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Blepharonasofacial malformation syndrome

INSERM

Source

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

Blepharonasofacial malformation syndrome. ORPHA:1252

Blepharonasofacial syndrome is a rare otorhinolaryngological malformation syndrome characterized by a distinctive mask-like facial dysmorphism, lacrimal duct obstruction, extrapyramidal features, digital malformations and intellectual disability.