Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome. ORPHA:1112

Aphalangy-hemivertebrae-urogenital-intestinal dysgenesis syndrome is an extremely rare congenital limb malformation syndrome, described in only 3 patients to date,characterized by the association of hypoplasia or aplasia of the hand and foot phalanges, hemivertebrae and various urogenital and/or intestinal abnormalities (i.e. dysgenesis of the urogenital tract and rectum). There have been no further descriptions in the literature since 1991.