Holoprosencephaly

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

What is Holoprosencephaly?

Holoprosencephaly is a complex brain malformation resulting from incomplete cleavage of the brain during early fetal development. This condition is characterized by the failure of the prosencephalon (the forebrain of the embryo) to properly divide into the double lobes of the cerebral hemispheres, leading to a spectrum of abnormalities affecting the brain and facial features.
Holoprosencephaly is classified into four types, based on the severity of the brain's division:
Alobar Holoprosencephaly: The most severe form, where there is no separation of the cerebral hemispheres, resulting in a single cerebral ventricle and a single lobed brain.
Semi-lobar Holoprosencephaly: The cerebral hemispheres are partially separated, and the brain's structure is somewhat intermediate between alobar and lobar.
Lobar Holoprosencephaly: The least severe form, with better separation of the cerebral hemispheres and more normal brain structure.
Middle Interhemispheric Variant (Syntelencephaly): The hemispheres are not separated in the middle of the brain but may be more normally divided in the anterior and posterior aspects.

What causes Holoprosencephaly?

The causes of holoprosencephaly are diverse and can include genetic abnormalities, environmental factors, and complications during pregnancy. Some cases are linked to chromosomal disorders, such as trisomy 13, while others may be associated with mutations in specific genes. Environmental factors that increase the risk include maternal diabetes, certain medications, and infections during pregnancy.

Should I have more tests done?

You might be referred for specialist neurosonography (ultrasound of the fetal brain and central nervous system) and magnetic resonance imaging (MRI). Further targeted ultrasound may be ordered to determine if other anomalies are present. You might consult with your caregiver and/or a maternal-fetal medicine specialist to determine whether continuing the pregnancy is the right choice for you and your family. Genetic counseling and testing could be recommended. This may include tests like amniocentesis or chorionic villus sampling (CVS) to check for chromosomal abnormalities and other genetic diagnostic tests such as chromosomal microarray testing or whole exome sequencing.

What are the things to watch for during my pregnancy?

In continuing pregnancies affected by Holoprosencephaly, you should attend your regular prenatal care and have regular ultrasounds to assess the baby's well-being and development. Your caregiver will advise you as your pregnancy progresses, for example, pay attention to your baby's movements. Significant changes in the pattern or frequency might need to be evaluated.

What does it mean for my baby after it is born?

The symptoms and physical manifestations of holoprosencephaly vary widely, depending on the severity of the condition. Some babies with holoprosencephaly may have severe facial abnormalities, such as closely spaced eyes (hypotelorism), a single eye (cyclopia), a flat nose or no nose, and cleft lip or palate. There may be neurological Issues, including intellectual disability, developmental delay, seizures, and problems with movement and coordination. Endocrine disorders related to the hypothalamus and pituitary gland, affecting temperature regulation, growth, and metabolism, may be present. You may be referred for consultation with a multidisciplinary team including pediatric neurology, craniofacial surgery, endocrinology, to discuss the implications of your baby's condition. The prognosis for holoprosencephaly varies greatly. In severe cases, the prognosis is very poor, with many infants not surviving beyond the first six months. In milder cases, children can survive into adulthood but may have various neurological and developmental challenges.

Will it happen again?

Each pregnancy is different, and the likelihood of a recurrence of holoprosencephaly can vary greatly for different families. Genetic counseling can provide more personalized risk assessment and information based on your family's history, the potential involvement of genetic factors, and any known environmental influences. This assessment might include discussions about genetic testing for the parents and potentially the affected child to identify specific mutations or chromosomal abnormalities.

What other questions should I ask?

- What are the long-term implications of holoprosencephaly for my child's health, development, and quality of life?
- Are there any additional tests or evaluations that need to be performed to assess my baby's condition and prognosis further?
- What are my options regarding continuing the pregnancy?
- Where should I deliver my baby?
- Can you tell me about available support for my baby after birth?
- Can you provide information about the medical team and specialists who will be involved in my delivery and the baby's care after birth?

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