Treacher Collins Syndrome

National Cancer Institute

Source

National Cancer Institute. Treacher Collins Syndrome. NCI Thesaurus. Code C75018.

A rare autosomal dominant syndrome caused by mutations in the TCOF1 gene. Its characteristics include underdevelopment of the facial bones, small jaw and chin, absent or small ears, defects in the middle ear resulting in hearing loss, and downward sloping palpebral fissures.