Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Osteochondrodysplatic dwarfism-deafness-retinitis pigmentosa syndrome Karłowatość osteochondrodysplastyczna - głuchota - zwyrodnienie barwnikowe siatkówki Osteochondrodysplatic dwarfism-hearing loss-retinitis pigmentosa syndrome Osteochondrodysplatic nanism-hearing loss-retinitis pigmentosa syndrome

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
2653	-	Q87.1

Kod ICD11
LD2H.Y

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*Źródło

orphanet