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Spondylocostal dysostosis-hypospadias-intellectual disability syndrome

INSERM

Source

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

Spondylocostal dysostosis-hypospadias-intellectual disability syndrome. ORPHA:329252

Spondylocostal dysostosis-hypospadias-intellectual disability syndrome is a rare, genetic, bone developmental disorder characterized by generalized vertebral segmentation and fusion defects, disproportionate short stature (with predominant truncal shortness) and thoracolumbar scoliosis, associated with mild intellectual disability, hypospadias, partial cutaneous finger syndactyly and mild swan neck-like deformities of the fingers.