Sirenomelia (the Mermaid Syndrome): Report of Three Cases

Sirenomeli (Mermaid Sendromu) Üç Olgu Sunumu

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ABSTRACT Sirenomelia is a rare and lethal malformation. It has been considered as an extreme form of caudal regression syndrome. This sporadic defect occurs in approximately 1 in 60000 newborn infants and has male predominance. Maternal diabetes mellitus is accepted to be an important predisposing factor and urogenital, gastrointestinal, cardiopulmonary and central nervous system defects are seen together with the anomaly. In this case presentation, we reported three cases of sirenomelia with multiple organ malformations. Variety of the associated anomalies and the pathological features of these rare cases may enlighten diagnosis of similar cases.

Key Words: Ectromelia; abnormalities

ÖZET Sirenomeli, nadir ve letal bir malformasyondur. Kaudal regresyon sendromlarının az görülen bir formu olduğu düşünülmektedir. Daha çok erkek cinsiyette izlenen bu sporadik defekt 60000 doğumda bir görülür. Maternal diyabet en önemli predispozan faktör olup ürogenital, gastrointestinal, kardiyopulmoner ve santral sinir sistemi defektleri bir arada görülür. Biz makalemizde birden fazla organ malformasyonu gösteren 33, 23, 14 haftalık üç sirenomeli olgusunu benzeri olgulara ışık tutacak patolojik özellikleri nedeniyle sunuyoruz.

Anahtar Kelimeler: Ektromeli; anomali

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Sirenomelia is a rare and lethal polymalformation. Many authors consider sirenomelia as an extreme form of caudal regression syndrome.[1,2] This sporadic defect occurs in approximately 1 in 60000 newborn infants; it has male predominance. It is seen together with urogenital, gastrointestinal, cardiopulmonary and central nervous system defects. Maternal diabetes mellitus is considered as an important predisposing factor. Genetic predisposition and vascular hipoperfusion have been suggested as other possible causative factors.[3,4]

CASE REPORT

Case one is a 33 weeks fetus born from a 21 years old multiparous mother. He died 2 minutes after his birth. According to the last menstrual date, the fetus was 33 weeks old but the embryogenetic age is established by correlation of crown-rump length was consistent with 37 weeks. There was no maternal diabetes mellitus or consanguineous marriage. Ultrasonography was not diagnostic because of olygohydramniosis. Provacated abortus was performed

depending on the permanent nonstress test results. Nasal root depression, facial edema, ear deformation, flexion contracture on left hand, umbilical cord edema and nail base congestion was found in external examination. The most important finding was pruned shape of the lower extremities (Figure 1).

Internal examinations revealed sacral, gallbladder and renal agenesis. Anal atresia was also present.

Case two is a 23 weeks fetus born from a 24 years old primigravid mother. Maternal diabetes was manifest. Ultrasonography showed marked oligohidramnios and renal agenesis. Only one femur was detected in ultrasonography. The postmortem X-ray examinations revealed lack of radius in the right side where the ulna was angulated. The fused lower extremities tapering in distal end was contracted and contained a bony nidus.

Autopsy findings were similar to case one. Additionally, corpus callosum agenesis, liver and pulmonary hypoplasia and abnormal lobulation of lungs were found. External genitalia were absent (Figure 2). A rudimentary tissue dissected in pelvic area has shown to be a testis on histopathologic examination.

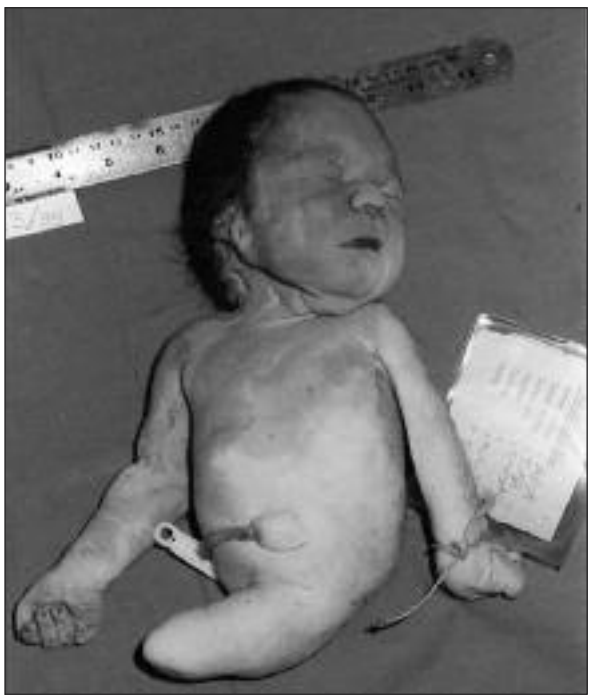

FIGURE 1: Case one with extreme rudimentary lower extremities.

