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Brachymorphism-onychodysplasia-dysphalangism syndrome

INSERM

Source

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

Brachymorphism-onychodysplasia-dysphalangism syndrome. ORPHA:1292

Brachymorphism-onychodysplasia-dysphalangism (BOD) is a very rare malformation syndrome that is characterized by short stature, hypoplastic fifth digits with tiny dysplastic nails, facial dysmorphism with coarse features including a wide mouth and broad nose, and mild intellectual disability. It has been suggested that Coffin-Siris syndrome (see this term) and BOD syndrome are perhaps allelic variants.