PP-06 Unicornuate uterus accompanied by unilateral tubal-agenesis in pregnancy a case presentation

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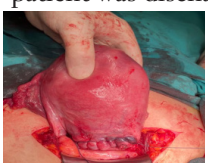
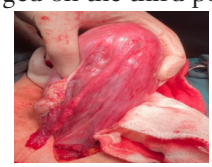
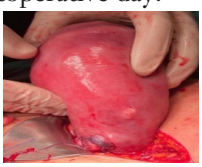
DOI:10.59215/prn.23.031supp021

Objective: Fusion of the Müllerian ducts occurs during weeks 6-11 of pregnancy. Disruptions in this fusion process can lead to Müllerian duct anomalies. Among these anomalies, unicornuate uterus is the most common and is associated with pregnancy complications such as preterm labor. In this case presentation, we aim to present a patient with a diagnosis of unicornuate uterus and pelvic kidney, accompanied by unilateral tubal-ovarian absence, who gave birth at our clinic.

Case: A 26-year-old, G2P0A1 woman presented to the clinic at 33+4 weeks of gestation with high blood pressure. Upon reviewing the patient's medical history, it was revealed that her first pregnancy ended in a miscarriage during the first trimester, and pelvic ultrasound detected an ectopic pelvic kidney. Further inquiry into the patient's family history indicated that her mother also had an ectopic pelvic kidney. The patient was being followed up in our clinic with the threat of preterm delivery due to high blood pressure and contractions in the non-stress test .Considering the potential risk of preterm birth, the patient was administered two doses of betamethasone 24 hours apart for lung maturation. As high blood pressure and uterine contractions persisted during follow-up, a decision was made for delivery. The patient underwent a cesarean section. A live baby weighing 2180 grams with an 8-10 APGAR score and umbilical cord blood gas pH of 7.40 was delivered. Intraoperatively, a unicornuate appearance of the uterus without a rudimentary horn was observed, and the left fallopian tube and ovary were not seen. No complications arose during the operation. The patient was discharged on the third postoperative day.

Conclusion: Mullerian duct anomalies are observed in approximately 1-5% of the general population. The prevalence increases to around 13-25% in patients with a history of recurrent miscarriages. Unicornuate uterus is the most commonly encountered type among müllerian duct anomalies. The variation without a rudimentary horn accounts for approximately 35% of all unicornuate uteri. Kidney anomalies accompany approximately 40% of patients with unicornuate uterus. Patients with unicornuate uterus are often asymptomatic, and ultrasound alone might not be sufficient for diagnosis. Complications related to unicornuate uterus primarily manifest during pregnancy. Pregnant individuals with unicornuate uterus are at an increased risk of preterm birth, first and second-trimester miscarriages, intrauterine fetal death, and cesarean delivery. Pelvic kidney generally remains asymptomatic and is incidentally diagnosed. Its prevalence is approximately 1-10/10,000. In cases where a pelvic kidney is detected in female patients, the possibility of müllerian duct anomalies should be considered, and patients should be informed about potential pregnancy complications, especially in the context of such anomalies.

Keywords: Unicornuate uterus, pelvic kidney, preterm birth

References

1. (Caserta, Mallozzi et al. 2014)
2. (Reichman, Laufer, & Robinson, 2009)

PP-07 TAR Syndrome case report

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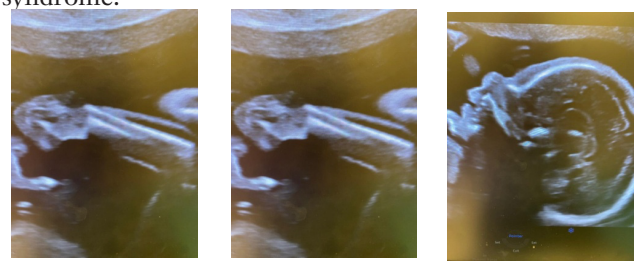
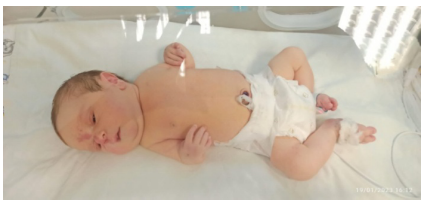
DOI:10.59215/prn.23.031supp022

Objective: "Thrombocytopenia - absent radii Syndrome" (TAR) is characterized by hypo-megakaryocytic thrombocytopenia and bilateral absence of the radius bones despite the presence of both thumbs. It is rare and follows an autosomal recessive inheritance pattern. In individuals with TAR syndrome, skeletal, cardiac, gastrointestinal, hematological, renal, and genital abnormalities can also accompany the condition. Skeletal anomalies include varying degrees of ulnar hypoplasia, as well as hypoplasia of phalanges and carpal bones. Thumbs are always present. Thrombocytopenia improves with age and usually resolves by school age. It is associated with a deletion in the proximal region of the gene locus 1q21.1. The syndrome, first described in 1929, has an estimated prevalence of 0.42/100,000. In this case presentation, we aim to discuss the diagnosis and management of TAR syndrome.

