Condition: CHARGE syndrome

Inheritance:

Usually sporadic, though autosomal dominant transmission can occur.

Genetic etiology:

Mutation in *CHD7* gene encoding chromodomain-helicase-DNA binding protein 7. Most mutations result in haploinsufficiency.

Frequency:

Approximately 1/10,000 births.

Clinical features:

CHARGE was coined as an acronym for the major features. These include choanal atresia, heart defects, esophageal atresia or tracheoesophageal fistula, facial palsy, hearing loss, colobomata, genitourinary anomalies, growth retardation, and developmental delay.

Management:

For non-lethal defects, the lesion is closed by surgery. Supportive care is then provided for neurological deficits and orthopedic problems.

Genetic counseling:

Most cases are sporadic, with low recurrence risk, though germline mosaicism is possible; genetic testing is available.

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