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Letter to the Editor

Antenatal diagnosis of sympus dipus by MRI

A 30 year old, G2P1L1 at 23 weeks period of gestation was referred to our facility for uncontrolled pre-gestational diabetes. Her glycosylated hemoglobin (HbA1c) was 6.9 %. She was started on insulin. Ultrasound done for fetal well-being revealed an-hydramnios, single umbilical artery, bilateral hypoplastic lungs with reduction in thoracic cavity diameters. Bilateral kidneys could not be visualized. Lower limbs were placed suspiciously close to each other. Due to uncertainty of diagnosis, fetal MRI was done which revealed excessive soft tissue between the two lower limbs. Bilateral femur and tibia were separate. Findings were suggestive of sympus dipus. In addition there was bilateral renal agenesis (Image 1a and b). MRI brain showed Viking helmet sign with bilateral parallel ventricles indicating absence of corpus callosum (Image 1c and d). Since patient was still within the legal limit, couple chose to undergo medical termination of pregnancy. Fetus (Image 2a–d) was had fused lower limbs with 10 digits at distal end, low set ears, Potter's facies, absent urogenital opening, contractures of middle finger in both hands and contracture of right upper limb. Fetal X Ray (Image 2e) showed well-formed bilateral femur, tibia and absent fibula with normal upper limbs. Fetal autopsy was not done as couple did not give consent.

Sirenomelia is a rare multi-organ congenital developmental disorder in which caudal part of the fetus is abnormally developed leading to fused lower limbs. Approximate incidence is estimated to be around 1 in 60,000–70,000 pregnancies [1]. Sirenomelia is also known as mermaid syndrome due to its similarity to a fabled marine creature with head and upper body of a human female and lower half of a fish. This disorder is associated with anomalies of the genito-urinary and gastro-intestinal tract, congenital heart defects, pulmonary hypoplasia and ambiguous external genitalia [2]. Combination of the above mentioned anomalies contributes to lethality of this syndrome. Bilateral renal agenesis leads to an-hydramnios. Single umbilical artery is a pathognomonic finding of sympus dipus. According to vascular steal hypothesis [3], single umbilical artery leads to deficient blood flow and nutrient supply to the caudal mesoderm, which in turn results in agenesis of midline structures and subsequent abnormal approximation of both lower limb fields. Defective blastogenesis hypothesis [4], states that a defect in development of caudal mesoderm occurs due to a teratogenic event during the gastrulation stage which interferes with formation of notochord, resulting in abnormal development of caudal structures.

Stocker & Heifetz classified mermaid syndrome into seven categories (Image 3) [5]. In Sympus dipus also known as symmelia, thigh and leg long bones are separate. In Sympus monopus or Uromelia, as the name describes femur start approximating and fusing from above downwards. In sympus apus or sirenomelia, bilateral lower limb long bones are fused. Type VII is the most lethal form of sirenomelia. Our patient had fetus with sympus dipus [symmelia type III].

There is a strong association of sirenomelia with pre-gestational diabetes mellitus and VACTREL syndrome. The relative risk of

