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

References

1. Chen CP. Meckel syndrome: genetics, perinatal findings, and differential diagnosis. Taiwan J Obstet Gynecol. 2007 Mar;46(1):9-14.
2. Barisic, I., Boban, L., Loane, M., Garne, E., Wellesley, D., Calzolari, E., … Verellen-Dumoulin, C.(2015). Meckel–Gruber Syndrome: A population-based study on prevalence, prenatal diagnosis, clinical features, and survival in Europe. European Journal of Human Genetics, 23, 746–752.
3. Weichert, J., Hoellen, F., Krapp, M., Germer, U., Axt-Fliedner, R., Kempe, A., … Gembruch, U. (2017). Fetal cephaloceles: Prenatal diagnosis and course of pregnancy in 65 consecutive cases. Archives of Gynecology and Obstetrics, 296, 455–463.
4. Hartill V, Szymanska K, Sharif SM, Wheway G, Johnson CA. Meckel-Gruber Syndrome: An Update on Diagnosis, Clinical Management, and Research Advances. Front Pediatr. 2017 Nov 20;5:244.
5. Erger, F., Bruchle, N. O., Gembruch, U., & Zerres, K. (2017). Prenatal ultrasound, genotype, and outcome in a large cohort of prenatally affected patients with autosomal-recessive polycystic kidney disease and other hereditary cystic kidney diseases. Archives of Gynecology and Obstetrics, 295, 897–906.

PP-18 Prenatal sonographic diagnosis of VACTERL syndrome

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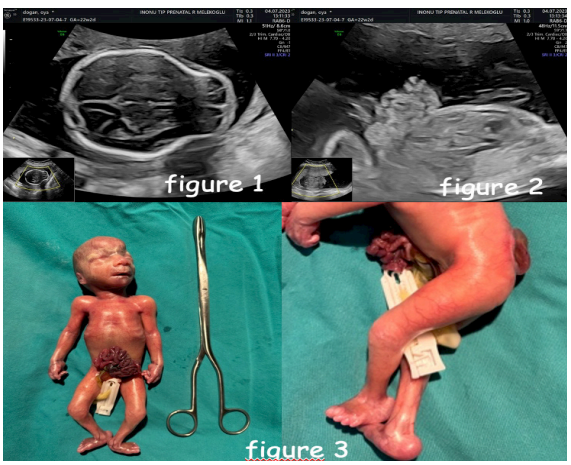
Objective: VACTERL syndrome is a genetic syndrome that occurs in 1 in 10,000 to 40,000 newborns. No specific genetic or chromosomal defect associated with VACTERL syndrome has been identified. It is defined by the presence of vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, 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).



Discussion: Numerous studies have investigated over 400 cases involving fetuses with VACTERL syndrome and partial caudal regression syndrome. The reported incidence rates of various abnormalities were as follows: spine abnormalities ranged from 60% to 80%, anal atresia from 55% to 90%, tracheoesophageal abnormalities from 50% to 80%, cardiac malformations from 40% to 80%, limb malformations from 40% to 50%, and kidney malformations from 50% to 80%. In conclusion, prenatal ultrasonic diagnosis plays an indispensable role in identifying VACTERL syndrome and partial caudal regression syndrome, offering valuable guidance for obstetric treatment. Its clinical implementation is highly warranted.[1]

Conclusion: VACTERL syndrome is typically defined by the presence of at least three of the following congenital malformations: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies and limb abnormalities. In addition to these main component features, patients may have other congenital anomalies. It is possible to suspect the diagnosis by antenatal ultrasonography. It is important to detect, suspect and investigate further when the first signs appear on a routine ultrasound scan.

