilepsy or encephalitis).

Treatment

The mainstay of treatment is prompt initiation of appropriate behavioral and early enrichment services. The treatments for ASD are aimed at improving social interactions and communication; not surprisingly, there is no set treatment that works for all. Rather, treatment of this disorder often requires coordination of medical, educational, and community services.

Prognosis

Patients with ASD respond to appropriate treatments over time, but these may be highly resource intensive. Some children with ASD, especially those who have normal verbal and intellectual