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ENCEPHALOCELE

Prevalence

Condition Description

Encephalocele (en-**SEF**-ah-lo-SEEL) is a type of neural tube defect (NTD) that occurs very early in fetal life where the embryo's cells that form the skull do not come together to close over the brain. The result is a defect in the bones of the skull, causing brain tissue to protrude from the skull. The protruding tissue may be located on any part of the head but is usually in the middle at the back of the head (midline occipital area). There may be meninges, brain tissue, parts of the ventricles and bone that protrude from the skull.

Alphafetoprotein levels are rarely elevated because almost all encephaloceles are covered by skin and therefore this identifying protein does not leak into the amniotic fluid as it does with most other NTD conditions.

Encephalocele is typically diagnosed by prenatal ultrasound. The best finding would be to find only fluid in the protruding sac.

Encephaloceles located on the front of the skull are more likely not to contain brain tissue and have better outcomes than those at the back of the head.

The cause of encephalocele is not known but it is probably a combination of factors such as maternal nutrition, a genetic predisposition, and exposure to toxins or infections very early in fetal life. Neural tube defects appear to be associated with maternal lack of folic acid, and supplementing the diets of all women of childbearing age with folic acid is decreasing the rate of these disorders.

Encephaloceles occur at a rate of 1 in 5,000 live births and are often associated with stillbirth before 20 weeks gestation. Only 20% of babies with encephaloceles are born alive, and only half of those survive. For an unknown reason, 70% of occipital encephaloceles occur in females, whereas anterior encephaloceles are more often seen in males.

Common Associated Conditions

Up to 40% of encephaloceles occur in babies who have chromosomal abnormalities. Other conditions that often occur together with encephaloceles are Dandy-Walker malformation (enlargement of the 4th ventricle with a cyst that extends into the brain's posterior fossa) and Chiari malformation (cerebellum of the brain extends into the spinal canal, blocking cerebral spinal fluid (CSF) flow and causing neurological symptoms). Meckel syndrome is a term used when an occipital encephalocele is associated with microcephaly, microphthalmia, cleft lip and palate, polydactyly, polycystic kidneys and ambiguous genitalia.

Short-term Treatment and Outcomes

A computerized tomogram (CT) scan will be done immediately after birth to determine the extent of the lesion and the location of major structures and blood vessels. Treatment will be immediate surgery to remove the sac and the neural tissue it contains, which will be abnormal. Hydrocephalus typically results and will need to be corrected by surgical shunting



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ENCEPHALOCELE – page 2

procedure. The outcome will depend on the amount of brain tissue in the encephalocele and whether child has other anomalies.

Long-term Treatment and Outcomes

Long-term treatment depends on extent of child's condition. Multi-stage corrective surgery may be needed, depending on location of encephalocele and structures involved.

Common Complications

Meningitis is a post-operative concern and antibiotics will be given before and after surgery. Hydrocephalus and shunting will typically be necessary to prevent build-up of cerebral spinal fluid. Because the vision center is located in the back of the brain, some babies who survive with encephalocele will be blind. Occulta (hidden) encephaloceles in the front of the skull and involving the bones of the nasal and sinus areas can sometimes cause problems later in life if brain tissue herniates through the bony defect.

Implications for Children's Development

At least half of survivors of encephalocele have significant cognitive deficits. The involved brain structures will determine the type of deficit, but significant motor and learning deficits are common. Children will need frequent interdisciplinary assessment and follow-up by subspecialists in neurology, neurosurgery and pediatric rehabilitation. Early intervention programs through the school system will assist the family to maximize the child's development and focus on developing the child's strengths.

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