Opis choroby *

Definicja

A rare lipoprotein metabolism disorder characterized biochemically by complete absence of apolipoprotein AI and extremely low plasma high density lipoprotein (HDL) cholesterol, and clinically by corneal opacities and xanthomas complicated with premature coronary heart disease (CHD).

Dane

Klasyfikacja
Choroba

Synonimy
ApoA-I deficiency
Hipoalfalipoproteinemia rodzinna
Niedobór ApoA-I
Rodzinny Niedobór apoA-I
Familial apoA-I deficiency
Familial hypoalphalipoproteinemia

Kod ORPHA
425

Kod OMIM
604091

Kod ICD10
E78.6

Kod ICD11
5C81.0

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*Źródło

orphanet