

Open Peer Review on Qeios

Ramon syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Ramon</u> <u>syndrome</u>. ORPHA:3019

A rare, genetic, primary bone dysplasia syndrome characterized by bilateral, painless swelling of the face extending from the mandible to the inferior orbital margins (cherubism), epilepsy, gingival fibromatosis (possibly obscuring teeth), and intellectual disability. Other associated variable features include hypertrichosis, stunted growth, juvenile rheumatoid arthritis, and development of ocular abnormalities (e.g. pigmentary retinopathy, optic disc pallor, Axenfeld anomaly). Radiological images typically show bilateral multifocal radiolucency involving the body, angle and ramus of the mandible and coronoid process.

Qeios ID: NNEAK0 · https://doi.org/10.32388/NNEAK0