Olgu Sunumu

Sirenomelia: A Case Of Severe Caudal Regression Syndrome

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ÖZET

Sirenomelia: Ağır Kaudal Regresyon Sendromlu Bir Olgu

Nadir olarak görülen;birleşik alt ekstremiteler, fibula yokluğu, bilateral renal agenezi ve geniş tek umbilikal arter ile karakterize ağır kaudal regresyon sendromu tanımlanmıştır. Yaşam şansı çok nadir olup yalnız bilateral renal agenezi yokluğunda mümkündür. Utrasonografi ile erken tanının ve ardından daha az travmatik terminas-yonun önemi tartışılmıştır.

Anahtar Kelimeler: *Sirenomelia, Kaudal regresyon sendromu, Antenatal tanı*

SUMMARY

A rare case of severe caudal regression syndrome including fused lower limbs, absence of fibula, anal atresia, bilateral renal agenesis, and a single large umbilical artery is described. Survival is extremely rare in such cases and only possible in the absence of bilateral renal agenesis.The importance of early diagnosis by ultrasonography and subsequently less travmatic termination are discussed.

Key Words: *Sirenomelia, Caudal regression syndrome, Antenatal diagnosis*

INTRODUCTION

Sirenomelia is a rare and most severe case of caudal regression syndrome. It is seen in 0.01 to 0.16/10000 births.(1) There is an abnormality in the development of the allantois at the third week of gestation. Cases are mostly sporadic and there is no known factor to be responsible for the disease. It is seen with increased incidence in diabetic mothers (200-400 times more than in normal pregnancies), but not pathognomonic for this group of patients. Sacral agenesis is the most characteristic anomaly associated with maternal diabetes mellitus. We must not forget that it can also be seen in fetuses of non-diabetic mothers as in our case. Caudal regression syndrome includes malformations ranging from mild forms of sacral agenesis to severe limb deformities. These deformities are related with multifactorial inheritance and examples of midline developmental field concept. There is an abnormality in the development of the allantois at the third week of gestation. Vascular abnormalities usually found in this condition can explain defects found distal to the malformed arteries. The common feature is the presence of a single large artery, arising high in the abdominal cavity, which assumes the function of the umbilical arteries and diverts nutrients from the caudal end of the embryo distal to the level of its origin. The “steal vessel” derives from the vitellin artery complex, an early embryonic vascular network that suplies the yolk sac. Arteries below the level of this “steal vessel” are underdeveloped and tissue dependent upon them for nutrient supply fail to develop, are malformed, or arrest in some incomplete stage. There are studies suggesting that the single lower extremity in sirenomelia arises from failure of the lower limb bud field to be cleaved into two lateral masses by an intervening allantois.(3) The severity of formed malformations are correlated with the extent of the field of the caudal axis distal to the malformed arteries. As our case shows the most severe form of this spectrum,it is found useful to review the current literature and share difficulties in diagnosing such anomalies.

CASE

A fetus with sirenomelia of non diabetic multigravida mother of her third pregnancy was diagnosed and terminated at 22 weeks of gestation. The mother had no alcohol abuse but was smoking 4-5 cigarettes per day. No history of drug usage during the pregnancy was reported. The patient complaint of the decreased fetal movements for the last two days. Ultrasonographic examination revealed a fetus of 21 weeks of gestation with fetal cardiac activity and severe oligohydramnios. The careful examination was very limited due to the presence of oligohydramnios . One week later, Level 2 Sonogram was performed and the findings of a fetus at 22 weeks of gestation with cardiac activity , anhydramnios and bilateral renal agenesis were reported. Since the prognosis was predicted to be very unfavorable, termination of the pregnancy was adviced. At the end of the procedure, a fetus with sirenomelic appearance and without cardiac activity was observed. The autopsy of the fetus revealed bilateral renal agenesis,urinary bladder agenesis, imperforate anal canal, one artery and one vein in the umbilical cord. In the lower extremities there were tibia, fibula, calcaneus and metatarsal agenesis on the left side but only femur on the right side **(Figure: 1, 2, 3)**.

