Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome. ORPHA:2653

Osteochondrodysplatic nanism-deafness-retinitis pigmentosa syndrome is characterized by severe dwarfism, progressive scoliosis and bilateral dislocation of the hip, associated with sensorineural deafness and retinitis pigmentosa. Radiographs show diffuse osteoporosis, severe bone-age delay and dysplasia of the femoral head. It has been described in two patients. Transmission is autosomal dominant variable penetrance.