OP-01 Primary microcephaly cases with molecular genetic basis

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Objective: Primary microcephaly (MCPH, for "microcephaly primary hereditary") is a disorder of brain development that results in a head circumference more than 3 standard deviations below the mean for age and gender. Our aim is to evaluate ultrasonographic findings, clinical features and molecular analysis of two cases of primary microcephaly

Case: Two cases at 26 and 23 weeks referred to our clinic due to small BPD and HC measurements for gestational age at second trimester screening ultrasonography

Discussion: The ongoing discovery and research on MCPH genes and their animal models will increase our knowledge in this rare non-progressive neuropediatric disorder. MCPH genes might play an essential role during evolution, and therefore, they are suitable candidates for studying normal brain development

Conclusion: All cases of microcephaly should be referred to the medical genetics service to confirm or rule out the diagnosis and to provide genetic counseling.

Keywords: Brain development, genetic, microcephaly

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OP-02 Serum prolactin levels in women with threatened miscarriage a prospective case-control study

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Objective: The aim of this study is to compare the serum prolactin levels of women with threatened miscarriage (TM) and women with uncomplicated pregnancies.

Results: There were statistically significant differences between groups in serum prolactin levels, significantly higher in group I (47.7 ± 42.6 vs 29.64 ± 13.01, respectively; p=0.003). Mean serum prolactin levels of women were increasing directly with gestational age (p=0.005)

	Threatened miscarrige (n:226)	Control Group (n: 215)	p
Prolactin (n/mL)	47.7 ± 42.6	29.64 ±13.01	0.003
TSH	1.75 ± 0.99	1.84 ± 1.04	0.356

Conclusion: Our results demonstrate that serum prolactin levels differed in women with TM in early weeks of pregnancy. Prolactin may be a future biomarker in predicting the outcome of TM.

Keywords: Biomarker, miscarriage, prolactin

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OP-03 Osteogenesis imperfecta type 3 with COL1A1 mutation prenatal diagnosis and management

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Objective: Osteogenesis Imperfecta (OI) is a rare genetic disease group consisting of defects in collagen synthesis. This disease is characterized by alteration of connective tissue structure of exposed patients, usually caused by mutation of type I collagen. Altered connective tissue structure leads to low bone mineral density, defective bone structure and strength, and multiple fractures, which are usually obtained after low-impact trauma.[1] It is a rare disease that is seen in 0.5-1 per 10000 live births in general.[2] There are several types of OI, type I being the mildest form of OI, which is usually asymptomatic, sometimes with scoliosis due to compression fractures of the vertebrae in adults. Type II is the most severe form and is generally incompatible with life, while type III is

the most severe form in surviving patients.[2-4]

OI is inherited in an autosomal dominant manner and heterozygous mutation in one of the COL1A1 or COL1A2 genes is the cause in more than 90% of cases.[2,5] Type III constitutes 5% of all OI cases.[5] With this case report, we aimed to present the antenatal findings and clinical management of an OI type III case diagnosed during the antenatal period.

Methods: Computer-based and ultrasonography records of a case of osteogenesis imperfecta who applied to the Perinatology outpatient clinic of Prof. Dr. Cemil Taşçıoğlu City Hospital at the 8th week of pregnancy were retrospectively scanned from the hospital system and the history of the ultrasonography device. Fetal ultrasonography examination was performed using Mindray Resona 7 device and its 1.2-6 MHz convex abdominal probe. Ultrasonography findings and patient history were noted.

