A case of phocomelia associated with diaphragmatic hernia

Cömert E H, Şal H, Güvendağ Güven E S, Güven S
Karadeniz Technical University, School of Medicine, Department of Obstetrics, TRABZON, Turkey

Objective

Phocomelia is a rare congenital anomaly seen in 4 million births. Many factors play a role in the etiology of phocomelia such as environmental-genetic factors, teratogen exposure and amniotic band syndrome. Thalidomide, alcohol, cyclophosphamide and retinoic acid have also been implicated in the pathogenesis. Phocomelia may be isolated or accompanied by syndromes. We present a case of isolated sporadic phocomelia with diaphragmatic hernia that was diagnosed and defined in our clinic.

Methods

This is a case report.

Results

A 35-year-old patient (Gravida 2 Parity 1) was admitted to our clinic at 21 weeks of gestation. A detailed ultrasound examination was performed, which showed total absence of the lower right extremity, absence of the distal part of the lower left extremity and diaphragmatic hernia. No family history of congenital abnormalities was reported. There was no consanguinity, maternal exposure to teratogens, smoking or alcohol, and no environmental impact. The prognosis of the pregnancy was explained to family. Termination was offered and performed. A 380 g immature male fetus was delivered by vaginal birth after medical induction. Chromosomal analysis and autopsy were suggested to the family and were declined; therefore cytogenetic analaysis could not be obtained. The left upper extremity of the fetus was normal, but the lower right extremity and the distal portion of the left lower extremity were not observed in the postmortem examination.

Conclusion

In this case, phocomelia may be sporadic because of no history of teratogenic drug use, no consanguinity and no family history of congenital abnormalities. As in our case, additional congenital anomalies can be accompanying phocomelia such as congenital diaphragmatic hernia. Early diagnosis of congenital anomalies can be made by detailed ultrasound examination in the prenatal period, including measurment of the extremities. As in our case, there may be absence of total and/or different parts of upper and lower extremities. Congenital anomalies that may be accompanying phocomelia should be considered. Families should be informed on the prognosis and risk of recurrence and early detailed ultrasound scans should be recommended for subsequent pregnancies.