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Mandibulofacial dysostosis-microcephaly syndrome

INSERM

Source

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

Mandibulofacial dysostosis-microcephaly syndrome. ORPHA:79113

Mandibulofacial dysostosis-microcephaly syndrome is a rare genetic multiple malformation disorder characterized by malar and mandibular hypoplasia, microcephaly, ear malformations with associated conductive hearing loss, distinctive facial dysmorphism, developmental delay, and intellectual disability.