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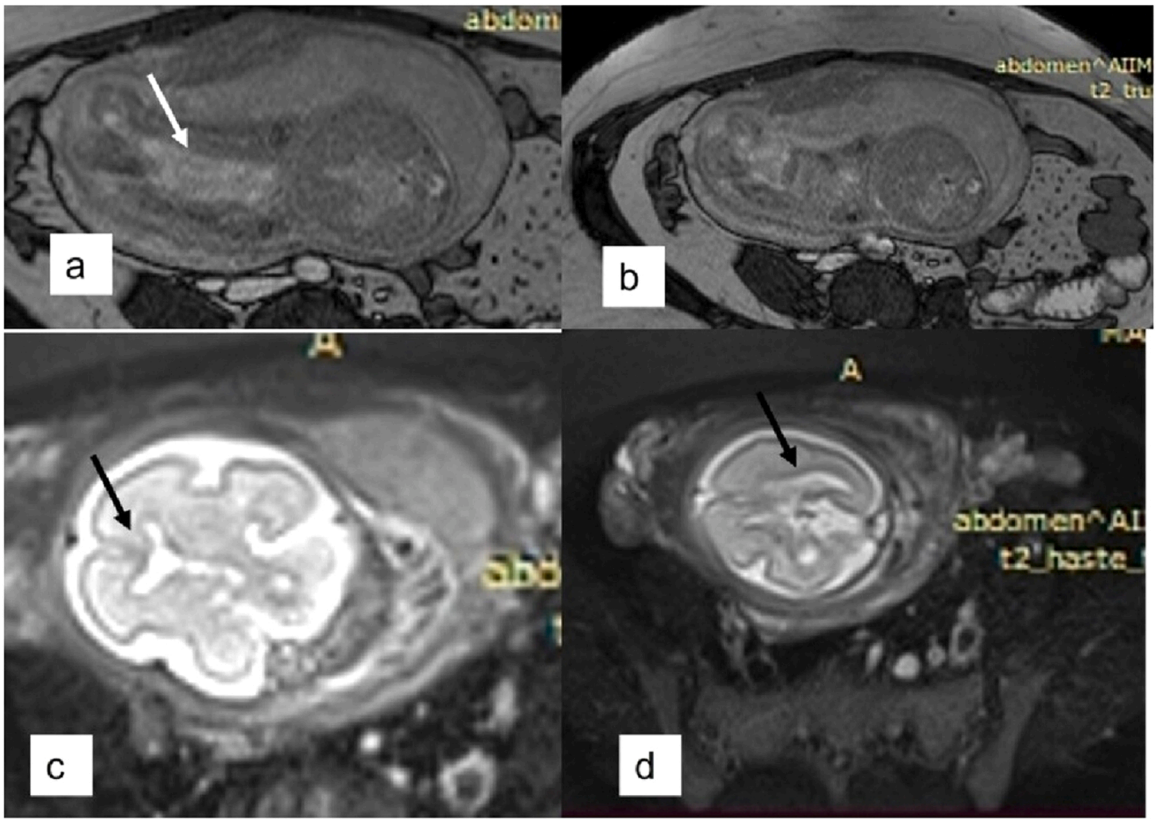

Image 1. Fetal MRI (a) T2w axial scan HASTE sequence depicting the hyperintense soft tissue in between lower limbs (arrow) suggestive of soft tissue sirenomelia. (b) Axial T2w axial scans Tru Fi sequence show absence of bilateral Kidneys. (c and d) Axial T2w TruFi sequence show Viking helmet sign (arrow in c) and bilateral parallel ventricles suggestive of corpus callosal agenesis. Anhydramnios is present. [HASTE–Half fourier Single-shot Turbo spin-Echo; Tru Fi: True Fast Imaging].

