### PP-15 Case report of isolated cardiac rhabdomyoma

Cagdas Nurettin Emeklioglu<sup>1</sup>, Enes Halk<sup>2</sup>, Mirac Ozalp<sup>3</sup> <sup>1</sup>Karabuk University Trainning and Research Hospital, Department of Obstetrics and Gynecology, Karabuk, Türkiye

<sup>2</sup>Prof. Dr. Cemil Tascioglu City Hospital, Department of Obstetrics and Gynecology, Istanbul, Türkive

<sup>3</sup>Prof. Dr. Cemil Tascioglu City Hospital, Department of Obstetrics and Gynecology, Perinatology Clinic, Istanbul, Türkiye

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**Objective:** Although fetal primary cardiac tumors are rare, they are usually detected during fetal echocardiography. The vast majority of primary cardiac tumors are benign, with rhabdomyomas being the most common cardiac tumors in the prenatal period, followed by teratoma, fibroma, and hemangioma. Cardiac rhabdomyomas are usually numerous, depending on the location, they can cause hemodynamic disorders and arrhythmias in the antenatal and early postnatal period. [1] Cardiac rhabdomyomas are frequently observed together with tuberous sclerosis. Other components of the tuberous sclerosis complex are tubercles and nodules in the brain, spots on the skin and renal angiomyolipomas, and fetuses with cardiac rhabdomyoma should be carefully screened for these anomalies. [1,2] With this case report, we aimed to share the clinical presentation and results of a cardiac rhabdomyoma case that we followed in our clinic.

Methods: Computer-based and ultrasonography records of an isolated cardiac rhabdomyoma case, who applied to Prof. Dr. Cemil Taşçıoğlu City Hospital Gynecology and Obstetrics Clinic - Perinatology outpatient clinic at the 34th week of pregnancy, were scanned retrospectively from the hospital software 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.

Case: The 30-year-old patient, who had no additional features in her obstetric history except for one abortion, was referred to our clinic with the suspicion of a cardiac mass. She was at 34 weeks of gestation according to the date of her last menstrual period. Fetal echocardiography revealed multiple rhabdomyomas, the largest of which was 12\*6.6 mm in the left ventricle, 10\*9 mm in the right ventricle, and 15\*10 mm in the apex of the interventricular septum (Figure 1-2). Although there was a rhabdomyoma of 7.7\*4.6 mm in front of the aortic valve at the exit of the left ventricle, the vessel sizes were normal in 3 vesselstrachea sections. No nodular image was observed in the cranial examination. Examination of the fetal kidneys was normal in size and anatomically. Umbilical artery, bilateral uterine artery and middle cerebral artery Doppler flows were normal. The patient and her husband did not want to have an invasive diagnostic test in the prenatal period for the differential diagnosis of tuberous sclerosis. Pediatric cardiovascular surgery was consulted about the patient, and postnatal emergency intervention was not considered as the current pathology did not cause any outlet obstruction. During the examination, on the patient's description of itching on the hands and feet, a diagnosis of intrahepatic cholestasis of pregnancy was

made in the further examination. One week later, due to pharmacotherapy-resistant pruritus, high total serum bile acids and unreliable NST, a live female baby was delivered at 35 weeks. The baby's 1st and 5th minute APGAR scores were 7 and 8, respectively. In the first examination, it was learned that there was no pathological cardiac listening finding. In the first echocardiographic examination, a large number of hyperechoic masses thought to be rhabdomyomas, the largest of which was 16\*18mm in diameter, were detected in the intracardiac region, which did not cause heart failure, ventricular tachycardia, valve stenosis, and left and right ventricular outflow tract stenosis. No intervention was required. It was learned that rhabdomyomas spontaneously regressed without the need for any treatment in the pediatric cardiology follow-up of the baby after birth.

