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Ataxia-photosensitivity-short stature syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ataxia-photosensitivity-short stature syndrome. ORPHA:1184

Ataxia-photosensitivity-short stature syndrome is a rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by cerebellar-like ataxia, photosensitivity (mainly of the face and trunk), short stature and intellectual disability. Additional features include clinodactyly, single palmar transverse crease, high-arched palate, pseudohypertrophy of the calves and aortic valve lesions. There have been no further descriptions in the literature since 1983.