

Holoprosencephaly

What is it?

Holoprosencephaly is a birth defect of the brain where the brain doesn't divide into right and left sections, called hemispheres. This can affect the head and face. There are three main types of holoprosencephaly:

- **Alobar:** Complete lack of division into right and left hemispheres of the brain. Babies with this type may also have a single eye, absent eyes, a very small eye, a flattened nose, and/or a cleft lip. This is the most severe type.
- **Semi-lobar:** The left side of the brain is fused to the right side at the front and sides of the brain. Babies with this type can also have small eyes, one eye, a flattened bridge and tip of the nose, one nostril, a cleft lip, and/or a cleft palate.
- **Lobar:** There is a right ventricle and a left ventricle, but the brain hemispheres are fused at the front. Babies also might have a cleft lip, closely spaced eyes, a depressed nose, or an almost normal-looking face.

Babies with any type of holoprosencephaly also may have seizures, water on the brain, neural tube defects, pituitary dysfunction, short height, feeding problems, developmental delay, and intellectual disability.

How common is it?

About 1 in every 16,000 babies is born with holoprosencephaly each year. It is more common in stillbirths and miscarriages.

What causes it?

The cause of holoprosencephaly is unknown. There may be many factors that cause it. More research is needed to understand the exact cause.

How is it diagnosed?

To confirm holoprosencephaly, imaging of the brain is done. During pregnancy, imaging can be done to look for birth defects and other conditions. After birth, imaging can be done to see the brain, too.

How is it treated?

Treatment for babies with this condition varies based on the type of holoprosencephaly. Your child's doctor will discuss appropriate treatment options with you.

For more information:

National Institutes of Health, Genetic and Rare Diseases
<https://rarediseases.info.nih.gov/diseases/6665/holoprosencephaly#diseaseDiagnosisSection>

National Human Genome Research Institute
<https://www.genome.gov/12512735/learning-about-holoprosencephaly/>