

Diagnostic criteria for Gorlin syndrome (or nevoid basal cell carcinoma syndrome, NBCCS)

(Veenstra-Knol H. E. et al., Eur. J. Pediatr. 164(3):126–30, 2005)

Presence of two major criteria or one major and two minor criteria

Major criteria

- Multiple (>2) basal cell carcinomas, or one diagnosed under 20 years
- Odontogenic keratocysts proven by histology
- ≥3 palmar or plantar pits
- Ectopic calcification (lamellar or early falx)
- Bifid, fused or markedly splayed ribs
- Family history of NBCCS

Minor criteria

- Macrocephaly determined after adjustment for height (≥97th percentile)
- Bridging of the sella turcica, vertebral anomalies such as hemivertebrae, fusion or elongation of the vertebral bodies, modeling defects of the hands and feet
- Cleft lip or palate, polydactyly
- Cardiac or ovarian fibroma
- Medulloblastoma
- Other skeletal abnormalities: Sprengel deformity, marked pectus deformity, marked syndactyly of the digits
- Eye anomaly: cataract, coloboma, microphthalmia
- *PTCH* mutation