Opis choroby *

Definicja

A rare, systemic amyloidosis characterized by a triad of ophthalmologic, neurologic and dermatologic findings due to the deposition of gelsolin amyloid fibrils in these tissues. Clinical manifestations include corneal lattice dystrophy, cranial neuropathy, especially affecting the facial nerve, bulbar signs, cutis laxa, increased skin fragility, and less commonly peripheral neuropathy and renal failure.

Dane

Klasyfikacja
Choroba

Synonimy
Familial amyloid polyneuropathy type IV
Amyloidoza gelsolinowa
Dziedziczna amyloidoza, typ fiński
Rodzinna amyloidoza, typ fiński
Rodzinna polineuropatia amyloidowa typu 4
Familial amyloidosis, Finnish type
Gelsolin amyloidosis
Hereditary amyloidosis, Finnish type

Kod ORPHA
85448

Kod OMIM
105120

Kod ICD10
E85.1

Kod ICD11
5D00.2Y

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*Źródło

orphanet