## **Condition:** Holoprosencephaly

---

### Inheritance:

Genetically heterogeneous, includes chromosomal and single gene causes.

### Genetic etiology:

Holoprosencephaly can result from a wide variety of genetic causes. It is a common component of trisomy 13 and many other syndromes. Nonsyndrome holoprosencephaly can result from mutation in any of several different genes, including *SHH*, *SIX3*, *TGIF,PTCH or ZIC2. Nonsyndromic holoprosencephaly tends to be dominantly transmitted.*

### Frequency:

Rare

### Clinical features:

Holoprosencephaly is a malformation of the central nervous system in which there is incomplete or absent separation of the cortex of the brain into two hemispheres. In its mildest form, it can manifest as absence of the olfactory bulbs and missing central maxillary incisors. In its severe form it is a severe brain malformation that is incompatible with normal development.

### Management:

Supportive care.

### Genetic counseling:

Depends on mechanism (e.g., chromosomal, single gene, sporadic). Genetic testing for several of the causative genes is available.

*Human Genetics and Genomics*, Fourth Edition. Bruce R. Korf and Mira B. Irons.