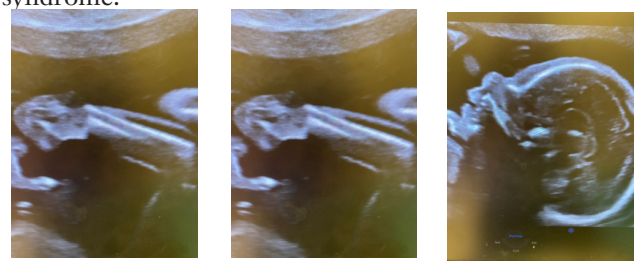
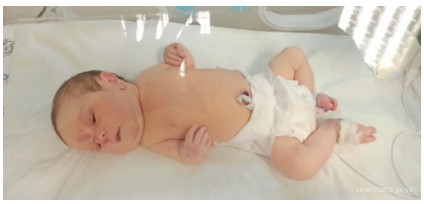
postnatal period, the infant was admitted to the neonatal intensive care unit due to a platelet count of 12000 and the presence of petechiae on the body. Absence of ulna and radius was observed on the right arm, while only a single bone consistent with ulna was seen on the left side. Postnatal microarray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

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PP-08 Meckel Gruber Syndrome a case report

Serem Kel[1], Nur Kuşcu[1] ,Süreyya Demir[2], Mehmet Nuri Duran[1], Bülent Demir[1]
[1]Canakkale 18 Mart University Hospital, Çanakkale, Türkiye
[2]Canakkale Mehmet Akif Ersoy State Hospital, Çanakkale, Türkiye

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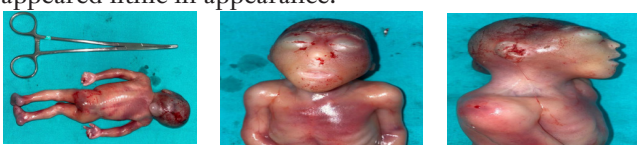
Objective: Meckel-Gruber Syndrome is a rare fetal anomaly characterized by multiple anomalies that are inherited in an autosomal recessive manner and are incompatible with life. In our clinic, we aimed to present the findings of a fetus with Meckel-Gruber Syndrome, for which we conducted pregnancy termination.

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

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