Holoprosencephaly

Holoprosencephaly is a disorder caused by the failure of the forebrain of the embryo (*prosencephalon*) to divide to form bilateral cerebral hemispheres. This causes defects in:

- Development of the face.
- Brain structure and function.

This disorder used to be called arhinencephaly. It consists of a spectrum of defects or malformations of the brain and face.

- Most cases are associated with severe malformations of the brain. These are incompatible with life. They often cause spontaneous intrauterine death.
- At the other end of the spectrum are individuals with normal or near-normal brain development and facial defects. These defects may affect the:
  - Eyes.
  - Nose.
  - Upper lip.

Seizures and mental retardation may occur.

Facial Abnormalities include:

- The most severe of the facial anomalies is cyclopia. This is an abnormality characterized by:
  - A single eye. It is located in the area normally occupied by the root of the nose.
  - A missing nose or a nose in the form of a tubular appendage (*proboscis*). It is located above the eye.
- Ethmocephaly is the least common facial anomaly. It consists of:
  - A proboscis separating narrow-set eyes with an absent nose.
  - Abnormal smallness of one or both eyes (*microphthalmia*).
- Cebocephaly is another facial anomaly. It is characterized by a small, flattened nose with a single nostril.
  - The nose is located below incomplete or underdeveloped closely set eyes.
- The least severe in the spectrum of facial anomalies is the median cleft lip. It is also called premaxillary absence (*agenesis*).

There are three classifications of holoprosencephaly.

- Alobar is the most serious form. In this form, the brain has no tendency to separate. It is usually associated with severe facial anomalies.
- Semilobar is an intermediate form of the disease. In this form, the brain's hemispheres have a slight tendency to separate.
- Lobar is the least severe form. With this, there is considerable evidence of separate brain hemispheres.
  - In some cases of lobar holoprosencephaly the patient's brain may be nearly normal.

TREATMENT

There is no standard course of treatment for this disorder. Treatment is symptomatic and supportive.

The likely outcome (*prognosis*) for individuals with the disorder is poor. Most of those who survive show no significant developmental gains.

RESEARCH BEING DONE

The NINDS supports and conducts a wide range of studies which focus on identifying and learning more about the genes and other factors involved in normal brain development. The knowledge gained from these fundamental studies provides the foundation for understanding how this process can go awry. It may eventually give clues to understanding disorders such as holoprosencephaly.

Research is continually being done to learn more about this problem.

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