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Thoracic dysplasia-hydrocephalus syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Thoracic dysplasia-hydrocephalus syndrome. ORPHA:1861

Thoracic dysplasia-hydrocephalus syndrome is an extremely rare primary bone dysplasia syndrome characterized by short ribs with a narrow chest and thoracic dysplasia, mild rhizomelic shortening of the limbs, communicating hydrocephalus, and developmental delay. There have been no further descriptions in the literature since 1987.