What is your diagnosis?

A 25-year-old primigravida, was admitted to the antenatal ward at 32 weeks gestation with decreased fetal movements. The patient lived in a remote hilly region and did not go for antenatal checkups because of the non-availability of transport due to the lockdown imposed during the coronavirus pandemic. The patient did not undergo an anomaly scan in the first or second trimester. The present pregnancy was conceived spontaneously, without any history of ovulation induction. There was no history of consanguineous marriage. There was no history of teratogenic drug exposure in the antenatal period. The patient did not have any risk factors for gestational diabetes, body mass index was 22.6 kg/m^2, and family history was not significant. Blood sugar profile was normal after admission, and during the intrapartum and postpartum periods while hemoglobinA1c was normal at 5.8%.

On examination, the fundal height corresponded to 26 weeks, and fetal parts were palpable superficially, suggesting decreased liquor and fetal growth restriction (FGR). The ultrasound showed a single live fetus in breech presentation, corresponding to gestational age 32 weeks with severe FGR, abdominal circumference less than the third centile, biparietal diameter and head circumference at the fifth centile and femur length at the tenth centile with placenta praevia and almost absent liquor. Due to grossly decreased liquor, the radiologist could not comment on fetal anatomy at this gestation. A Doppler study of the umbilical arteries suggested reversed end-diastolic flow with brain sparing effect. Cardiotocography was suggestive of prolonged late decelerations. After discussion with the parents, the patient was taken for lower segment caesarean section because of primigravida with placenta praevia, breech presentation and Stage 4 FGR with high suspicion of fetal acidosis (1).

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Answer

A live baby, weighing 1.2 kg and with Apgar scores of 3/3/1, was born through lower segment caesarean section. The baby had a phenotype suggestive of Potter sequence, with flattening of the nose, hypertelorism with prominent epicanthal fold, micrognathia, and dysplastic and low set ears. The baby also had webbed neck, flexion deformities of hands and wrist, and fused lower limbs without feet, suggestive of Sirenomelia (Figure 1). A bluish-coloured cystic swelling, approximately 4x4x5 cm in the lumbosacral region, suggestive of lumbar myelomeningocele, was present. The external genitalia and anal openings were absent (Figure 2). There was a single umbilical artery. The baby died 10 minutes after birth. The parents refused an autopsy.

Sirenomelia is a polytopic, multi-systemic congenital anomaly with an unknown etiology. The reported incidence is 1.5-4.2 per 100,000 births (2). It gets its name due to the analogy of the born fetus to the mythological siren or "mermaid" (3). In Greek mythology, these creatures were depicted as half woman/half fish and are believed to sing enchanting songs that lure sailors to death (4). Sirenomelia is characterized by varying degrees of fusion of the lower limbs, usually a single axially positioned lower limb, associated with anomalies of the lower spine and urogenital and lower gastrointestinal tracts. Although the exact etiopathology is unknown, the vascular steal phenomenon and defective blastogenesis (5) are the most widely accepted hypotheses regarding its origin. Maternal age of less than 20 or more than 40 years, diabetes mellitus, genetic predisposition, smoking and cocaine abuse and vascular mal-perfusion have been reported to be risk factors in a few studies (6,7).

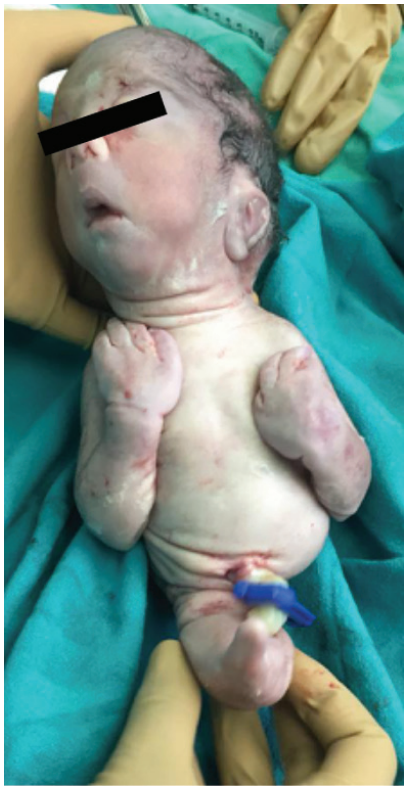

Figure 1. Note the presence of Potter facies, low set ears, micrognathia, club hands and fused lower limbs

Sirenomelia is uniformly associated with poor fetal prognosis because of the associated complications related to abnormal kidney, lung, heart, and bladder development and function (8). Other lethal congenital malformations include body stalk anomaly, anencephaly, autosomal recessive polycystic kidney disease, some forms of skeletal dysplasia, bilateral renal agenesis with pulmonary hypoplasia or fetuses with multiple anomalies, especially associated with a chromosomal abnormality. Ideally, if detected before 24 weeks of gestation, termination of pregnancy is the preferred option. It is always preferable to deliver vaginally in these conditions, and parents should be counselled regarding the poor neonatal prognosis. In the present case, caesarean section was performed due to placenta praevia and poor fetal condition.

