

Diagnostic criteria for CHARGE syndrome

Blake's diagnostic criteria (1998)

(Blake K.D. et al., 1998; Clin Pediatr (Phila), 37(3):159-73)

Major criteria (4 C's)

Coloboma -of iris, retina, choroid, disc; microphthalmia

Choanal atresia

Cranial nerve (especially VII and VIII) dysfunction

Characteristic ear abnormalities

Minor criteria

Genital hypoplasia

Developmental delay

Cardiovascular malformations

Growth deficiency

Orofacial cleft

Tracheoesophageal fistula

Characteristic face

Occasional criteria

Renal anomalies

Hand anomalies

Abdominal defects

Spine anomalies

Neck/shoulder anomalies

Thymic/parathyroid hypoplasia

Diagnostic criteria interpretation

Definite CHARGE: 4 major or 3 major and 3 minor criteria

Probable/possible CHARGE: 1 or 2 major and several minor criteria

Verloes' diagnostic criteria (2005)

(Verloes A. 2005; Am J Med Genet A. 15;133A(3):306-8)

Major criteria (3 C's)

Coloboma (iris or choroid, with or without microphthalmia)

Choanal atresia

Hypoplastic semi-circular Canals

Minor criteria

Rhombencephalic dysfunction (brainstem dysfunctions, cranial nerve VII to XII palsies and neurosensory deafness)

Hypothalamo-hypophyseal dysfunction (including GH and gonadotrophin deficiencies)

Abnormal middle or external ear

Malformation of mediastinal organs (heart, esophagus)

Mental retardation

Diagnostic criteria interpretation

Typical CHARGE: 3 major, or 2 major and 2 minor criteria

Partial/incomplete CHARGE: 2 major and 1 minor criteria

Atypical CHARGE: 2 major, or 1 major and 3 minor criteria