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Case Report

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Sirenomelia: a case report

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Abstract

Objective: It is aimed in this study to evaluate the characteristics of a case of sirenomelia diagnosed at our obstetrics department.

Case: A 23 years old woman (G2P1) with an intrauterine pregnancy of 35 weeks of pregnancy referred to our clinic with a cervical dilatation of 9 cm and delivered a baby (2,600 g and 1st and 5th minutes of Apgar scores of 3-0) with fetal anomalies such as single lower extremities, rudimentary foot and external genitalia which are in the shape of a small bud. The upper extremities and anal hiatus were normal. The initial diagnosis of the baby was sirenomelia. Autopsy also confirmed the diagnosis of sirenomelia. There were undeveloped bone pelvis, single lower extremities, fusion of the femur bones, rudimentary tibia bones and absence of fibulas. Urinary bladder, ureter and urethra were absent, and rectum was atresic. On microscopic evaluation there was haemorrhage at lungs, liver, heart and kidney. The placenta and umbilical cord were normal.

Conclusion: Sirenomelia is a rare and lethal congenital anomaly. It is important to diagnose this anomaly in order to give counseling to the family for termination of the pregnancy.

Key words: Sirenomelia, congenital malformations, autopsy.

Sirenomelia: Olgu sunumu

Amaç: Bu çalışmada obstetri kliniğimizde tanı konulan bir sirenomelia olgusunun tartışılması amaçlanmaktadır.

Olgu: Yirmi üç yaşında (G2P1), 35 haftalık gebe kadın servikal açıklığı 9 cm iken kliniğimize başvurdu. Normal vajinal yol ile 2600 g, 1. ve 5. dakika Apgar skorları 3-0 bir bebek doğurtuldu. Bebeğin anomalileri tek alt ekstremite, rudimenter ayak ve küçük bir tomurcuk şeklinde olan eksternal genitalya idi. Üst ekstremiteler ve anal açıklık normal idi. Bebeğin ilk tanısı sirenomelia idi. Otopsi de sirenomelia tanısını desteklemekteydi. Gelişmemiş kemik pelvis, tek alt ekstremite, femurların füzyonu, rudimenter tibialar ve fibulaların bulunmadığı tespit edildi. Mesane, üreter ve üretra mevcut değildi ve rektum da atrezik idi. Mikroskopik incelemede akciğerler, karaciğer, kalp ve böbreklerde hemoraji mevcuttu. Plasenta ve umbilikal kord normal idi.

Sonuç: Sirenomelia nadir görülen konjenital bir anomalidir. Ölümcül bir anomali olması nedeniyle, erken prenatal tanı konulması durumunda aileye terminasyon açısından danışmanlık verilebilir.

Anahtar sözcükler: Sirenomelia, konjenital anomaliler, otopsi.

Introduction

Sirenomelia is a congenital anomaly seen in 1 out of 60,000 -100,000.[1] The rate of male/female fetus is 2.7/1 and it is prevalent among monozygotic twins.[2] Sirenomelia is defined as a lethal anomaly in which severe urogenital malformations are seen together with the fusion, rotation and hypotrophy of lower extremities.[3] This anomaly may be a variant of caudal regression syndrome (CRS); however, it is distinguished by the presence of two umbilical arteries and non-lethal renal anomalies, non-existence of the fusion of lower extremities and tracheoesophageal, neural tube and cardiac anomalies.[4] In this study, our purpose is to evaluate the characteristics of sirenomelia case which was established intrapartum diagnosis and delivered in our clinic.

