Opis choroby *

Definicja

Autosomal recessive myogenic arthrogryposis multiplex congenita is a rare inherited neuromuscular disease characterized by prenatal presentation (usually in the second trimester) of reduced fetal movements and abnormal positioning resulting in joint abnormalities that may involve both lower and upper extremities and is usually symmetric, severe hypotonia at birth with bilateral club foot, motor development delay, mild facial weakness without opthalmoplegia, absent deep tendon reflexes, normal motor and sensory nerve conduction velocities, no cerebellar or pyramidal involvement, and progressive disease course with loss of ambulation after the first decade of life.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive myogenic AMC
AMC zwiazana z SYNE1
Artrogrypoza związana z SYNE1
Autosomalna recesywna miogeniczna AMC
SYNE1-related AMC
SYNE1-related arthrogryposis multiplex congenita

Kod ORPHA
319332

Kod OMIM
618484

Kod ICD10
Q74.3

Kod ICD11
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*Źródło

orphanet