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Sitosterolemia

National Cancer Institute

Source

National Cancer Institute. *Sitosterolemia*. NCI Thesaurus. Code C125694.

An extremely rare autosomal recessive inherited disorder caused by mutations in the ABCG5 or ABCG8 genes. It is characterized by a defective sterolin transporter that impairs the elimination of plant sterols and, to a lesser degree, cholesterol from the body. These fatty substances build up in the tissues including arteries and skin, resulting in atherosclerosis and xanthomas.