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Albright Hereditary Osteodystrophy with Multiple Hormone Resistance

National Cancer Institute

Source

National Cancer Institute. *Albright Hereditary Osteodystrophy with Multiple Hormone Resistance*. NCI Thesaurus. Code C129721.

Parathyroid hormone (PTH) resistance caused by heterozygous inactivating mutation(s) of the maternal allele of the GNAS gene encoding Gs-alpha, resulting in expression of PTH from only the paternal allele. Clinical manifestations include Albright hereditary osteodystrophy, early-onset obesity, and, in some cases, resistance to thyroid-stimulating hormone, gonadotropins, and growth hormone-releasing hormone, reflecting additional manifestations of Gs-alpha deficiency.