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Osteopetrosis with Renal Tubular Acidosis

National Cancer Institute

Source

National Cancer Institute. *Osteopetrosis with Renal Tubular Acidosis*. NCI Thesaurus. Code C118438.

A rare, autosomal recessive inherited disorder caused by mutation in the CA2 gene. It is characterized by osteopetrosis, renal tubular acidosis, and cerebral calcifications. It results in growth failure, mental retardation, and fractures.