Holoprosencephaly

BIRTH DEFECT RESEARCH FOR CHILDREN

What is holoprosencephaly (HPE)?

Holoprosencephaly is a serious brain defect that occurs during the first few weeks after conception. In HPE, the forebrain of the developing embryo does not divide to form the right and left sides of the brain. This results in defects in the development of the brain structure and function and often facial defects as well.

Holoprosencephaly

How is holoprosencephaly classified?

Lobar (mild) - The least frequent form of HPE. The brain is divided but there is some fusion of structures and mild abnormalities.

Semilobar (moderate) - The brain is partially divided. There are two hemispheres in the rear, but not the front of the brain. There are some moderate abnormalities with this form of HPE. One fourth of the babies with HPE have the semilobar form.

Alobar is the most serious form. The brain is not divided and there are severe abnormalities including facial abnormalities. Almost two-thirds of babies with HPE have the alobar form.

Children with HPE may also have other abnormalities including microcephaly (a small head), hydrocephalus (excessive fluid in the brain), seizures, endocrine abnormalities, or malformations of other organ systems like the heart, skeleton, genitourinary and gastrointestinal systems.

Many children with HPE have facial abnormalities including a nose that is flat with only one nostril, close-set eyes, cleft lip and/or palate or just a single upper middle tooth. More severe facial defects may include a single eye located in the middle of the face, a nose located on the forehead or missing facial features.

What causes HPE?

Although the cause of holoprosencephaly is not known, it has been linked to both genetic alterations and environmental exposures. Some environmental risk factors include maternal diabetes, infections during pregnancy (herpes, rubella, toxoplasmosis, syphillis, cytomegalovirus) and various drugs taken during pregnancy including alcohol, aspirin, lithium, thorazine, anticonvulsants, hormones, retinoic acid, and possibly others.

In some families, there is evidence that HPE is genetically transmitted through autosomal dominant, autosomal recessive or X-linked inheritance. In some children with HPE, specific chromosomal abnormalities have been identified.

How common is HPE?

Between 1 in 5,000-10,000 babies are born with HPE each year. Since only about 3% of fetuses with HPE survive till birth, the actual frequency may be as high as 1 in 200-250.

What are the risks of reoccurrence?

The risk of reoccurrence is small in most families, but genetic counseling is recommended.

What is the prognosis for a child with HPE?

Babies with the most severe forms of holoprosencephaly often die before they are born or shortly after birth. However some children even with the most severe forms of HPE may live several months or years after birth. Children with the mildest forms of HPE may have a normal life span.

Fact Sheet by:

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