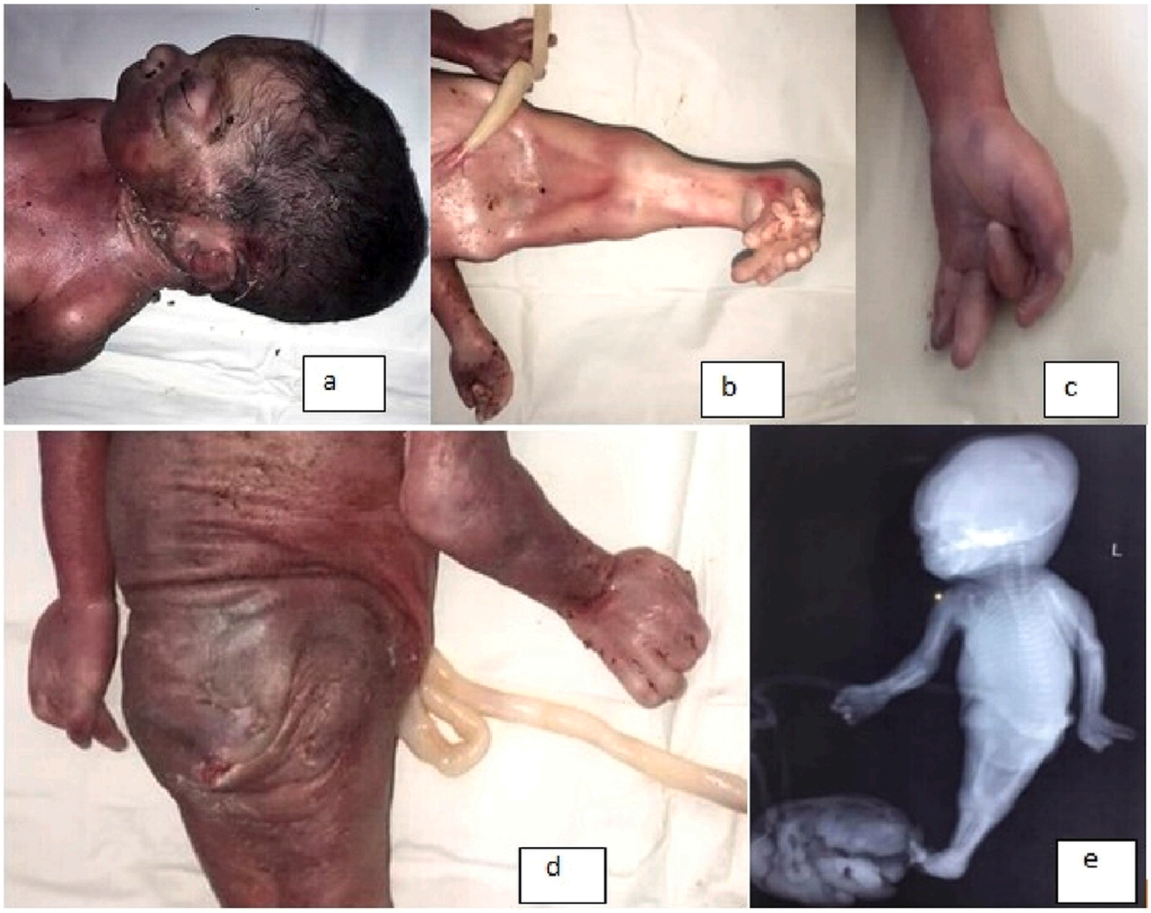

Image 2. (a) Swollen face with distinct features of Potter's facies, low set ears; (b) Absent urogenital opening; both lower limb fused, 10 digits at end of fused lower limbs; (c) Contractures of middle finger in both upper limbs (d) Contracture of right upper limb, ambiguous anal opening, (e) Infantogram: femur and tibia were well formed, bilateral upper limbs bones well formed.

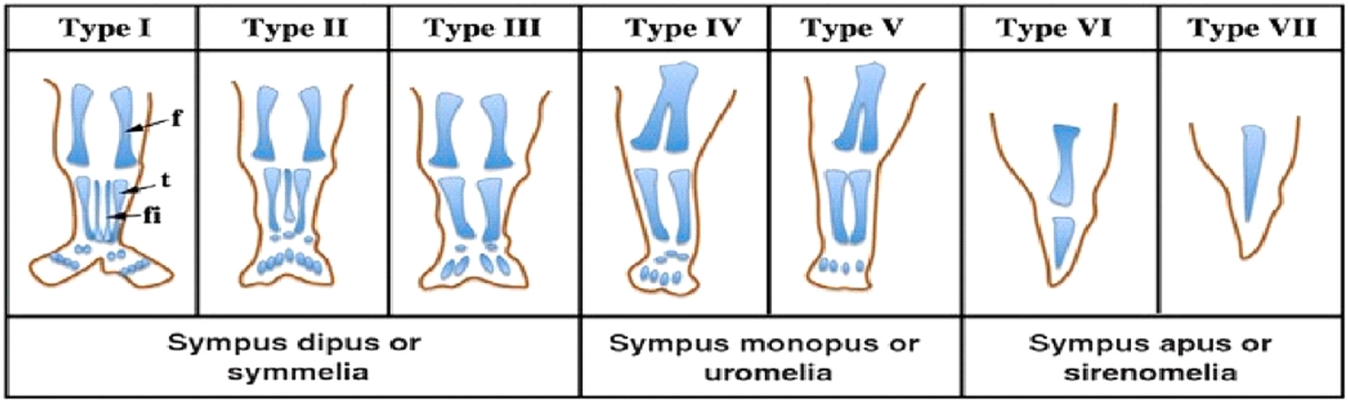

Image 3. Stocker & Heifetz classification of Mermaid syndrome [6]. (f = femur, t = tibia, fi = fibula).

sirenomelia among women with uncontrolled diabetes mellitus during period of organogenesis is 1 in 200–250 [2].

Being a rare and almost always a lethal anomaly, a high index of suspicion should be maintained if ultrasound is suggestive of anhydramnios/severe oligohydramnios with single umbilical artery. The inability to show separate lower limbs with decreased mobility on ultrasonography should raise suspicion of fused lower extremities, though final diagnosis may be difficult due to associated severe oligohydramnios which hampers proper evaluation of fetus. Fetal MRI may be done to confirm the findings. MRI raises diagnostic confidence for detection of anomalies with sensitivity and diagnostic accuracy of 96.6 % and added benefits in cases of severe oligohydramnios/anhydramnios and maternal obesity which often leads to inconclusive reporting on ultrasound.

Conflict of Interest

There is no conflict of interest.

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