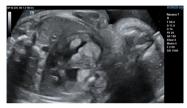


Fig 1. Prenatal cardiac rhabdomyomas



Fig 2. Rhabdomyoma adjacent to the pulmonary valve

**Results:** Rhabdomyomas may be in the form of multiple intramural lesions in the right and left ventricles, or they may be located in the interventricular septum or atrium. Although they can be seen as a single mass, they often involve the heart multiple or diffusely. Fetal echocardiography is a sensitive method for detecting cardiac tumors. Especially during the second-trimester ultrasonography, 4-chamber and 3-vessels-trachea sections are valuable in detecting cardiac tumors. Tuberous sclerosis accompanies 30-50% of cardiac rhabdomyomas, which is why these patients deserve a detailed evaluation and follow-up. [1,2]

Conclusion: Cardiac rhabdomyomas may be symptomatic depending on the size and location of the tumor, although by their nature they tend to regress spontaneously after the end of the hormonal effect of pregnancy after delivery. For these reasons, the growth status of the tumor should be evaluated in antenatal follow-ups and it should be recommended to give birth in a tertiary center by deciding with the family.

Keywords: Rhabdomyoma, cardiac tumors, tuberous sclerosis

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## PP-16 The importance of early diagnosis in maternal syphilis

Sevim Tuncer Can<sup>1</sup>, Atalay Ekin<sup>1</sup>
<sup>1</sup>University of Health Sciences İzmir Tepecik Training and Research Hospital, Department of Gynecology and Obstetrics, İzmir, Türkiye

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**Objective:** Infection is of particular concern during pregnancy because of the risk of transplacental transmission to the fetus. In this case report; Prenatal diagnosis and management of syphilis is discussed.

Case: A 20-year-old G1P0 patient at 12w5d gestational week was refered to our department with a positive Venereal Disease Research Laboratory (VDRL) test result. His history was obtained and systemic examination was performed. There was no recent acute febrile infection or vaccination history. No additional systemic disease was observed. There were no signs of infection including maculopapular rash, genital ulcer and oral aphthae. The patient had symptoms of groin pain, burning during urination, chills and shivering. Anti-HIV test result was negative. T. pallidum particle agglutination test (TPPA) was used as a confirmatory test because of the possibility of false positive screening test result. The diagnosis of syphilis was confirmed with a positive TPPA test result (26.32 s/co) and 2.4 million units of Penicillin G benzathine was initiated intramuscularly.

**Discussion:** Treponema Pallidum easily infects the placenta and causes amniotic fluid infection, placentomegaly and fetal anemia, thrombocytopenia, ascites, hydrops and intrauterine growth retardation by transplacental transmission. Manifestations of congenital infection are affected by gestational age, maternal syphilis status, maternal treatment, and fetal immunological response.

**Conclusion:** Screening all pregnant women for syphilis infection at the first trimester is recommended. Benzathine Penicilline G is effective in preventing transmission of infection to the fetus in most settings.

Keywords: Pregnacy, prenatal management, syphilis

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# PP-17 A case of Meckel-Gruber syndrome diagnosed in the first trimester

Büsra Berfin Polat<sup>1</sup>, Rauf Melekoglu<sup>1</sup>

<sup>1</sup>Inonu University Faculty of Medicine, Department of Obstetrics and Gynecology, Malatya, Türkiye

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**Objective:** Meckel-Gruber syndrome (MGS), an autosomal recessively inherited hereditary syndrome from the group of ciliopathies, is characterized by occipital encephalocele, large polycystic kidneys and postaxial polydactyly, resulting from the involvement of multiple genes and therefore has 15 phenotypes, occurring in 1 in 13,250-140,000 live births worldwide. Prenatal ultrasonography is the best available method to diagnose MGS. In this report, we aimed to present the prenatal diagnosis of a first trimester MGS case.

Case: A 25-year-old patient with gravida 3, parity 1, 14 weeks and 1 day gestation according to the last menstrual period was admitted to the prenatal diagnosis and treatment unit of our clinic for a first trimester screening test. In the ultrasonographic examination of the patient who had a history of MGS in her previous pregnancy, fetal cranial evaluation revealed acrania, exencephaly, anencephaly sequence (figure 1) and open spina bifida anomaly. Fetal abdominal evaluation revealed bilateral cystic kidneys (figure 2). Fetal extremity evaluation revealed no clear evaluation for postaxial polydactyly. The family was informed in detail about the possible poor fetal/neonatal prognosis of the fetus, which was primarily diagnosed as MGS according to the sonographic findings, and invasive prenatal diagnostic testing and pregnancy termination were presented as options. The family decided to terminate the pregnancy without invasive prenatal diagnostic testing. After termination of pregnancy, genetic examination of the abortion material was requested. Macroscopic examination of the abortion material confirmed prenatal findings (figure 3). The family was referred to the genetics outpatient clinic.



