Hypoparathyroidism-Retardation-Dysmorphism Syndrome

National Cancer Institute

Source

National Cancer Institute. Hypoparathyroidism-Retardation-Dysmorphism Syndrome. NCI Thesaurus. Code C133727.

An autosomal recessive condition caused by mutation(s) in the TBCE gene, encoding tubulin-specific chaperone E. It is characterized by congenital hypoparathyroidism, mental retardation, seizures and developmental delay.