Cranioectodermal Dysplasia

National Cancer Institute

Source

National Cancer Institute. Cranioectodermal Dysplasia. NCI Thesaurus. Code C129305.

An autosomal recessive disorder associated with mutation(s) in at least one of four genes (WDR35, IFT122, WDR19, or IFT43). It is characterized by distinctive abnormalities of the face and skull, in association with developmental abnormalities of the structures derived from ectodermal tissues.