Case: A 28-year-old woman, gravida 4, parity 1, abortion 1, was placed under routine pregnancy surveillance. During first-trimester screening, the absence of bilateral forearm segments and an increased nuchal translucency (3.8mm) were detected, prompting further monitoring. Amniocentesis was performed for the patient. In the second trimester, an obstetric ultrasound revealed the absence of the right forearm during upper extremity examination. Only a single bone consistent with the ulna was observed on the left side. Throughout the ultrasound examination, limited hand movement, significant wrist extension, and flexion contractures at the knees were noted. Additionally, there was pes equinovarus observed on the left foot. Differential diagnoses considered TAR syndrome, Roberts syndrome, and Fanconi syndrome. The amniocentesis yielded a normal fetal karyotype. At 40+1 weeks gestation by last menstrual period, the patient underwent a normal vaginal delivery with a birth weight of 3070g (19th percentile) and an 8-9 APGAR score. In the

postnatal period, the infant was admitted to the neonatal intensive care unit due to a platelet count of 12000 and the presence of petechiae on the body. Absence of ulna and radius was observed on the right arm, while only a single bone consistent with ulna was seen on the left side. Postnatal microarray analysis was consistent with TAR syndrome.





Conclusion: TAR syndrome is an exceedingly rare condition. Prenatal diagnosis can be achieved through ultrasound evaluation of limb anomalies at around 16 weeks of gestation, combined with complete blood count and genetic analysis via cordocentesis. In neonates, when encountering thrombocytopenia and hemolytic anemia, TAR syndrome should be considered as a differential diagnosis.

Keywords: TAR syndrome, absent radius, thrombocytopenia, hemolytic anemia, phocomelia.

References

1. **Hall, J. G. Thrombocytopenia and absent radius (TAR) syndrome. Journal of medical genetics. 24, 1987**
2. **Bozkurt Turhan, A., Barsan Kaya, T., Tekin, A. N., Anıl, H., Özdemir, C., & Akşit, M. A. Trombositopeni-Radius Yokluğu Sendromlu Yenidoğanda Hemolitik Anemi ve İnek Sütü İntoleransı Birlikteliği. Journal of Pediatric Disease/Cocuk Hastaliklari Dergisi. 2017**

PP-08 Meckel Gruber Syndrome a case report

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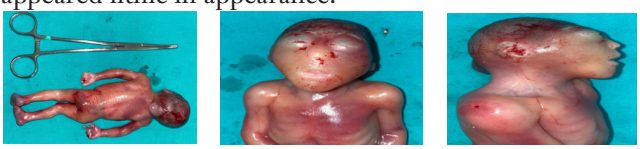
DOI:10.59215/prn.23.031supp023

Objective: Meckel-Gruber Syndrome is a rare fetal anomaly characterized by multiple anomalies that are inherited in an autosomal recessive manner and are incompatible with life. In our clinic, we aimed to present the findings of a fetus with Meckel-Gruber Syndrome, for which we conducted pregnancy termination.

Case: A 37-year-old patient with a history of G9P8Y7PPEX1 was referred to our center at 18+2 weeks of gestation due to cranial anomalies identified through a structural anomaly scan. Obstetric ultrasound revealed a single viable fetus consistent with 18+1 weeks of gestation. In intracranial evaluation, the fetus exhibited a single cavity in the lateral ventricles, thalamic fusion, absence of the cavum septum pellucidum, and alobar holoprosencephaly. A cystic hygroma with septations measuring 18x13 mm was observed in the fetal neck. Microcephaly and significant hypertelorism were also present. Ultrasonographic findings were suggestive of Meckel-Gruber Syndrome. Following the decision made by the board and with the consent of the family, the pregnancy termination was carried out. The fetus was aborted vaginally and weighed 110 grams, measuring 18 cm in length. Macroscopic examination of the fetus revealed micrognathia, hypertelorism, flattened nasal bridge, low-set ears, and encephalocele, consistent with female external genitalia. Autopsy examination of the fetus indicated an enlarged liver with absence of the gallbladder, spleen, and pancreas. Other internal organs appeared lithic in appearance.



Conclusion: Meckel-Gruber Syndrome is a rare fetal anomaly with an autosomal recessive inheritance pattern, occurring at a frequency of 1 in 13,250 to 140,000 births. It follows a severe and lethal course. While the classic triad includes cystic renal dysplasia, encephalocele, and polydactyly, the syndrome can also manifest with additional anomalies. Differential diagnoses should include Trisomy 13 and Smith-Lemli-Opitz syndrome, given their similar clinical presentations. Careful consideration is necessary in making a differential diagnosis due to the overlapping features with these two conditions. Definitive diagnosis requires autopsy, as karyotype analysis might yield normal results. The recurrence rate is 25%, underscoring the importance of prenatal diagnosis and monitoring. Mortality associated with this syndrome is 100%. Families should be counseled about the risk of mortality in the current pregnancy and the possibility of recurrence in subsequent pregnancies. First-trimester ultrasound evaluation between 11 and 14 weeks is strongly recommended for subsequent pregnancies.

Keywords: Meckel-gruber, encephalocele, polydactyly

References

1. **(Alexiev, Lin, Sun, & Brenner, 2006)**
2. **(Hartill, Szymanska, Sharif, Wheway, & Johnson, 2017)**