

Open Peer Review on Qeios

Brachydactyly-arterial hypertension syndrome

INSFRM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

<u>Brachydactyly-arterial hypertension syndrome</u>. ORPHA:1276

Brachydactyly - arterial hypertension is a rare genetic brachydactyly syndrome characterized by the association of brachydactyly type E (see this term) with hypertension (due to vascular or neurovascular anomalies) as well as the additional features of short stature and low birth weight (compared to non-affected family members), stocky build and a round face. The onset of hypertension is often in childhood and, if untreated, most patients will have had a stroke by the age of 50.

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