

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

*Źródło

orphanet