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Pseudoxanthoma Elasticum

National Cancer Institute

Source

National Cancer Institute. *Pseudoxanthoma Elasticum*. NCI Thesaurus. Code C85036.

A rare, progressive, autosomal recessive inherited disorder caused by mutations in the ABCC6 gene. It is characterized by calcification and fragmentation of the elastic fibers of the skin, retina, and cardiovascular system. Signs and symptoms include skin plaques and bumps, thickened skin, retinal hemorrhage and obstruction of the blood vessels.