

## References

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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 referred 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:** Pregnancy, 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.