The diagnosis is usually easily made by ultrasound at the first-trimester nuchal translucency scan. However, in low and middle-income countries, it is not uncommon for a patient to visit the physician for the first time at advanced gestations. The diagnosis at these times can be challenging due to associated severe oligohydramnios. Any pregnancy, presenting with severe oligohydramnios in the late second or third trimester should be studied in detail with colour Doppler imaging to map the fetal vasculature. Imaging for mid-trimester an-hydramnios should include colour Doppler for renal arteries, especially in a patient who had not undergone an anomaly scan in the first or second trimester. Congenital renal abnormalities in the antenatal period have the highest probability of being associated with oligohydramnios. Aberrant abdominal vasculature or absent

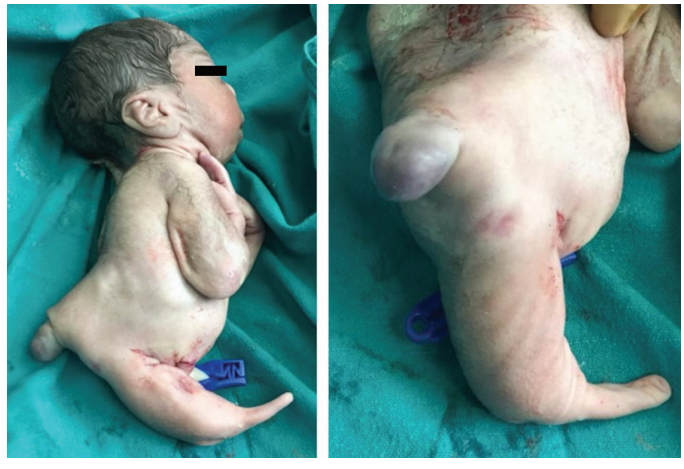

Figure 2. Bluish swelling in the sacral region, suggestive of lumbosacral myelomeningocele

renal arteries may be important clues to the possible diagnosis of sirenomelia (9).

A high index of suspicion is needed to diagnose such cases because, at advanced gestation, the associated oligohydramnios makes it challenging to study the fetal anatomy. Although Sirenomelia is a rare entity, it should be included in the differential diagnosis, when severe oligohydramnios or anhydramnios is detected on antenatal ultrasound.

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References

1. Figueras F, Gratacós E. Update on the diagnosis and classification of fetal growth restriction and proposal of a stage-based management protocol. Fetal Diagn Ther 2014; 36: 86-98.
2. Browne M, Fitchev P, Adley B, Crawford SE. Sirenomelia with an angiomatous lumbosacral myelocystocele in a full-term infant. J Perinatol 2004; 24: 329-31.
3. Boer LL, Morava E, Klein WM, Schepens-Franke AN, Oostra RJ. Sirenomelia: A multi-systemic polytopic field defect with ongoing controversies. Birth Defects Res 2017; 109: 791-804.
4. Romano S, Esposito V, Fonda C, Russo A, Grassi R. Beyond the myth: The mermaid syndrome from Homerus to Andersen. A tribute to Hans Christian Andersen's bicentennial of birth. Eur J Radiol 2006; 58: 252-9.
5. Garrido-Allepuz C, González-Lamuño D, Ros MA. Sirenomelia Phenotype in Bmp7;Shh Compound Mutants: A Novel Experimental Model for Studies of Caudal Body Malformations. PLoS One 2012; 7: e44962.
6. Tilahun T, Desta D. Successful expectant management of the anomalous fetus with sirenomelia in twin pregnancy: A case report and literature review. Int Med Case Rep J 2021; 14: 229-32.
7. Kucuk Ş, Kucuk İG. Sirenomelia (Mermaid syndrome): A case report. Turk Patoloji Derg 2020; 36: 256-60.
8. Shojaee A, Ronnasian F, Behnam M, Salehi M. Sirenomelia: two case reports. J Med Case Rep 2021; 15: 217.
9. Patel S, Suchet I. The role of color and power Doppler ultrasound in the prenatal diagnosis of sirenomelia. Ultrasound Obstet Gynecol 2004; 24: 684-91.