Otopalatodigital Syndrome Type 1

National Cancer Institute

Source

National Cancer Institute. Otopalatodigital Syndrome Type 1. NCI Thesaurus. Code C118845.

A rare, X-linked dominant inherited syndrome caused by mutations in the FLNA gene. It is characterized by hearing loss caused by malformations in the ossicles, cleft palate, wide-set eyes, prominent brow ridges, small and flat nose, and skeletal abnormalities in the fingers and toes. Males usually experience more severe symptoms than females.