

Sirenomelia, which is also known as mermaid syndrome, is an extremely rare congenital developmental disorder characterized by anomalies of the lower spine and the lower limbs. Affected infants are born with partial or complete fusion of the legs. Additional malformations may also occur including genitourinary abnormalities, gastrointestinal abnormalities, anomalies of the lumbarsacral spine and pelvis and absence or underdevelopment (agenesis) of one or both kidneys. Affected infants may have one foot, no feet or both feet, which may be rotated externally. The tailbone is usually absent and the sacrum is partially or completely absent as well. Additional conditions may occur with sirenomelia including imperforate anus, spina bifida, and heart (cardiac) malformations. Sirenomelia is often fatal during the newborn period. The exact cause of sirenomelia is unknown, most cases occur randomly for no apparent reason (sporadically). Sirenomelia affects males more often than females by a ratio of 2.7-1. The exact incidence is unknown, but sirenomelia is estimated to occur in approximately 1 in 60,000 to 100,000 births. Sirenomelia occurs with greater frequency in one twin of identical (monozygotic) twins than it does in fraternal (dizygotic) twins or individuals. A diagnosis of sirenomelia can be made prenatally, most often during the second trimester, by fetal ultrasound. An ultrasound is an exam that uses high-frequency sound waves to produce an image of the developing fetus. A fetal ultrasound can detect some of the defects associated with sirenomelia.