Shprintzen-Goldberg Craniosynostosis Syndrome

National Cancer Institute

Source

National Cancer Institute. Shprintzen-Goldberg Craniosynostosis Syndrome. NCI Thesaurus. Code C124840.

A rare, autosomal dominant inherited syndrome often caused by mutations in the SKI gene. It is characterized by premature fusion of skull bones and distinctive facial features, including a long, narrow head, hypertelorism, exophthalmos, downslanting palpebral fissures, a high, narrow palate, micrognathia, and low-set ears. The bodies of affected individuals resemble those of people with Marfan syndrome.