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Cleft palate-short stature-vertebral anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cleft palate-short stature-vertebral anomalies syndrome. ORPHA:2015

Cleft palate- short stature- vertebral anomalies is a multiple congenital anomalies syndrome described in a father and son characterized by the association of cleft palate, peculiar facies (asymmetrical appearance, inner epicanthal folds, short nose, anteverted nostrils, low and back-oriented ears, thin upper lip and micrognathism), short stature, short neck , vertebral anomalies and intellectual disability. The transmission is presumed to be autosomal dominant. There have been no further descriptions in the literature since 1993.