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Progressive Heterotopic Heteroplasia

National Cancer Institute

Source

National Cancer Institute. *Progressive Heterotopic Heteroplasia*. NCI Thesaurus. Code C131193.

Loss of function mutation(s) in the paternal allele of the GNAS gene, encoding guanine nucleotide-binding protein G(s) subunit alpha isoforms short resulting in dermal ossification beginning in infancy, followed by increasing and extensive heterotopic bone formation in deep muscle and fascia.