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Hermansky-Pudlak Syndrome 1

National Cancer Institute

Source

National Cancer Institute. *Hermansky-Pudlak Syndrome 1*. NCI Thesaurus. Code C150367.

An autosomal recessive sub-type of Hermansky-Pudlak syndrome caused by mutation(s) in the HPS1 gene, encoding Hermansky-Pudlak syndrome 1 protein. This sub-type is associated with pulmonary fibrosis.