Open Peer Review on Qeios

Schimke Immunoosseous Dysplasia

National Cancer Institute

Source

National Cancer Institute. Schimke Immunoosseous Dysplasia. NCI Thesaurus. Code C135087.

An autosomal recessive condition caused by mutation(s) in the SMARCAL1 gene, encoding SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A-like protein 1. It is characterized by short stature, intrauterine growth restriction, microdontia, depressed nasal bridge, skeletal dysplasia, immune complex nephritis and immune deficiency.