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Deafness-epiphyseal dysplasia-short stature syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Deafness-epiphyseal dysplasia-short stature syndrome. ORPHA:3218*

This syndrome is characterised by sensorineural deafness, short stature, femoral epiphyseal dysplasia, umbilical and inguinal hernias and developmental delay (growth retardation and mild intellectual deficit).