Kenny-Caffey Syndrome Type 1

National Cancer Institute

Source

National Cancer Institute. Kenny-Caffey Syndrome Type 1. NCI Thesaurus. Code C130992.

An autosomal recessive form of Kenny-Caffey syndrome due to mutation(s) in the TBCE gene, encoding tubulin-specific chaperone E. This condition is characterized by hypoparathyroidism with hypocalcemia, marked growth retardation, craniofacial anomalies, absent diploic space in the skull, cortical thickening of long bones with medullary stenosis, and small hands and feet.