Opis choroby *

Definicja

Olivopontocerebellar atrophy-deafness syndrome is characterised by infancy-onset olivopontocerebellar atrophy, sensorineural deafness and speech impairment. It has been described in less than 15 children. Most cases were sporadic, but autosomal recessive inheritance was suggested in three cases.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Olivopontocerebellar atrophy-hearing loss syndrome

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
2732	-	Q04.8

Kod ICD11
LD2H.Y

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*Źródło

orphanet