

DISEASE:

Ear-patella-short stature syndrome

NAME:	Ear-patella-short stature syndrome
DESCRIPTION:	A rare microcephalic primordial dwarfism characterized by the association of bilateral microtia (severe hypoplasia of ear pinnae), absent patellae, short stature 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).
ORPHACODE:	2554
SYNONYMS:	Meier-Gorlin syndrome
XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM MeSH MedDRA ICD-10 OMIM

ANALYTE(S):	<u>ORC4</u> <u>ORC6</u> <u>CDT1</u> <u>CDC6</u> <u>ORC1</u> <u>GMNN</u> <u>CDC45</u>
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