Opis choroby *

Definicja

A rare, hereditary amyloidosis with primary renal involvement characterized by renal interstitial and medullary deposition of amyloid, low plasma levels of ApoA-1 and slow disease progression. Main clinical signs and symptoms are hypertension, proteinuria, hematuria and edema due to chronic renal insufficiency leading to end stage renal disease. Hepatosplenomegaly, progressive cardiomyopathy and involvement of skin, testes and adrenals (hypergonadotropic hypogonadism) have also been reported.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	Apolipoprotein A-I amyloidosis Amyloidoza związana z apolipoproteiną A - wariant I Dziedziczna amyloidoza nerek spowodowana wariantem I apolipoproteiny A Dziedziczna nefropatia amyloidowa spowodowana wariantem I apolipoproteiny A Rodzinna amyloidoza nerek spowodowana wariantem I apolipoproteiny A Rodzinna nefropatia amyloidowa spowodowana wariantem I apolipoproteiny A Familial amyloid nephropathy due to apolipoprotein A-I variant Familial renal amyloidosis due to apolipoprotein A-I variant Hereditary amyloid nephropathy due to apolipoprotein A-I variant Hereditary renal amyloidosis due to apolipoprotein A-I variant

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
93560	-	E85.0

Kod ICD11
5D00.2Y

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*Źródło

orphanet