Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome. ORPHA:86818

Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome is characterised by the association of Alport syndrome, midface hypoplasia, intellectual deficit and elliptocytosis. It has been described in two families. The syndrome is transmitted as an X-linked trait is caused by a contiguous gene deletion in Xq22.3 involving several genes including COL4A5, FACL4 and AMMECR1.