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Cartilage Hair Hypoplasia

National Cancer Institute

Source

National Cancer Institute. *Cartilage Hair Hypoplasia*. NCI Thesaurus. Code C61245.

A rare autosomal recessive inherited disorder caused by mutation in the gene for RNAase RMRP. It is characterized by short-limb dwarfism, presence of fine sparse hair, and T- and B-cell immunodeficiency.