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Desbuquois Dysplasia

National Cancer Institute

Source

National Cancer Institute. *Desbuquois Dysplasia*. NCI Thesaurus. Code C124056.

A rare osteochondrodysplasia characterized by short stature, joint laxity, multiple dislocations, vertebral and metaphyseal abnormalities, and advanced carpotarsal ossification. Two forms have been identified: type 1 caused by mutation in the gene CANT1 and type 2 caused by mutations in the gene XYLT1.