Loeys-Dietz Syndrome

National Cancer Institute

Source

National Cancer Institute. Loeys-Dietz Syndrome. NCI Thesaurus. Code C75006.

A rare autosomal dominant syndrome caused by mutations in the TGFBR1 or TGFBR2 genes. It is characterized by vascular abnormalities including aortic and arterial aneurysms, aortic dissection, and tortuosity of the arteries. Other findings include scoliosis, long fingers, and joint hypermobility. Patients with TGFBR1 gene mutations also exhibit hypertelorism, bifid uvula, and early fusion of the skull bones.