Holoprosencephaly

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Holoprosencephaly Information Page.

Holoprosencephaly is a disorder caused by the failure of the *prosencephalon* (the embryonic forebrain) to sufficiently divide into the double lobes of the cerebral hemispheres. The result is a single-lobed brain structure and severe skull and facial defects. In most cases of holoprosencephaly, the malformations are so severe that babies die before birth. In less severe cases, babies are born with normal or near-normal brain development and facial deformities that may affect the eyes, nose, and upper lip.

There are three classifications of holoprosencephaly. Alobar, in which the brain has not divided at all, is usually associated with severe facial deformities. *Semilobar*, in which the brain's hemispheres have somewhat divided, causes an intermediate form of the disorder. *Lobar*, in which there is considerable evidence of separate brain hemispheres, is the least severe form. In some cases of lobar holoprosencephaly the baby's brain may be nearly normal.

The least severe of the facial anomalies is the median cleft lip (*premaxillary agenesis*). The most severe is *cyclopia*, an abnormality characterized by a single eye located in the area normally occupied by the root of the nose, and a missing nose or a proboscis (a tubular-shaped nose) located above the eye. The least common facial anomaly is *ethmocephaly*, in which a proboscis separates closely-set eyes. *Cebocephaly*, another facial anomaly, is characterized by a small, flattened nose with a single nostril situated below incomplete or underdeveloped closely-set eyes.