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Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome

INSERM

Source

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

Spondylocostal dysostosis-anal atresia-genitourinary malformation syndrome.

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Spondylocostal dysostosis-anal and genitourinary malformations syndrome is characterised by the association of spondylocostal dysostosis with anal and genitourinary malformations (anal atresia and agenesis of external and internal genitalia). To date, only four cases have been described in the literature. Autosomal recessive inheritance has been suggested.