Open Peer Review on Qeios

Chondroectodermal dysplasia with night blindness

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Chondroectodermal dysplasia with night blindness. ORPHA:319195

Chondroectodermal dysplasia with night blindness is a rare genetic bone development disorder characterized by proportionate short stature, nail dysplasia (enlarged, convex, hypertrophic nails), hypodontia and night blindness. Osteopenia, a tendency to present fractures, talipes varus with abnormal gait, ear infections, and watering eyes due to narrow tear ducts are frequently associated. Radiologically patients present delayed bone age on wrist X-rays, platyspondyly, and broad metaphyses of humeri with dense and thickened growth plates.