Keywords: Cardiac malformation, limb anomaly, vertebral defect, VACTERL

References

1. Van de Putte, R., van Rooij, I.A.L.M., Marcelis, C.L.M. et al. Spectrum of congenital anomalies among VACTERL cases: a EUROCAT population-based study. Pediatr Res87, 541–549 (2020).
2. Zhang W, Chen J. Prenatal diagnosis of VACTERL association in the first trimester. Prenat Diagn. 2022 Apr;42(4):482-483.
3. Solomon BD. VACTERL/VATER association. Orphanet J Rare Dis. 2011;6:56.
4. Guthrie JD, DeCosta L. Prenatal Diagnosis of VACTERL Association. Journal of Diagnostic Medical Sonography. 2012;28(5):245-249.

PP-19 Prenatal diagnosis of isolated bilateral congenital cataract

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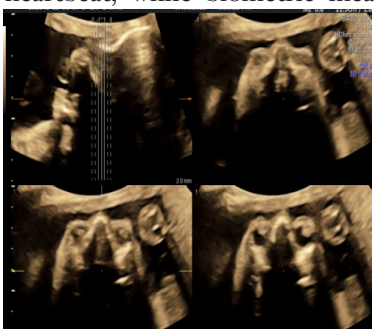
Objective: Congenital cataract stands as an orbital anomaly characterized by lens opacity, manifesting unilaterally or bilaterally, with an incidence of 1 in 10,000 births. Genetic syndromes are identifiable in roughly 10% of cases; nonetheless, congenital infections have been discerned in approximately 30% of instances. In the presence of unilateral or bilateral congenital cataracts, a comprehensive ultrasound evaluation, encompassing neurosonography, invasive prenatal diagnostic testing for karyotyping, and maternal TORCH panel analysis for fetal infections, becomes imperative. Furthermore, maternal utilization of pharmaceuticals (including steroids), radiation exposure, or exposure to potential teratogens, along with any underlying metabolic conditions, should be meticulously investigated for their potential etiological implications. In the context of this report, our objective is to present a case involving the prenatal diagnosis of an isolated instance of bilateral congenital cataract..

Case: A 36-year-old patient, gravida 6, parity 4, at 24 weeks and 4 days of gestation, was admitted to the prenatal diagnosis and treatment unit at our clinic for fetal anomaly screening. The patient had a history of pregestational diabetes mellitus and was meticulously monitored. Ultrasonographic examination confirmed a positive fetal heartbeat, while biometric measurements aligned with the gestational age, and amniotic fluid volume remained within normal ranges. Notable findings in fetal facial assessment included increased opacity observed in bilateral lenses within the coronal plane and orbital assessment (figure 1). Fetal neurosonography and abdominal evaluation yielded no evidence of periventricular calcification, hepatic/splenic calcification, or hyperechogenic bowel. Given the patient's history of congenital cataract in a previous child, an isolated diagnosis of congenital cataract was considered. Upon fetal cardiac evaluation, borderline myocardial hypertrophy and perimembranous ventricular septal defect (VSD) were identified. At this juncture, the option of invasive prenatal diagnostic testing or cell-free fetal DNA analysis in maternal blood was presented. Maternal blood TORCH panel analysis yielded no abnormal findings, and in light of the patient's preferences, she chose not to undergo invasive prenatal diagnostic testing. Throughout this process, the family was provided with comprehensive information regarding potential neonatal complications.



Discussion: Genetic testing and prenatal ultrasound have become primary methods of diagnosing congenital cataracts. The analysis by Yue Qin et al found a total of 41 cases of congenital cataracts diagnosed prenatally among 788,751 women who underwent the mid-tirmester fetal anatomical scan. Based on sonographic features, 16/41 (39.0%) had an intense echogenic pattern, 15/41 (36.6%) had a hyperechogenic spot, and 10/41 (24.4%) had a "double ring" sign. 17/41 (41.5%) were isolated and 24/41 (58.5%) had associated intraocular and extraocular findings. Microphthalmia, cardiac abnormalities and central nervous system abnormalities were the most common associated abnormalities. Potential etiology regarding the disease, 6 cases had known family history of congenital cataracts, 4 cases had confirmed congenital rubella infection, and 2 cases had aneuploidy.[3]