Sirenomelia

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Abstract:

Sirenomelia is a rare malformation of caudal part of embryo. It is characterised by complete or partial fusion of the legs into a single lower limb. Abnormalities of the kidneys, large intestines and genitalia are common. Sirenomelia cases have only one umbilical artery and one vein. Upper body birth defects are rare and include abnormalities of heart, lungs, arms, spine and brain. Here we report a case of sirenomelia with uncommon upper body birth defects involving right forearm and hand, and the rib cage. Vascular steal phenomenon cannot explain the upper body birth defects.

Case report

A 22-year-old primigravida with nonconsanguineous marriage without any prior ultrasonographic screening of pregnancy status presented with obstructed labour at 34 weeks of gestation.

She had a normal antenatal course except poor progression of abdominal girth. There was no history of medication in the early pregnancy. She was nondiabetic. Her past medical history and family history were unremarkable. Caesarian section was done. A 1500 gm, stillborn baby of undetermined sex was born. Head circumference was 26 cm.

The infant had flattened facies, fused lower extremities, single umbilical artery, absent anal orifice and absent external genitalia (Figures 1–3). Right forearm was short with fused fingers. The fetus was diagnosed to be a case of sirenomelia.

Postmortem radiograph showed hypoplastic pelvis, fused femurs at the proximal end, two tibias and one fibula. In right forearm a small radius/ulna and a piece of small bone were present with absence of carpal, metacarpal and phalangeal bones. Rib cage on right side was poorly developed with fewer numbers of ribs (Figure 4).

Autopsy revealed bilateral renal agenesis, absent ureters, urinary bladder and urethra, absent internal genitalia, colorectal agenesis with blind ended caecum, caudal tapering of abdominal aorta below the origin of only umbilical artery and right lung hypoplasia.
Figures 1–3. Photographs showing a stillborn baby with single lower limb with imperforate anus, absent genitalia, flat face, and short right forearm with fused fingers (the authors obtained consent from the baby’s mother for publication of these images)

Figure 4. Radiograph showing single lower limb with two femurs fused at proximal end, two tibias and one fibula. In right forearm there is a small radius or ulna with absence of carpal, metacarpal and phalangeal bones. Rib cage on right side was poorly developed with fewer numbers of ribs
Discussion

Sirenomelia, also known as Mermaid Syndrome, is a lethal birth defect of the lower body characterised by apparent fusion of the legs into a single lower limb. Infants resemble the siren of Greek mythology. Other birth defects are always present in sirenomelia. The most common abnormalities are of kidneys, large intestines and genitalia. Exact cause of sirenomelia is not known. It is non-hereditary. Some undetermined teratogenic agents may be responsible for it.

Failure of caudal mesoderm blastogenesis, mechanical compression by amniotic bands or oligohydramnios, and unknown genetic mechanism have also been postulated as causes of sirenomelia. There are some associations with in-vitro fertilisation, twin pregnancies and diabetic mothers. It was also thought to be an extreme case of caudal regression syndrome. Defect in primitive streak had been proposed as a causative factor. Attention have been drawn to the overlap in phenotypic features of sirenomelia and VATER. Vascular steal phenomenon is a proposed mechanism causing relative ischaemia below a persistent vitelline artery that diverts blood from the abdominal aorta and caudal structures to the placenta.

Spectrum of sirenomelia varies from simple cutaneous fusion of lower limbs to absence of all bones (except fused femur). Pelvic bones may also be absent, fused or poorly formed. Back of the knee and foot (if present) may face forward due to rotation. Urogenital abnormalities are common including renal agenesis, absence of bladder and urethra, and absent or poorly formed internal and external genitalia. Blind ending colon and imperforate anus may also present. Only one umbilical artery is present.

Oligohydramnios is frequent in mother, which can interfere with development of lungs of the fetus and cause a flattened face from compression against the mother's abdomen. Upper body birth defects are very rare and include abnormalities in heart, lungs, arms, spine and brain. The present case of sirenomelia also had uncommon upper body birth defects involving right forearm and hand, and the rib cage.

Single umbilical artery in sirenomelia is thought to arise from primitive vitelline arteries of embryo and has direct continuation with abdominal aorta. This vitelline umbilical artery steals blood and nutrition from the lower body and diverts it to the placenta. As a result, urogenital and gastrointestinal systems and lower extremities do not form properly. Though vascular steal theory can explain the lower body defects, it can not explain upper body birth defects including cardiac, cranial and radial malformations.

Single umbilical artery occurs in about 1% of all live-born infants but association of other birth defects is relatively low. Persutte and Hobbins described congenital malformations associated with single umbilical artery into three groups:

- Malformations identified with prenatal ultrasound;
- Difficult to be diagnosed prenatally;
- Unlikely to be diagnosed prenatally.
They concluded that nearly two-thirds of all congenital malformations associated with single umbilical artery could be missed on a prenatal ultrasound examination.\textsuperscript{11}

Antenatal diagnosis of sirenomelia is done by ultrasonography. Oligohydramnios can prevent a clear view of the fetus by ultrasound. Observation of lower extremity fusion and bilateral renal agenesis are helpful for diagnosis of sirenomelia. Blood vessel anomalies can be detected by colour-flow imaging. Serum marker for antenatal diagnosis of sirenomelia is currently not available.

Sirenomelia is fatal. Babies are stillborn, or live-born with survival for few minutes to few days. Babies born alive with functioning kidneys may survive with appropriate surgical management.\textsuperscript{12,13}

Antenatal ultrasonography is important for detecting fetal anomalies. Diagnosis of sirenomelia was missed in our case because of lack of antenatal check-ups including ultrasonography. Diagnosis by first trimester is helpful for planning early termination of pregnancy and minimises the trauma of termination at advanced gestation.

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References:
