Encephalocele

BIRTH DEFECT RESEARCH FOR CHILDREN

What is Encephalocele?

Encephalocele is a birth defect that results in a hole in the skull through which brain tissue protrudes. In the Western Hemisphere, the most common Encephaloceles, called Occipital Encephaloceles, are located in the back of the head. Occurring less frequently are Frontoethmoidal Encephaloceles which are located in the area around the eyes and nose. This type of Encephalocele is most common in Asia. A small percentage of Encephaloceles are located around the nasal cavity. Other brain malformations and craniofacial abnormalities often occur with Encephaloceles. Associated abnormalities and symptoms of Encephalocele may include hydrocephalus (excessive accumulation of cerebrospinal fluid in the brain), microcephaly (abnormally small head), paralysis of the arms and legs, seizures, developmental delays, mental and growth retardation, and vision problems.

Encephalocele

How many children have Encephalocele?

Encephalocele is a rare disorder affecting one to four babies per 10,000 live births in the United States. It is more common in girls than in boys. At least 60% of the babies with Encephalocele have other malformations and/or chromosomal anomalies. Twenty percent of fetuses with Encephalocele are stillborn.

How do you know if your child has Encephalocele?

Most cases are diagnosed prenatally with ultrasonography or MRI (magnetic resonance imaging). Diagnosis generally is not possible before skull ossification begins at ten weeks gestation. Once diagnosed, a thorough search for associated abnormalities is usually performed. If you know in advance that your baby may be born with Encephalocele and other abnormalities, you can arrange for prompt and expert medical treatment. Usually Encephaloceles are deformities apparent at birth, but diagnosis of a Frontoethmoidal Encephalocele may be delayed until a nasal mass, nasal congestion, or a facial abnormality become apparent. Children with Frontoethmoidal Encephaloceles are often neurologically normal.

What causes Encephalocele?

Encephaloceles are caused by failure of the neural tube to close completely during fetal development. The neural tube is the structure that gives rise to the central nervous system (the spinal cord and brain). Neural tube defects (NTDs) are one of the most common birth defects causing infant death or serious disability. Genetic and geographic factors have been implicated in the cause of Encephalocele, and it often occurs in families with a history of NTDs including spina bifida (a defect of the spinal column) and anencephaly (underdeveloped brain and incomplete skull).

Environmental triggers for NTDs that are being studied include viruses, chemicals, drugs, vitamin and mineral deficiencies, and maternal illness like diabetes. Research has suggested that the majority of NTDs could be prevented if women consume supplements containing folic acid prior to and in the early months of pregnancy. Experts now recommend that, under doctor supervision, all women of childbearing age who might become pregnant consume 400 mcg of folic acid daily.

How can you help a child with Encephalocele?

Most babies undergo surgery to place the tissues back into the skull and to correct any craniofacial abnormalities. If hydrocephalus is present, surgical treatment with a shunt may be necessary. Other treatment may be necessary depending on the symptoms and associated abnormalities.

What's in the future for a child with Encephalocele?

Your baby's prognosis will depend on the size and location of the Encephalocele, the type of brain tissue involved, the ease of surgical correction, and the presence of other associated abnormalities. Approximately one-half of the babies born with Encephaloceles survive, and approximately 75% of these babies have mental retardation. Babies with Anterior Encephaloceles and no associated abnormalities have the best prognosis. Babies with Posterior Encephaloceles and other abnormalities have less favorably prognoses.

Fact Sheet by:

Birth Defect Research Children, Inc.
www.birthdefects.org