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# Upper limb defect-eye and ear abnormalities syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Upper limb defect-eye and ear abnormalities syndrome. ORPHA:2489*

Upper limb defect - eye and ear abnormalities syndrome associates upper limb defects (hypoplastic thumb with hypoplasia of the metacarpal bone and phalanges and delayed bone maturation), developmental delay, central hearing loss, unilateral poorly developed antihelix, bilateral choroid coloboma and growth retardation.