

17.96 per 100,000 live births, thus making this one of the most common birth defects in the United States (Matthews, 2009; Wolff, Witkop, Miller, et al, 2009). Increased use of prenatal diagnostic techniques and termination of pregnancies have also affected the overall incidence of NTDs (see also Prevention, later in chapter).

**Anencephaly**, the most serious NTD, is a congenital malformation in which both cerebral hemispheres are absent. The condition is usually incompatible with life, and many affected infants are stillborn. For those who survive, no specific treatment is available. The infants have a functional portion of the brainstem and are able to maintain vital functions (e.g., temperature regulation and cardiac and respiratory function) for a few hours to several weeks but eventually die of respiratory failure.

**Myelodysplasia** refers broadly to any malformation of the spinal canal and cord. Midline defects involving failure of the osseous (bony) spine to close are called **spina bifida (SB)**, the most common defect of the CNS. SB is categorized into two types—SB occulta and SB cystica.

**Spina bifida occulta** refers to a defect that is not visible externally. It occurs most frequently in the lumbosacral area (L5 and S1) (see Fig. 30-5, B). SB occulta may not be apparent unless there are associated cutaneous manifestations or neuromuscular disturbances.

**Spina bifida cystica** refers to a visible defect with an external saclike protrusion. The two major forms of SB cystica are **meningocele**, which encases meninges and spinal fluid but no neural elements (see Fig. 30-5, C), and **myelomeningocele** (or **meningomyelocele**), which contains meninges, spinal fluid, and nerves (see Fig. 30-5, D). Meningocele is not associated with neurologic deficit, which occurs in varying, often serious, degrees in myelomeningocele. Clinically, the term *spina bifida* is used to refer to myelomeningocele.

## Pathophysiology

The pathophysiology of SB is best understood when related to the normal formative stages of the nervous system. At approximately 20 days of gestation, a decided depression, the neural groove, appears in the dorsal ectoderm of the embryo. During the fourth week of gestation, the groove deepens rapidly, and its elevated