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Achondroplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Achondroplasia. ORPHA:15

Achondroplasia is the most common form of chondrodysplasia, characterized by rhizomelia, exaggerated lumbar lordosis, brachydactyly, and macrocephaly with frontal bossing and midface hypoplasia.