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Upper limb mesomelic dysplasia

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base*. [Upper limb mesomelic dysplasia](#). ORPHA:2497

This syndrome is an isolated upper limb mesomelic dysplasia. It has been described in four patients from two unrelated families (a man and his daughter, and a Lebanese man and his son). Patients present with ulnar hypoplasia with severe radial bowing, but normal stature. The mode of transmission is likely to be autosomal dominant with variable expressivity.