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Short stature-valvular heart disease-characteristic facies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Short stature-valvular heart disease-characteristic facies syndrome. ORPHA:2868

Short stature-valvular heart disease-characteristic facies syndrome is characterised by severe short stature with disproportionately short legs, small hands, clinodactyly, valvular heart disease and dysmorphism (ptosis, high-arched palate, abnormal dentition). It has been described in a mother and two daughters. This syndrome is probably transmitted as an autosomal dominant trait.