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# Autosomal Recessive Osteopetrosis

National Cancer Institute

## Source

National Cancer Institute. *Autosomal Recessive Osteopetrosis*. NCI Thesaurus. Code C129733.

An autosomal recessive form of osteopetrosis caused by mutation(s) in at least 8 genes related to osteoclast function. This condition is characterized by the failure of osteoclasts to resorb bone, resulting in impaired bone modeling/remodeling, and skeletal fragility despite increased bone mass; it is also associated with hematopoietic insufficiency, hypocalcemia, disturbed tooth eruption, nerve entrapment syndromes, and growth impairment. Some cases are also associated with progressive neurological deterioration.