Opis choroby *

Definicja

Duane anomaly-myopathy-scoliosis syndrome is characterised by the association of bilateral Duane anomaly type 3, severe scoliosis of early onset, congenital myopathy with hypotonia without muscular weakness, delayed motor development, and short stature. It has been described in one pair of sibs. The Duane type 3 anomaly consists of eye abduction and adduction palsy, globe retraction and narrowing of the palpebral fissure. Muscular biopsy shows aspecific myopathy. Intellectual development is normal. The syndrome is most likely inherited in an autosomal recessive manner. It differs from the Crisfield-Dretakis-Sharpe syndrome, in which short stature and muscular features are absent. Surgery of the scoliosis is necessary. Functional prognosis depends on the severity of the visual handicap.

Dane

Klasyfikacja
Choroba

Synonimy
Verloes-Deprez syndrome
Zespół Verloesa i Depreza

Kod ORPHA
50817

Kod OMIM
-

Kod ICD10
Q87.5

Kod ICD11
-

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*Źródło

orphanet