Marshall Syndrome

National Cancer Institute

Source

National Cancer Institute. Marshall Syndrome. NCI Thesaurus. Code C128115.

An autosomal dominant condition caused by mutation(s) in the COL11A1 gene, encoding collagen alpha-1(XI) chain. The syndrome may be characterized by facial dysmorphism, cataracts, myopia, hearing loss, and short stature. Mutation(s) in the COL11A1 gene are causative in Stickler syndrome, but the phenotype of Marshall syndrome is more mild.