

CHARGE Syndrome

Characteristics

- **Coloboma**
 - Unilateral or bilateral coloboma of the iris, retina-choroid, and/or disc with or without microphthalmos (80%-90% of individuals)
- **Heart Defects (75-85%)**
 - Almost every type of congenital heart defect has been described including VSD, ASD, PDA
 - Many have multiple and complex defects: aortic arch defects, shunts, AV canal and others. Cardiology of CHARGE
- **Atresia of the choanae**
 - Unilateral or bilateral choanal atresia or stenosis (50%-60%)
- **Retarded growth and development**
 - Growth deficiency (70%-80%)
 - Developmental Delay
- **Genital abnormalities**
 - Cryptorchidism in males and hypogonadotrophic hypogonadism in both males and females
- **Ear anomalies**
 - Abnormal outer ears, ossicular malformations, Mondini defect of the cochlea and absent or hypoplastic semicircular canals (>90%)
 - impaired hearing and/or swallowing problems (70-80%)
- **Other problems**
 - Cranial nerve dysfunction resulting in hyposmia or anosmia, unilateral or bilateral facial palsy (40%), swallowing problems (70-90%)
 - Orofacial clefts (15%-20%)
 - Tracheoesophageal fistula (15%-20%)

Major Characteristics: More specific for CHARGE

- Ocular Coloboma
- Choanal atresia or stenosis
- Hearing loss
- Cranial nerve abnormalities

Minor Characteristics: Less specific for CHARGE, more important if many of these present

- Heart Defects
- Growth and Developmental Delay

- Cleft Lip and or Palate
- Genital abnormalities
- Tracheoesophageal fistula

Incidence

- Estimated to occur in 1:8,500 to 1:10,000 people

Genetics

- Inheritance Pattern
 - Autosomal Dominant, most with new mutation
- Genes
 - CHD7 found to cause more than 50% of cases
 - CHD7 gene product regulates gene expression by chromatin remodeling.
 - CHD7 mutations can have varied and widespread effects during embryonic development

Ocular Considerations

- Retinal Detachment Risk: 8% in a study by Keech and others-1997
- Vision may be affected or not depending upon the position and extent of the coloboma
- Trial of amblyopia treatment may be warranted
- Cataract may accompany coloboma

Resources

[NLM Genetics Home Reference](#)
[GeneReviews](#)
[GeneTests Labs for Gene Testing](#)
[Cardiology of CHARGE](#)

[syndrome](#)

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