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CHARGE Syndrome

National Cancer Institute

Source

National Cancer Institute. *CHARGE Syndrome*. NCI Thesaurus. Code C75100.

A rare autosomal dominant syndrome usually caused by mutations in the CHD7 gene. The term CHARGE is an acronym for the following unusual congenital abnormalities that are associated with this syndrome: coloboma of the eye, heart defects, choanal atresia, growth and developmental retardation, genital, and ear abnormalities.