Hereditary bullous dystrophy, macular type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hereditary bullous dystrophy, macular type. ORPHA:1867

Bullous dystrophy, macular type is a genetic disorder characterised by formation of bullae without traumatic origin, alopecia, hyperpigmentation, acrocyanosis, short stature, microcephaly, intellectual deficit, tapering fingers and nail abnormalities. Two families (one of whom was Dutch and the other Italian) have been described up to now, in which only males were affected. Transmission is X-linked recessive. The bullous dystrophy locus has been mapped to Xq26.3 in the Italian family and to Xq27.3 in the Dutch family.