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Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome

INSERM

Source

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

Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome. ORPHA:1964

Extrasystoles-short stature-hyperpigmentation-microcephaly syndrome is a rare, genetic, malformation syndrome with short stature characterized by microcephaly, borderline intellectual disability, hyperpigmentation of the skin, short stature, and ventricular extrasystoles. Cardiac syncope may also be associated. There have been no further descriptions in the literature since 1975.