Ruvalcaba syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ruvalcaba syndrome. ORPHA:3121

Ruvalcaba syndrome is an extremely rare malformation syndrome, described in less than 10 patients to date, characterized by microcephaly with characteristic facies (downslanting parpebral fissures, microstomia, beaked nose, narrow maxilla), very short stature, narrow thoracic cage with pectus carinatum, hypoplastic genitalia and skeletal anomalies (i.e. characteristic brachydactyly and osteochondritis of the spine) as well as intellectual and developmental delay.