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Microphthalmia-ankyloblepharon-intellectual disability syndrome

INSERM

Source

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

Microphthalmia-ankyloblepharon-intellectual disability syndrome. ORPHA:85275

Microphthalmia-ankyloblepharon-intellectual disability syndrome is characterized by microphthalmia, ankyloblepharon and intellectual deficit. It has been described in seven male patients from two generations of a Northern Ireland family. The causative gene is localized to the Xq27-q28 region. The syndrome is transmitted as an X-linked recessive trait.