Case: A 40-year-old primagravid patient who became pregnant by in vitro fertilization first applied to our clinic for an 11-14 week ultrasound examination at 14 weeks of age. In the patient's anamnesis, it was learned that he had a brain aneurysm 2 years ago and had hypothyroidism. The result of the non-invasive prenatal test performed by the patient in an external center was reported as normal. In the ultrasound examination of the patient at the 14th week, hypomineralization was observed in the skull bones (Figure 1). It was observed that the head shape was deformed by the compression of the ultrasound probe. Osteogenesis imperfecta, hypophosphatasia and achondrogenesis were considered as the differential diagnosis in the patient. The patient was informed and called for follow-up 2 weeks later. In the ultrasound examination performed at the 16th week, hypomineralization was observed in the skull bones, the mineralization of the vertebral column was normal, shortness in all long bones, contractures in the hands and feet, and short costa were observed (Figure 2). The family was informed about amniocentesis and amniocentesis was performed. A heterozygous variant in the COL1A1 gene was detected in the fetus in whole-exome sequencing analysis, and this variant was reported as a pathogenic variant associated with Osteogenesis Imperfecta Type 3. The family decided to terminate. Termination was performed at 20 weeks (Figure 3,4). The patient, who had no bleeding or complaints after termination, was discharged with recommendations.

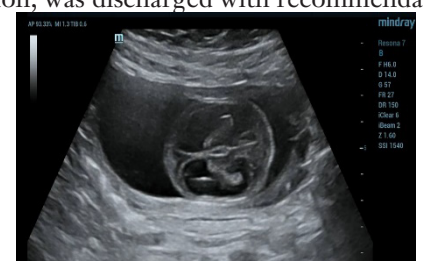

Fig 1. Hypomineralitation of skull bones during ultrasonoghrapic examination

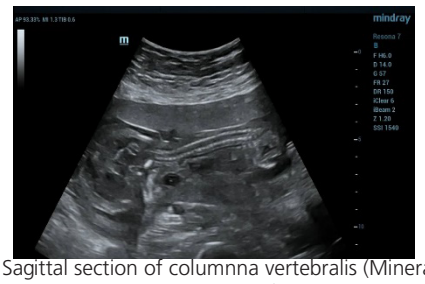

Fig 2. Sagittal section of columnna vertebralis (Mineralization of columnna vertebralis as it should be.)

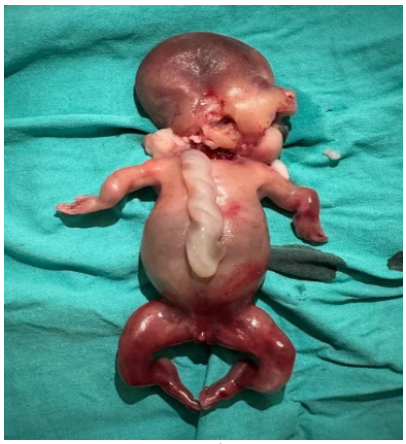

Fig 3. Post-termination view of the case

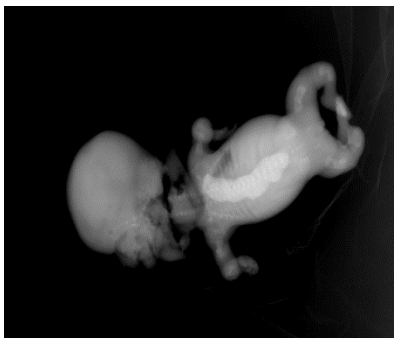

Fig 4. X-ray of the case after termination

Results: OI is a persistent connective tissue disease with autosomal dominant inheritance. OI Type III is shown as the most severe form in survivors.[2] Although there are treatment options such as pamidronate, zolidronate or risedronate in the postnatal period, these patients are faced with nearly 100 bone fractures and the necessity of many operations both during birth and until adolescence. For these reasons, the family should be informed in detail about the prognosis and possible risks in patients diagnosed with OI Type III, who were diagnosed with invasive diagnostic tests during the antenatal period, and the decision to continue or terminate the pregnancy should be made and managed together.

