Loeys-Dietz Syndrome Type 2

National Cancer Institute

Source

National Cancer Institute. Loeys-Dietz Syndrome Type 2. NCI Thesaurus. Code C114768.

A rare autosomal dominant inherited disorder of connective tissue caused by mutations in either the TGFBR1 or TGFBR2 gene. Like Loeys-Dietz syndrome type I the disease is characterized by enlargement of the aorta and other arteries, and arterial tortuosity, but skeletal signs are typically less severe or absent in type 2. Skin abnormalities, such as velvety skin are often present in type 2.