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Ear-patella-short stature syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ear-patella-short stature syndrome. ORPHA:2554

Ear-patella-short stature syndrome is an association of malformations including bilateral microtia (severe hypoplasia of ear pinnae), absent patellae, short stature, poor weight gain, and characteristic facial features such as high forehead, micrognathism with full lips and small mouth, and accentuated nasolabial folds (smile wrinkles linking the nostrils to the labial commissure).