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Congenital heart defect-round face-developmental delay syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Congenital heart defect-round face-developmental delay syndrome. ORPHA:1355*

Heart defect round face congenital developmental delay is very rare syndrome described in three sibs of one Japanese family and characterized by congenital heart disease, round face with depressed nasal bridge, small mouth, short stature, and relatively dark skin and typical dermatoglyphic anomalies, and intellectual deficit.