Encephaloceles

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Encephaloceles Information Page.

Encephaloceles are rare neural tube defects characterized by sac-like protrusions of the brain and the membranes that cover it through openings in the skull. These defects are caused by failure of the neural tube to close completely during fetal development. The result is a groove down the midline of the upper part of the skull, or the area between the forehead and nose, or the back of the skull. When located in the back of the skull, encephaloceles are often associated with neurological problems. Usually encephaloceles are dramatic deformities diagnosed immediately after birth, but occasionally a small encephalocele in the nasal and forehead region can go undetected. Encephaloceles are often accompanied by craniofacial abnormalities or other brain malformations. Symptoms and associated abnormalities of encephaloceles may include *hydrocephalus* (excessive accumulation of cerebrospinal fluid in the brain), *spastic quadriplegia* (paralysis of the arms and legs), *microcephaly* (abnormally small head), *ataxia* (uncoordinated movement of the voluntary muscles, such as those involved in walking and reaching), developmental delay, vision problems, mental and growth retardation, and seizures. Some affected children may have normal intelligence. There is a genetic component to the condition; it often occurs in families with a history of spina bifida and anencephaly in family members.