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Prenatal Diagnosis of Sporadic Phocomelia Associated with Diaphragmatic Hernia: A Case Report

Diyafragma Hernisine Eşlik Eden Sporadik Fokomelinin Prenatal Tanısı: Olgu Sunumu

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ABSTRACT

Phocomelia is a rare conjenital anomaly and seen in 4 million births. Many factors play a role in etiology of phocomelia such as environmental-genetic factors, teratogen exposure, amniotic band syndrome, thalidomide and also alcohol, cyclophosphamide and retinoic acid have been implicated in the pathogenesis. Phocomelia may be isolated or accompained by syndromes. A 35-years-old woman was admitted to our clinic for detailed ultrasonographic examination. Absence of some extremities and congenital diaphragmatic hernia was observed. There was no factors for phocomelia in family because of that, we suggested this case may be isolated. In this case, we present a case of isolated sporadic phocomelia with diaphragmatic hernia that was diagnosed and defined in our clinic.

Keywords: Diaphragmatic hernia; phocomelia; prenataly; sporadic; termination

ÖZET

Fokomeli, 4milyon doğumda bir görülen etiyolojisinde pek çok etkenin rol oynadığı nadir bir hastalıktır. Fokomeli etiyolojisinde çevresel-genetik faktörler, teratojen maruziyeti, amniyotik band sendromu gibi faktörler yer alabildiği gibi, izole olarak da gözlenebilmektedir. Talidomid dışında alkol, siklofosfamid ve retinoik asitte patogenezinde suçlanmıştır. Fokomeli izole olabileceği gibi Robert-SC fokomeli, Furhmann sendromu, Al-Awadi/Roos-Rthschild/Schinzel fokomeli gibi bazı sendromların parçası olabilir. 35 yaşında bir kadın ayrıntılı ultrasonografik inceleme için kliniğimize başvurdu.Sağ alt ekstemite total olarak sol ekstremitede ise distal kısım yokluğuna ek olarak diyafragma hernisi gözlenmiştir. Ailede fokomeli için hiçbir risk faktörü yoktu, bu yüzden bu vakanın izole olabileceğini öne sürdük. Sunduğumuz bu olguda kliniğimizde tanı ve teşhis edilen diyafragma hernisine ek olarak eşlik eden izole sporadik fokomeli olgusunu sunmak istedik.

Anahtar Kelimeler: Diyafragmatik herni; fokomeli; prenatal; sporadik; terminasyon

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Phocomelia (seal fish extremity) is a rare congenital anomaly in which the extremities are more or less absent in the long bones of the extremity and the hands and/or feet are directly attached to the body. Fetal extremity anomalies are seen at 20,000 births.[1,2] Limb defects may be isolated or accompanied by syndromes associated with multiple congenital anomalies.[3]

Early diagnosis of congenital extremity anomalies in the intrauterine period by ultrasonography is important. In this case, we want present a rare case of phocomelia that was diagnosed at 21st week of gestation in our clinic.

CASE REPORT

A 35-year-old case (Gravida 2 Parity 1) was admitted to our clinic on the 21st gestational week of gestation. Extremity anomaly was observed on detailed ultrasonographic examination. Absence of total lower right extremity and the distal part of left extremity and in addition the diaphragmatic hernia was observed during ultrasonography. There was no history of congenital anomaly in the parents and their families. And also no history such as relationship between parents, exposure to teratogenicity, smoking-alcohol and environmental impact. About the prognosis of the pregnancy was explained to family. Termination was present to family and pregnancy termination decision was taken to the direction of the wishes. 380 gr ex immature male fetus was delivered by vaginal birth after medical induction. Chromosome analysis and autopsy were suggested to family but they didnt accepted therefore cytogenetic analaysis could not be obtained. The left upper extremity of fetuses normally, totaly of the lower right extremity and the distal portion of the left extremity were not observed in the postmortem examination.

DISCUSSION

Phocomelia is a rare diasease seen in 4 million births and many factors play a role in the etiology (4). Some factors such as environmental-genetic factors, teratogen exposure, amniotic band syndrome may be observed in phocomelia etiology and also can be observed as isolated. An increase in the incidence of phocomelia has been reported after the use of thalidomide-based medication for sedation in pregnancy at the end of 1951s. After the use of this drug was restricted, there was a decrease in the number of phocomelia cases.[5,6] In additon to tolidomide, alcohol, cyclophosphamide and reti noic acid have been implicated in the pathogenesis. İn studies, phocomelia has been reported after use of retinoic acid in pregnant mice.[7] Phocomelia can be isolated, or it may be part of some syndromes such as Robert-SC phocomelia, Furhmann syndrome, Al-Awadi/Roos-Rthschild/ Schinzel phocomelia.[8]

Early ultrasonographic examination and karyotype analysing in the intrauterine period may be helpful in diagnosing of phocomelia cases. Phocomelia can be diagnose earliest at 12th gestational week by transvaginal ultrasonography during antenatal period, it has been reported in the literatüre.[9] In our case,phocomelia may be sporadic because of no history of teratogenic drug use, no relation between parents, absence of anomaly birth history in family and their relatives. As in our case, additional congenital anomalies can be accompanying to phocomelia such as congenital diaphragmatic hernia.

Early diagnosis of congenital anomalies can be made by detailed ultrasonographic examinations in the prenatal period. The extremities should be measure as comparative and severally in detailed ultrasonographic examination. As in our case, there may be absence a total and/or diffrent part of different extremities. Congenital anomalies that can accompanying to phocomelia as we are in our case should be remembered and detailed ultrasonographic examinations should be done. Families should be informed about the prognosis. Parents should be informed about the risk of recurrence in subsequent pregnancies, and early detailed ultrasound should be recommended for subsequent pregnancies.

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