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)

**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