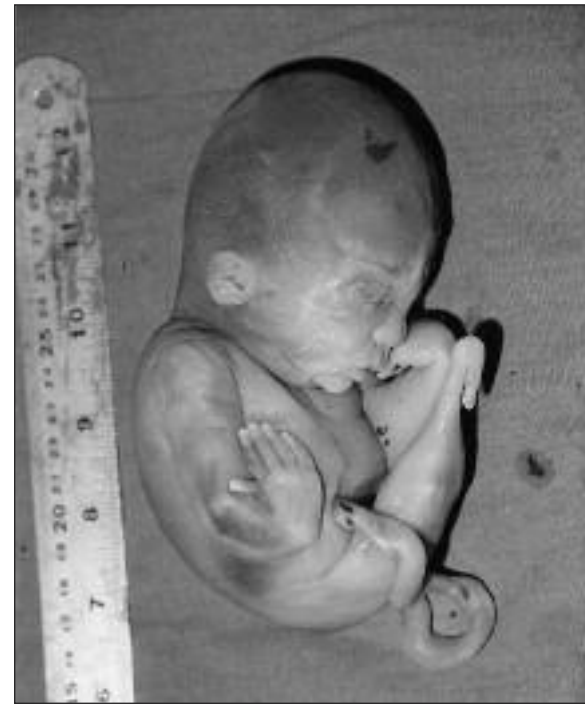

FIGURE 2: Note to the upper extremities' contracture in case two.

Case three is a 14 weeks fetus spontaneously aborted. Ultrasonography was not performed. Macroscopic examination of the macerated fetus included midface defect (cleft lip), nuchal edema, flexion contracture and oligodactily of bilateral hands. The main feature was the fusion of the lower extremities (Figure 3). Autolysis and degenerations interfered the evaluation of organs, but we found large bowel hipoplasia, renal, gallbladder and genitalia agenesis and anal atresia. Dissection of the caudal part of the fetus revealed a single femur and tibia with a rudimentary foot that had three fingers. Umblical cords of all fetuses were normal. Placental examination was not possible because of the missing delivery of the material from the obstetric clinic. Genetic counseling that was performed to case 1 and 2 showed no particular chromosomal abnormalities.

CONCLUSION

Sirenomelia manifests with fusion, rotation, hypotrophy or atrophy of lower limbs in combination with severe urogenital malformations and is usually a lethal disorder.[5] In English literature there are only five living sirenomelic infants.[6] Etiology and patho-

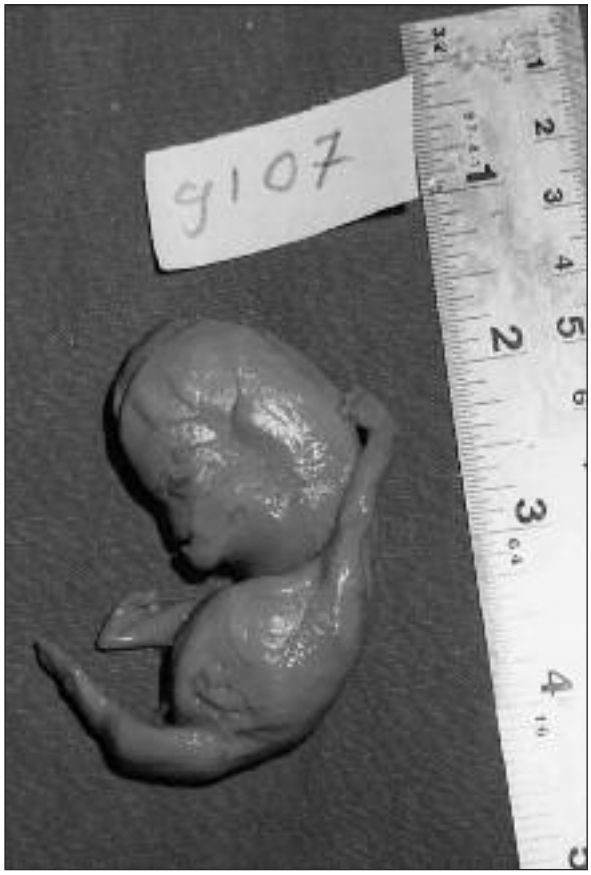

FIGURE 3: Macerated fetus (case 3); posterior nuchal edema, oligodactily.

genesis of the disease has been poorly understood. An early alteration of the embryological vascular network that damages the caudal mesoderm is thought to lead to arrested development of the lower limbs and the other affected organs.[7-9]

Our cases had most of the expected anomalies. Besides these anomalies, we also detected some relatively rare ones such as upper extremity anomalies, caudal agenesis and midface defect. Single artery in the umbilical cord is a common finding in the sirenomelia cases. However, umblical cords of all our fetuses were normal.

Bilateral renal agenezis appears to be a part of several malformation syndromes. It has been frequently reported in sirenomelia cases. Perinatal ultrasonographical diagnosis should be obtained as early as possible and anhydramniotic cases with maternal diabetes need special care. Color Doppler is helpful to confirm the diagnosis. Coexisting abnormalities should be taken into consideration in autopsy.

Here, we reported three cases of sirenomelia with multiple organ malformations.

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