

Cephalocele

Patient Information Series – What you should know, what you should ask.

What is Cephalocele?

Cephalocele is a rare birth defect characterized by a protrusion of brain tissue and its surrounding membranes through an opening in the skull. This occurs when the neural tube, which forms the brain and spinal cord, does not close completely during early fetal development.

What Causes Cephalocele?

The exact cause of cephalocele is not fully understood but is related to a combination of genetic and environmental factors that affect neural tube development in the first days of fetal development. Factors that may increase the risk include certain medications, nutritional deficiencies (such as folic acid), and genetic mutations.

Should I Have More Tests Done?

After an initial diagnosis through routine ultrasound, further testing is often recommended to assess the condition's extent and determine if other anomalies are present. You can consult with your caregiver and/or a maternal-fetal medicine specialist to decide whether terminating the pregnancy is the right choice for you and your family. You may be referred to a maternal-fetal medicine or other specialist for detailed ultrasound of the fetal brain and central nervous system, as well as fetal MRI for detailed imaging of the fetus. Genetic counseling and testing, such as amniocentesis or chorionic villus sampling (CVS) with chromosomal microarray or whole exome sequencing, can help determine if the cephalocele is part of a genetic syndrome.

What Are the Things to Watch for During My Pregnancy?

Throughout your pregnancy, regular medical check-ups are important to monitor the health of both you and your baby. Your healthcare provider may suggest more frequent ultrasounds or other monitoring techniques to watch for complications such as hydrocephalus (excess fluid in the brain), which can accompany cephalocele.

What Does It Mean for My Baby After It is Born?

After birth, the treatment for cephalocele typically involves surgery to repair the skull defect and reposition any protruded brain tissue. Each baby is unique. The prognosis for your baby depends on the size and location of the cephalocele, whether other brain structures are involved, or other organ systems affected. Early intervention and rehabilitation services can help address developmental delays or motor impairments.

Will It Happen Again?

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If you have had a child with a cephalocele, the risk of this condition occurring in a future pregnancy may be slightly increased. Discuss with your genetic counselor or specialist to understand your specific risk and possible preventive measures, such as increasing folic acid intake before conception.

What other questions should I ask?

- How will the cephalocele affect my baby's development and quality of life?
- What specialists will be involved in my baby's care after birth?
- Are there any specific symptoms or signs I should report immediately during my pregnancy?
- What support services are available for families of children with special needs?

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