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Single Umbilical Artery, Mermaid Syndrome in a Diabetic Mother : Sirenomelia a Rare Congenital Deformity

KEYWORDS

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Introduction

Pachajoa and Isaza definedSirenomelia as a rare congenital malformation characterized by a single fused lower limb, its prevalence is estimated at 0.2 per 100,000 birth with different degrees of fusion of the lower extremities and is considered as the extreme form of the anomalous caudal process.[1]Taori KB, Mitra K, Ghonga NP, Gandhi RO, Mammen T and Sahu J found that male to female ratio is 3: 1, when sex can be determined. This deformity is synonymous with Mermaid, Monopodia, Symmelia, Sympus, Symposia, and Uromelia.[3] Rougemont AL, Bouron-Dal Soglio D, De´silets V reported the pathognomonic features include renal dysgenesis with absent external genitals, two-vessel umbilical cord, imperforate anus, and variable degrees of malformations of the lower limbs.[4]Tanha FD, Googol N and Kaveh Mconcluded etiology remains controversial however it is strongly associated with maternal diabetes with a relative risk of 1:200-250.[5]Blaicher W, Lee A and Deutinger J notedfew teratogenic factors like cadmium, cocaine, ionizing radiation and isotretinoin are reported to be associated with sirenomelia.[6]Castori M, Silvestri E, Cappellacci S,Binni F, Sforzolini GS and Grammatico P showed thatthe anomaly is characterized by fusion and hypotrophy of lower limbs, with severe urogenital anomalies, leading to oligohydramnios in the second half of pregnancy, making the sonographic diagnosis even more difficult.Prognosis of sirenomelia is fatal because of pulmonary hypoplasia and renal agenesis.[7]We report a neonate born at 36-37 week with this anomaly to a mother with gestational diabetes mellitus at tertiary care center of northern India.

Case report

A newborn was born to a 24 year old, third gravida hindu mother (married non consangiously) who had an unsupervised and uneventful antenatal period. The mother was detected to have deranged blood sugar readings consistent with gestational diabetes mellitus at 36 week of gestation. Baby was born by vaginal delivery and weighed 1.8 kg and had an APGAR score of 2/10 at 5 and 10 minutes of life. Head circumference, Chest circumference and length of the baby were 30, 27 and 48 cm respectively.Baby was grossly malformed with fusion of lower limbs with single rudimentary foot, imperforate anus, absence of external genitalia with no anal opening and single umblical artery apparent on examination. Upper limbs were well in place (Figs. 1 and 2).The baby expired within one hour of birth. Postmortem radiological investigations revealed malformed sacrum, fused lower limb bones, lung hypoplasia and renal agenesis with intraabdominal testis. Large intestine ended with a blind sigmoid colon. Umbilical artery was single in number and abdominal aorta continued downward as median sacral arteryinstead of dividing into two iliac arteries. In the present case absence of iliac arteries and endof aorta as median sacral artery were unique in its findings.

FIG 1

Discussion

Castori M, Silvestri E, Cappellacci S et. al, explained two main etiological hypotheses- the vascular steal theory and defect in mesodermal cell migration of the caudal embryo region during primitive streak period. Single umblical artery arising from abdominal aorta diverting the blood supply and nutrients from lower part of body and leading to growth arrest or variable degree of abnormalities in lower part of body. Maternal diabetes mellitus,genetic predisposition, environmental factors have been proposed as causative factors.[6]Thottungal AD, Charles AK, Dickinson JE and Bower C opined thatsirenomelia is a severe form of caudal regression syndrome and VACTERL (vertebral defects, anorectal atresia, cardiac abnormalities, tracheo-oesophageal fistula, renal and limb abnormalities) association due to overlapping features and maternal diabetes is associated with both conditions.[7] Valenzano M, Paoletti Rand Rossi A found thatdiagnosis of this lethal condition is possible in the first trimester, which become difficult in second trimester because of significant oligohydroamnios due to renal agenesis.The differential diagnosis of sirenomelia in the first trimester should be taken into account in the presence of bilateral renal agenesis, malformed lower limbs with reduced movements, and single umbilical artery. [8]Langer B, Stoll C and Nicolau Rclassified sonographic appearance of the fusion of the lower limbs (symelia) into three types: apus—one tibia, one femur, no feet; unipus—two femurs, two tibiae, two fibulae, one foot; dipus—two fused legs with two feet like a mermaid.[9]Sepulveda W, Corral E and Sanchez J supportedthat sonography at the end of the first trimester together with the power Doppler imaging enables the detection of abnormal vasculature: single aberrant umbilical artery as a branch of persistent vitelline artery and absent renal arteries.[10]Van Keirsbilck J, Cannie M and Robrechts C were also of the opinion that

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severe oligohydramnios hinders a reliable sonographic visualization of the lower extremitiesin second trimester.High-quality MRI imaging is better option in second trimester to diagnose this condition[11]

The condition is lethal because of bilateral renal agenesis which leads to severe oligohydraminos and lung hypoplasia.There is no treatment available for sirenomelia. However prevention is possible and should be the goal.Because of strong association of sirenomelia and maternal diabetes mellitus maternal blood glucose level should be regularly monitored in preconceptual period to prevent this anomaly.

Conclusion

We conclude that the correct diagnosis of sirenomelia as a lethal polymorphic anomaly with a

very poor prognosis is already feasible at the end of the first trimester. Dosedla E, Calda P and Kacerovsky M showed that two/three/four-dimensional US of complex malformations facilitates prenatal diagnosis and parental counseling. [12]Detection of rare congenital anomaly like Sirenomelia among mothers with gestational diabetes is essential to plan early termination of pregnancy and to reduce the adverse psychological impact on parents. This was not possible in our case as the mother's pregnancy was unsupervised.

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