Diaphanospondylodysostosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Diaphanospondylodysostosis. ORPHA:66637

Diaphanospondylodysostosis is characterized by absent ossification of the vertebral bodies and sacrum associated with variable anomalies. It has been described in less than ten patients from different families. Manifestations include a short neck, a short wide thorax, a reduced number of ribs, a narrow pelvis, and inconstant anomalies such as myelomeningocele, cystic kidneys with nephrogenic rests, and cleft palate.