Splenogonadal fusion-limb defects-micrognathia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Splenogonadal fusion-limb defects-micrognathia syndrome. ORPHA:2063

Splenogonadal fusion-limb defects-micrognatia syndrome is a rare dysostosis syndrome characterized by abnormal fusion of the spleen with the gonad (or more rarely with remnants of the mesonephros), limb abnormalities (consisting of amelia or severe reduction defects leading to upper and/or lower rudimentary limbs) and orofacial abnormalities such as cleft palate, bifid uvula, microglossia and mandibular hypoplasia. It could also be associated with other malformations such as cryptorchidism, anal stenosis/atresia, hypoplastic lungs and cardiac malformations.