Encephalocele is one of a group of birth defects known as neural tube defects (NTD). It occurs very early in fetal life where the neural tube does not close properly resulting in brain tissue protruding from the skull into a sac. Encephalocele occurs in about 1 to 2 of every 10,000 births in the United States. Of those babies, only 20% of them are born alive, and only half of those survive. For an unknown reason, 70% of occipital (back of the skull) encephaloceles occur in females, whereas anterior (front of the skull) encephaloceles are more often seen in males.

Associated Conditions

Up to 40% of encephalocele cases occur in babies who have chromosomal abnormalities. The brains of children with encephalocele do not form normally, which may result in developmental delay, learning problems, or mental retardation.

Causes & Risk Factors

The causes for encephalocele are unknown. However, it has been shown that genetic and environmental risk factors play an important role. Mothers with the following characteristics have been found to be at higher risk for having a baby with encephalocele:

- Maternal lack of folic acid and vitamins
- Previous baby with NTD (1 in 25)
- Family history of NTD
- Maternal obesity
- Diabetes during pregnancy
- Maternal stress
- Woman who has epileptic seizures

Prevention

There are a number of steps a woman can take to reduce her risk of having a baby with a birth defect. Please see CBDMP’s fact sheet on Reducing the Risk of Birth Defects.

Preconception & Prenatal Care

Folic acid is a B vitamin that plays an important role in the development of the fetal brain and spinal cord during very early pregnancy, often before many women know they are pregnant. For this reason and because many pregnancies are unplanned, it is recommended that all women of childbearing age take 400 (0.4mg) micrograms of folic acid per day through dietary supplements of foods that are fortified with folate. It has been shown that taking the recommended dose can reduce the risk of neural tube defects by up to 70%. Encephalocele may be detected during the middle of the second trimester of pregnancy using high-level ultrasound examination. It may not be detected by prenatal blood testing because alphafetoprotein levels are not typically elevated when the defect is covered by skin. If an encephalocele is detected, a specialist that handles high-risk pregnancies should be consulted. Women who think that they may be in a high risk category should have genetic counseling. For more information, please visit the California Prenatal Screening Program website.

Medical Care

Treatment may include immediate surgery to remove the sac and replace the brain tissue into the skull. A computerized tomogram (CT) scan or other studies may be done to determine the extent of the lesion. Long-term treatment depends on the extent of the defect. The fetus should be delivered at a facility that has personnel capable of handling all aspects of neonatal complications.

Infection (meningitis) is a post-operative concern. Because the vision center is located in the back of the brain, some babies who survive may be blind.

Long Term Outcomes

Multi-stage surgery may be needed, depending on location of the encephalocele and structures involved. Survival for children with encephalocele is in the range of 50 - 95%. At least half of survivors will grow up to have significant developmental delays. Early intervention programs are available to assist families to maximize the child’s development and focus on the child’s strengths.
Resources For Families

California Children's Services (CCS)

California Children's Services (CCS) is a state program for children with certain diseases or health problems. Through this program, children up to 21 years old can get the health care and services they need. CCS will connect you with doctors and trained health care people who know how to care for your child with special health care needs. (Service eligibility is income-based, but you may be eligible for services if the health care costs exceed 20% of your income.)

Find your local CCS Office –
• In the phone book under California Children's Services or County Health Department
• Or online at: www.dhs.ca.gov/ccs

Family Resource Centers Networks of California

Their mission is to support families of children with disabilities, special healthcare needs, and those at risk by ensuring the continuance, expansion, promotion and quality of family-centered, parent-directed, family resource centers.

Find your local Family Resource Center –
• In the phone book business pages under Social and Human Services for Families and Individuals or Family Resource Centers
• Or online at: http://www.frcnca.org/

California Healthy Families Program:

Healthy Families is low cost insurance for children and teens. It provides health, dental and vision coverage to children who do not have insurance and do not qualify for free Medi-Cal.

• Phone: 1-800-880-5305 if you do not already have a child enrolled in Healthy Families.
• E-mail: HealthyFamilies@MAXIMUS.com
  Be sure to include your name and phone number.
• In person: Many community organizations have people who are trained to help you apply. Call to find a Certified Application Assistants, or CAAs in your area: 1-888-747-1222. The call is free.
• Website: www.healthyfamilies.ca.gov

Birth Defects Research for Children, Inc.

Their mission is to support families of children with disabilities, special healthcare needs, and those at risk by ensuring the continuance, expansion, promotion and quality of family-centered, parent-directed, family resource centers.

Find your local Family Resource Center –
• In the phone book business pages under Social and Human Services for Families and Individuals or Family Resource Centers
• Or online at: http://www.frcnca.org/

Birth Defects Research for Children is a resource for free birth defect information, parent networking and birth defect research through the National Birth Defect Registry.

• Birth Defect Research for Children, Inc.
  800 Celebration Avenue, Suite 225
  Celebration, FL 34747
• Phone: 407-566-8304
• Fax: 407-566-8341
• Website: http://www.birthdefects.org/

Other Information & Support Groups

• Regional Centers in California: nonprofit private corporations that contract with the Department of Developmental Services to coordinate services and provide support for individuals with developmental disabilities. www.dds.ca.gov/RC/Home.cfm
• March of Dimes Birth Defects Foundation: 1-888-422-2437 www.marchofdimes.com
• National Center for Birth Defects and Developmental Disabilities: 1-770-488-7160 www.cdc.gov/ncbddd
• California Prenatal Screening Program: http://www.cdph.ca.gov/programs/pns/Pages/default.aspx

References