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Radial hypoplasia-triphalangeal thumbshypospadias-maxillary diastema syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Radial</u> <u>hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome</u>. <i>ORPHA:2252

Radial hypoplasia-triphalangeal thumbs-hypospadias-maxillary diastema syndrome is characterised by symmetric, nonopposable triphalangeal thumbs and radial hypoplasia. It has been described in eight patients (five females and three males) spanning generations of a family. The affected males also presented with hypospadias. The syndrome is inherited as an autosomal dominant trait.