Osteoporosis Pseudoglioma Syndrome

National Cancer Institute

Source

National Cancer Institute. Osteoporosis Pseudoglioma Syndrome. NCI Thesaurus. Code C130998.

An autosomal recessive condition caused by homozygous or compound heterozygous inactivating mutation(s) in the gene LRP5, encoding low-density lipoprotein receptor-related protein 5. This condition is characterized by severe juvenile-onset osteoporosis and congenital or juvenile-onset blindness due to a vascularized retinal mass that resembles a glioma.