Figure 1: Anterior view of the sirenomelic fetus

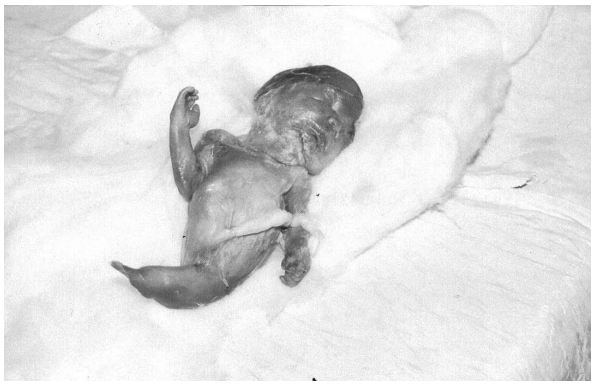


Figure 2: Posterior view of the sirenomelic fetus

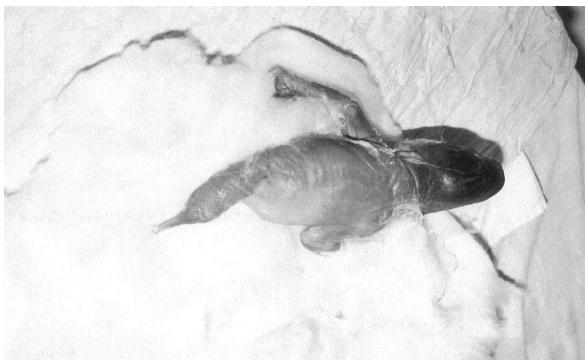
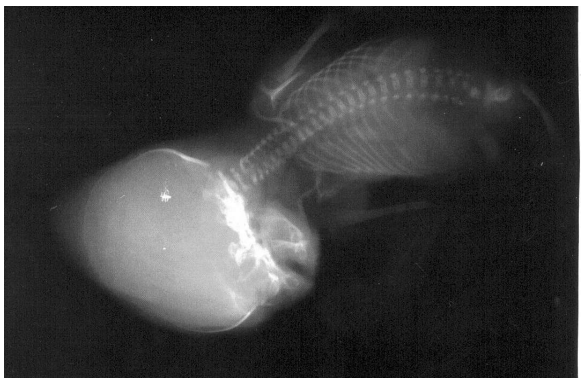


Figure 3: Fetal radiography after the termination



DISCUSSION

The exact cause sirenomelia, rare and mostly fatal malformation with multisystem anomalies, is not known but diabetes mellitus, genetic causes, teratogenic substances have been proposed as etiological factors for the disease. In our case any predisposing factor in the history could not be elucidated.

Although caudal regression is seen as a sequela of maternal diabetes the exact association is contraversial(4). The recent articles investigating pathologic/autopsy diagnosis of caudal regression or sirenomelia suggest that caudal regression is a separate entity distinct from sirenomelia but this classification is still debatable(5). The advances in the knowledge of axial mesodermal patterning during early embryonic development indicate that sirenomelia represents the most severe form of the caudal regression spectrum(6). This extreme example of the caudal regression syndrome called sirenomelia prompted us to focus on the need for the early diagnosis of such anomalies. Early antenatal ultrasound is important because of the bad prognosis and correct diagnosis allows for earlier, less traumatic termination of pregnancy.

Anhydramnios as a usual finding seen with bilateral renal agenesis in the second trimester should allert us for the malformations in the urinary tract. In the literature it is stated that 45 % of the cases can be correctly diagnosed by ultrasound; in the remainder only bilateral agenesis is visible(6). Third-trimester ultrasound diagnosis is usually impaired by severe oligohydramnios related to bilateral renal agenesis, whereas during the early second

trimester the amount of amniotic fluid may be sufficient to allow diagnosis (7). In our case, this syndrome was unfortunately missed during gestation due to severe oligohydramnios. The ultrasound is sometimes unable to recognize even such severe forms of fetal malformations due to anhydramnios and the exact diagnosis is made after birth of the fetus(8). A case of living female neonate with dipodic simelia (fusion of well-developed legs) was presented. In this case the fetus died at 7 weeks of age due to renal failure(9).

