Opis choroby *

Definicja

A rare hereditary amyloidosis with primary renal involvement characterized by variable onset of renal insufficiency with edema, hypertension, proteinuria, and azotemia, eventually leading to end-stage renal disease. Amyloid cardiomyopathy and histopathological evidence of amyloid deposition in other organs, such as the spleen, liver, adrenal glands, and pancreas, among others, have also been described.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	Apolipoprotein A-II amyloidosis Amyloidoza apolipoproteinowa A-II Dziedziczna amyloidoza nerek z powodu wariantu apolipoproteiny A-ll Dziedziczna nefropatia amyloidowa z powodu wariantu polipoproteiny A-ll Rodzinna amyloidoza nerek z powodu wariantu apolipoproteiny A-ll Rodzinna nefropatia amyloidowa z powodu wariantu apolipoproteiny A-ll Familial amyloid nephropathy due to apolipoprotein A-II variant Familial renal amyloidosis due to apolipoprotein A-II variant Hereditary amyloid nephropathy due to apolipoprotein A-II variant Hereditary renal amyloidosis due to apolipoprotein A-II variant

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
238269	-	E85.0

Kod ICD11
5D00.2Y

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*Źródło

orphanet