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PP-16 The importance of early diagnosis in maternal syphilis

Sevim Tuncer Can[1], Atalay Ekin[1]

[1]University of Health Sciences İzmir Tepecik Training and Research Hospital, Department of Gynecology and Obstetrics, İzmir, Türkiye

DOI:10.59215/prn.23.031supp031

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 syphılıs 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[1], Rauf Melekoglu[1]

[1]Inonu University Faculty of Medicine, Department of Obstetrics and Gynecology, Malatya, Türkiye

DOI:10.59215/prn.23.031supp032

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.

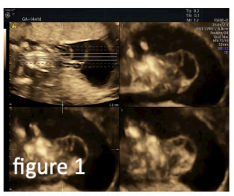
figure 1

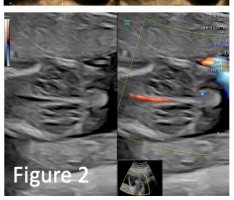
Figure 2

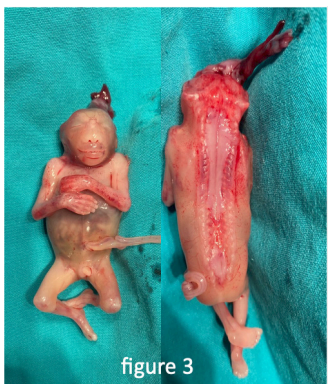
figure 3

Discussion: Prenatal features of MKS, such as postaxial polydactyly, encephalocele, and polycystic kidneys, are often profound and easily detectable in the first trimester. The findings of a large population-based review that estimated the incidence of typical symptoms were as follows: encephalocele, 83.8%; polydactyly, 87.3%; and cystic kidney disease, 97.7%.[2] Therefore, targeted prenatal diagnosis of MKS is usually triggered by these findings. However, the presence of encephalocele is not specific to MKS. Only 21% of fetuses diagnosed with prenatal encephalocele will have MCS,[3] and the same is true for polycystic kidney finding. The majority of confirmed hereditary cystic kidney disease detected prenatally is autosomal recessive polycystic kidney disease (ARPKD), diagnosed in 81% of cases. Meckel-Gruber syndrome was found in only 8% of such cases.[5]

Conclusion: MGS is a rare autosomal recessive condition with a mortality of 100%; diagnosis is possible antenatally even in the first trimester of pregnancy by prenatal sonographic examination. Given its mortality, early diagnosis of MGS and other such lethal anomalies has a significant impact on family counseling, especially when it comes to termination of pregnancy. Early prenatal diagnosis and genetic counseling are important in the management of this case with a first trimester prenatal diagnosis due to the high recurrence rate of 25% in subsequent pregnancies of the mother.

Keywords: Ensephalocele, fetal kidney, meckel gruber syndrome, polydactyly

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PP-18 Prenatal sonographic diagnosis of VACTERL syndrome

Büsra Berfin Polat[1], Rauf Melekoglu[1]
[1]Inonu University Faculty of Medicine, Department of Obstetrics and Gynecology, Malatya, Türkiye
DOI:10.59215/prn.23.031supp033

Objective: VACTERL syndrome is a genetic syndrome that occurs in 1 in 10,000 to 40,000 newborns. No specific genetic or chromosomal defect associated with VACTERL syndrome has been identified. It is defined by the presence of vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies and at least three of the limb abnormalities seen on ultrasonography. In this report, we aimed to present the prenatal diagnosis of a case of VACTERL syndrome with multiple fetal anomalies.[2]

Case: A 30-year-old patient with a gravida 2, parity 1, 22 weeks and 3 days gestation according to the last menstrual period was admitted to the prenatal diagnosis and treatment unit of our clinic for fetal anomaly screening. Ultrasonography showed positive fetal heartbeat, normal amniotic volume and small biometric measurements according to the gestational age. Fetal neurosonography showed lemon sign and banana sign (figure 1) and vertebral evaluation revealed open spina bifida (meningomyelocele) at the S1-S4 level of the sacral vertebrae. Fetal abdominal examination revealed gastroschisis (figure 2) and horseshoe kidney anomaly. Fetal echocardiography showed complete AVSD. Fetal extremity examination revealed bilateral pes equinovarus and genital examination revealed anal atresia. The family was informed in detail about the possible poor fetal/neonatal prognosis of the fetus with VACTERL syndrome in the foreground and invasive prenatal diagnostic test and termination of pregnancy were presented as options. The patient underwent amniocentesis. Without waiting for the results of amniocentesis, the patient and her husband requested termination of the pregnancy due to multiple fetal anomalies present in the fetus. After termination of pregnancy, fetal autopsy confirmed the prenatal findings (figure 3).