Opis choroby *

Definicja

A rare mitochondrial disease characterized by a highly variable phenotypic spectrum comprising delayed motor development, peripheral neuropathy, cataract, short stature due to growth hormone deficiency, nystagmus, sensorineural hearing loss, dysmorphic facial features, and skeletal abnormalities consistent with spondyloepimetaphyseal dysplasia. Hyperextensible joints, achalasia, and telangiectasia have also been described. Cognition is normal. Atrophy of the pituitary gland has been observed in brain imaging.

Dane

Klasyfikacja
Choroba

Synonimy
CAGSSS
CAGSSS

Kod ORPHA
436174

Kod OMIM
616007

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet