Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome. ORPHA:2057

Blepharophimosis-ptosis-esotropia-syndactyly-short stature syndrome is characterised by the association of blepharophimosis and ptosis, V-esotropia, and weakness of extraocular and frontal muscles with syndactyly of the toes, short stature, prognathism, and hypertrophy and fusion of the eyebrows. It has been described in six members of three related families. Transmission is autosomal recessive.