Keywords: Osteogenesis imperfecta, COL1A1, collagen, bone mineralization, fetal anomalies

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OP-04 A study of fetal sacrococcygeal teratoma cases perinatal and postnatal evaluation

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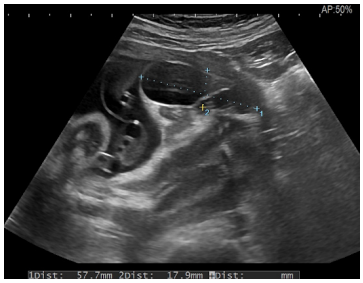
Objective: This study aimed to evaluate the perinatal and postnatal outcomes of fetuses diagnosed with sacrococcygeal teratoma during the antenatal period.

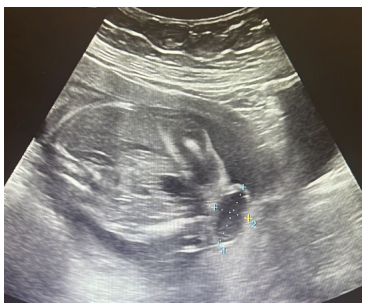
Methods: The records of patients who presented to our clinic between 2020-2023 and were diagnosed with fetal sacrococcygeal teratoma were retrospectively reviewed. Patient data including gestational age at diagnosis and delivery, teratoma size and type, presence of additional structural anomalies, genetic examination results, as well as the presence of polyhydramnios and hydrops, were collected. The sacrococcygeal tumors were classified into types 1 through 4. A multidisciplinary perinatology council team assessed the cases.

Results: The study included nine patients diagnosed with sacrococcygeal teratoma via ultrasound and confirmed postpartum. Of the sacrococcygeal teratomas, four were type 1, four were type 2, and one was type 3.Of the nine fetuses, six survived, while three died within the first week of life without surgery. Surgical intervention was performed during the first week of life for the surviving neonates. The teratoma types in the deceased patients were type 3 and type 2.

Conclusion: Although fetal sacrococcygeal teratoma cases are rare, successful results can be obtained with an early and accurate prenatal diagnosis, appropriate surgical intervention and frequent follow-up approach. The management of such cases is a complex process that requires a multidisciplinary approach. The size, content, growth rate and type of the tumor are the determining factors in terms of proper treatment and management planning.

Keywords: Ultrasound, perinatal diagnosis, sacrococcygeal teratoma





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OP-05 Prenatal diagnosis of left ventricle aneurysm

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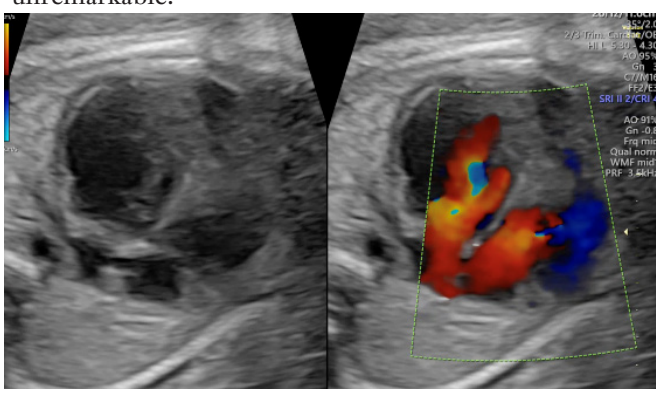
Objective: Ventricular aneurysm of the fetal heart is a rare cardiac anomaly that occurs after a defect in the myocardium. In this case report; Prenatal diagnosis and management of ventricle aneurysm is discussed.

Case: The patient was presented for routine screening at 22 weeks of gestation, and the prenatal history was unremarkable.



Discussion: Ventricular aneurysm of the fetal heart is a rare cardiac anomaly that occurs after a defect in the myocardium.Unlike a diverticulum, it is larger and the contractility of the heart wall is impaired.It is more common in the left than in the right ventricle.

Conclusion: Left ventricular aneurysm is one of the rare cardiac anomalies that can be diagnosed in the prenatal period.The size of the lesion, impaired cardiac function and arrhythmia are the criteria for a poor prognosis.