Opis choroby *

Definicja

A rare arthrogryposis syndrome characterized by the association of arthrogryposis multiplex congenita and a severe form of motor neuron disease with loss of anterior horn cells in the spinal cord. Patients present with fetal akinesia deformation sequence with multiple contractures and facial anomalies, such as low-set ears, hypoplastic jaw, and short neck, as well as hypotonia and respiratory insufficiency. Some patients may survive into childhood and show developmental delay, markedly decreased muscle bulk, dystonic and involuntary movements, ataxia, and poor speech.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
AAHD
Choroba Vuopala
LAAHD
Vuopala disease

Kod ORPHA
53696

Kod OMIM
611890

Kod ICD10
Q68.8

Kod ICD11
LD2F.1Y

---

*Źródło

orphanet