Sirenomelia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Sirenomelia. ORPHA:3169

Sirenomelia is a rare, genetic, developmental defect during embryogenesis disorder characterized by fusion of the lower limbs and associated with some degree of lower extremity reduction and persistent vitelline artery. Patients also present severe malformations of the musculoskeletal system (e.g. sacral agenesis), as well as the urogenital and lower gastrointestinal tracts (e.g. renal agenesis, absent bladder, rectal/anal atresia, and absent internal genitalia). Most cases are stillborn, or die during, or shortly after, birth.