Sitosterolemia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Sitosterolemia. ORPHA:2882

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.