

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Schmitt-Gillenwater-Kelly syndrome Zespół Schmitta, Gillenwatera i Kelly'ego

Kod ORPHA	Kod OMIM	Kod ICD10
2252	179250	Q87.2

Kod ICD11

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*Źródło

orphanet