Roifman syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Roifman syndrome. ORPHA:353298

Roifman syndrome is a rare, genetic immuno-osseous dysplasia disorder characterized by pre- and post-natal growth retardation, hypotonia, borderline to moderate intellectual disability, retinal dystrophy, spondyloepiphyseal dysplasia (epiphyseal dysplasia, epiphyses ossification delay, vertebral changes) and skeletal anomalies (brachydactyly, fifth finger clinodactyly), as well as humeral immunodeficiency characterized by inability to generate specific antibodies and low circulating B-cells. Craniofacial dysmorphism, that typically inlcudes microcephaly, hypertelorism, long palpebral fissures, prominent eyelashes, a narrow, tubular, upturned nose with hypoplastic alae nasi, long philtrum and thin upper lip, are also associated.