Opis choroby *

Definicja

A rare, genetic, metabolic disease with connective tissue and eye involvement, characterized by progressive ectopic mineralization and fragmented elastic fibers in the skin, retina and vascular walls.

Dane

Klasyfikacja
Choroba

Synonimy
Gronblad-Strandberg-Touraine syndrome
PXE
Zespół Gronblada, Strandberga i Touraine'a
PXE

Kod ORPHA
758

Kod OMIM
264800

Kod ICD10
Q82.8

Kod ICD11
EC40

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*Źródło

orphanet