Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome. ORPHA:397927

Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome is a rare, genetic, neural tube defect malformation syndrome characterized by sacral agenesis and abnormal vertebral body ossification with normal vertebral arches associated with notochord canal persistence on ultrasonography. Additional findings include bilateral clubfoot, oligohydramnios, single umbilical artery and, in some, increased nuchal translucency.