

Opis choroby *

Definicja

Sitosterolemia is a rare autosomal recessive sterol storage disease characterized by the accumulation of phytosterols in the blood and tissues. Clinical manifestations include xanthomas, arthralgia and premature atherosclerosis. Hematological manifestations include hemolytic anemia with stomatocytosis and macrothrombocytopenia. The disease is caused by homozygous or compound heterozygous mutations in *ABCG5* (2p21) and *ABCG8* (2p21) genes.

Dane

Klasyfikacja

Choroba

Synonimy

Phytosterolemia

Fitosterolemia

Ksantomatoza z sitosterolemią

Kod ORPHA

2882

Kod OMIM

618666

Kod ICD10

E78.0

Kod ICD11

5C52.1Y

*Źródło

orphanet