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Case Report

Twenty-three-year-old woman (G2P1) with an intrauterine pregnancy of 35 weeks of pregnancy referred to our clinic with a cervical dilatation of 9 cm. There was no prominent characteristic in her obstetric history and it was observed that she had a kin marriage. It was found in her family history that one of her second degree relatives delivered a baby with anencephaly and another relative had two deliveries which were neural tube defects. The patient delivered by normal vaginal way and delivered a baby with fetal anomalies (2,600 g and 1st and 5th minutes of Apgar scores of 3-0). The anomalies of the baby were single and attached lower extremities, rudimentary foot and undeveloped external genitalia which are in the shape of a small bud. Upper extremities and anal hiatus were normal. Initial diagnosis of the baby was sirenomelia. Autopsy also confirmed the diagnosis of sirenomelia. There were undeveloped bone pelvis, single lower extremities, fusion of the femur bones, rudimentary tibia bones, and absence of fibulas. Urinary bladder, ureter and urethra were absent, and rectum was atresic (**Figures 1** and **2**). On microscopic evaluation, there was haemorrhage at lungs, liver, heart and kidney. The placenta and umbilical cord were normal. No sacral agenesia, hypoplastic fetus, combined femur and tibia, and absence of fibula were detected in the postpartum radiography (**Figure 3**).

Discussion

Sirenomelia is a congenital anomaly seen in 1 out of 60,000 -100,000[1] and is defined as a lethal anomaly in which severe urogenital malformations are seen together with the fusion, rotation and hypotrophy or atrophy of lower extremities.[3]

Maternal diabetes and genetic predisposition are the predisposing factors for sirenomelia.[5,6] Only five

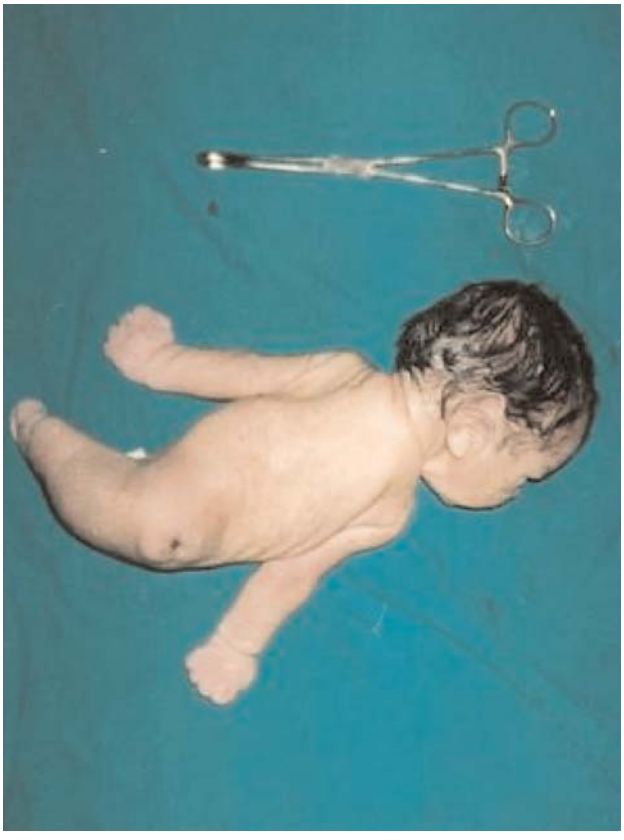

Figure 1. Dorsal view of the infant; caudal regression and fusion on lower extremities are seen.