Amnioinfusion may assist in differentiating sirenomelia from bilateral renal agenesis or multicystic dysplasia. Doppler ultrasound has been used to correctly identify the persistent midline vitellin artery. It can also be useful to visualize absent or non-functional renal arteries in fetuses with severe oligohydramnios(1). In the literature, it is also stated that some cases of sirenomelia can be detected on prenatal ultrasonograms by demonstration of a single lower extremity, oligohydramnios and bilateral renal agenesis (6). Ultrasound as a diagnostic tool has a crucial role in diagnosing this rare anomaly. If the ultrasound examination is done as early as possible in the second trimester, it is more likely to visualize the sirenomelic appearence of the fetus. As the gestation progresses, oligohydramnios due to bilateral renal agenesis makes it difficult to diagnose sirenomelia in utero. It was focused on the importance of early diagnosis of such rare syndromes such as sirenomelia achieved only by early ultrasound examination of the fetus. We must not forget the association of caudal regression syndrome and diabetes mellitus(4). Association of omphalocele, exomphalos, pentolology of Cantrell and limb bodywall complex in sirenomelia cases suggests a common etiology similar to vascular steal leading to ventral developmental defects in these conditions(10). In our opinion, obstetricians should have basal knowledge about the allerting signs of the disease such as single lower extremity, bilateral renal agenesis and sacral agenesis. It should not be not forgotten to observe the presence of both lower extremities during early second trimester ultrasound in order to diagnose rare caudal regression syndrome or sirenomelia. The earlier the diagnosis, the less traumatic the termination of the pregnancy, either physically or psychologically for the mother.

REFERENCES

1.*Fleischer AC,Manning FA,Jeanty P, RomeroR.Sonography in Obstetrics and Gynecology: Principles and practice; 1996(Fifth edition):Appleton&Lange; Stamford, Connecticut: Fetal Sceletal Anomalies: 478-92.5.*

2. *Valenzano M,Paoletti R, Rossi A, Farinini D, Garlaschi G, Fulcheri E. Sirenomelia: Pathological features, antenatal ultrasonographic clues, and a review of current embryogenic theories. Hum Reprod Update 1999;5(1): 82-6.*

3. *Stevenson RE, Jones KL, Phelan MC at al. Vascular steal: the pathogenetic mechanism producing sirenomelia and associated defects of the viscera and soft tissues. Pediatrics, 78(3): 1986 Sep: 451-7.*

4. *Lynch SA, Wright C; Sirenomelia, limb reduction defects, cardiovascular malformation, renal agenesis in infant born to a diabetic mother. Clin Dysmorphol, 1997; 6(1):75-80*

5. *Twickler D, Budorick N, Pretorius D, Grafe M, Currarino G. Caudal regression versus sirenomelia: sonographic clues; J Ultrasound Med 1993;12(69): 323-30*

6. *Sirtori M, Ghidini A, Romero R, Hobbins JC. Prenatal Diagnosis of Sirenomelia; J Ultrasound Med, 8(2): 1989; 83-8*

7. *Valenzano M, Paoletti R, Rossi A, Farinini D, Garlaschi G, Fulcheri E. Sirenomelia. Pathological features, antenatal ultrasonographic clues and review of current embryogenic theories. Hum Reprod Update 1999;5(1): 82-6*

8. *Sancı M, Güler A, Meydan E, Dicle N, Başoğlu N, Sipahi Ç. Sirenomelia İki Olgunun Sunumu; Klinik Bilimler ve Doktor, Kasım 2001, Cilt 7, Sayı 6, 854-856*

9. *Lutz N, Meyrat BJ, Guignard JP, Hohlfeld J. Mermaid syndrome: virtually no hope for survival. Pediatr Surg Int. 2004;19:330-4*

10. *Kulkarni ML, Abdul Manaf KM, Prasannakumar DG, Kulkarni PM: Sirenomelia with radial displasia, Indian J Pediatr. 2004;71(5):447-9*