

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 AAHD

Synonimy

Choroba Vuopala

LAAHD

Vuopala disease

Kod ORPHA

53696

Kod OMIM

611890

Kod ICD10

Q68.8

Kod ICD11

LD2F.1Y

*Źródło

orphanet