Opis choroby *

Definicja

A rare genetic, multisystemic lysosomal disease characterized by specific cutaneous (angiokeratoma), neurological (pain), renal (proteinuria, chronic kidney failure), cardiovascular (cardiomyopathy, arrhythmia), cochleo-vestibular and cerebrovascular manifestations (transient ischemic attacks, strokes). The phenotypic expression depends on age of onset and, in females, the level of X-inactivation.

Dane

Klasyfikacja
Choroba

Synonimy
Alpha-galactosidase A deficiency
Choroba Andersona i Fabry'ego
FD
Niedobór alfa-galaktozydazy A
Rogowiec naczyniowy rozlany
Rozsiane zmiany o charakterze angiokeratoma
Anderson-Fabry disease
Angiokeratoma corporis diffusum
Diffuse angiokeratoma
FD

Kod ORPHA
324

Kod OMIM
301500

Kod ICD10
E75.2

Kod ICD11
5C56.01

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*Źródło

orphanet