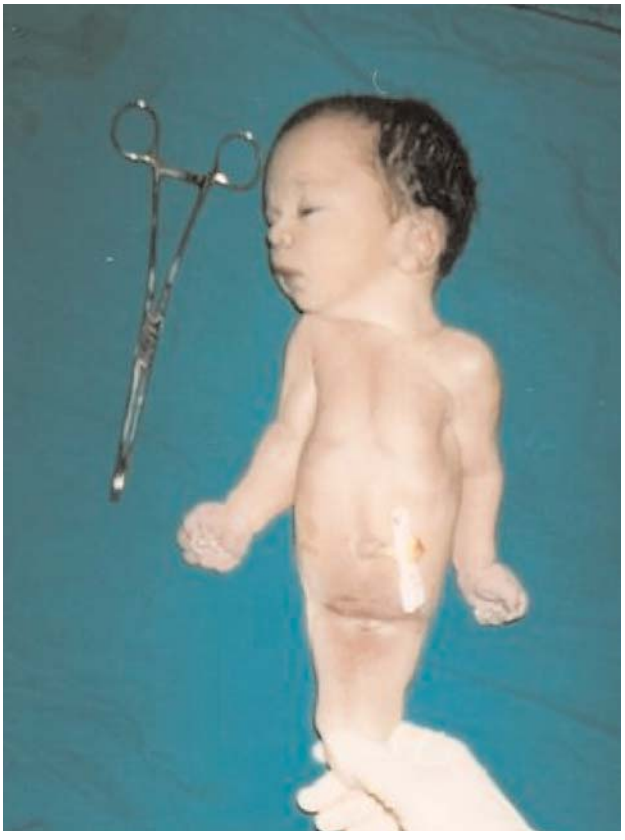

Figure 2. Fusion on lower extremities and typical sirenomelia are seen on the frontal view of the infant.

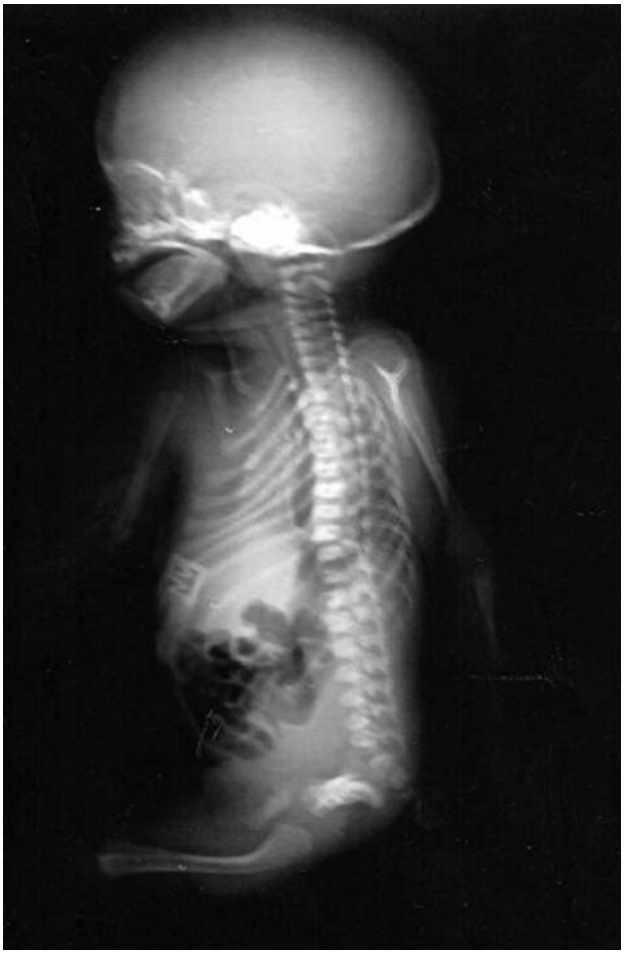

Figure 3. Sacral agenesia, hypoplastic fetus, combined femur and tibia, and absence of fibula are seen in the postmortem radiography.

sirenomelia cases which lived were defined in the English literature.[7] Pathogenesis of sirenomelia is still not known; only maternal disease known as associated with sirenomelia is diabetes mellitus.[8] In our case, there was no evidence related with diabetes mellitus. It was reported that defects similar to sirenomelia appeared after the exposure to etretinate (synthetic vitamin A analogue) and ochratoxin (fungal toxin) applied on animals experientially.[9,10] In our case, antenatal was free of problems and there was no medical/surgical disease history or medication history.

The existence of anencephaly and neural tube defect in family history of our case makes us to consider that the etiology is genetic; however, genetic analysis was not performed since the family did not accept it.

A prenatal ultrasonography including oligohydramnios, malformed lower extremities and normal upper extremities, and single umbilical artery should cause clinician to suspect in favor of sirenomelia. Oligohydramnios is caused by renal agenesia and it generally occurs during second trimester.[11]

Conclusion

Sirenomelai is a rare congenital anomaly. Since it is a lethal anomaly, counseling may be provided to the family for termination of pregnancy in case of an early perinatal diagnosis.

Conflicts of Interest: No conflicts declared.

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