CAMOS syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. CAMOS syndrome. ORPHA:83472

CAMOS syndrome is characterised by the association of a non-progressive congenital ataxia, severe intellectual deficit, optic atrophy and structural anomalies of the skin vessels. It has been described in five children from a large consanguineous Lebanese family. Short stature and microcephaly were also reported. Transmission is autosomal recessive.