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Turner syndrome presenting with bilateral multicystic dysplastic kidney disease

Rachel Rayburn, MS, CGC[1]; Alan Donnenfeld, MD[2]
[1]Labcorp, Louisville, KY; [2]Labcorp, Philadelphia, PA

Background

Turner syndrome is diagnosed when a phenotypic female is missing all or part of their second X chromosome.[1] The features of Turner syndrome include short stature, ovarian dysgenesis, delayed puberty, infertility, congenital anomalies, endocrine disorders, autoimmune conditions, Type 2 diabetes and neurocognitive conditions.[2] Common ultrasound findings associated with Turner syndrome include cystic hygroma, hydrops fetalis, subcutaneous edema, short femur, horseshoe kidney, omphalocele, narrowed aortic arch, ventricular septal defect and large fourth ventricle.[3] Multicystic dysplastic kidneys are known to be associated with genetic syndromes and chromosome conditions.[4] Cystic renal disease is seen in 1.76% of individuals with Turner syndrome.[5]

Case Presentation

A 38-year-old G3P1 patient was referred for genetic counseling due to an abnormal ultrasound finding of bilateral multicystic dysplastic kidney disease and anhydramnios. The patient previously had normal NIPS for trisomies 21, 18 and 13. The patient's family history was non-contributory. Amniocentesis could not be performed due to anhydramnios. NIPS including genome analysis and carrier screening including testing for autosomal recessive polycystic kidney disease (ARPKD) were performed for the patient. NIPS results showed 45,X. Carrier screening results were negative for ARPKD. The patient was provided with pregnancy management options and elected to continue the pregnancy.

Discussion

The possibility of 45,X being an incorrect or incomplete diagnosis cannot be excluded due to the limitations of NIPS testing and the fact that amniocentesis could not be performed due to lack of fluid. However, isolated bilateral multicystic dysplastic kidneys are an interesting and uncommon presentation for a fetus with Turner syndrome.

Conclusions

Bilateral multicystic dysplastic kidneys are often sporadic abnormalities of embryologic development. In a small proportion of cases, genetic etiologies are identified through microarray analysis, genetic testing using a congenital anomalies of the kidney and urinary tract (CAKUT) panel or whole exome sequencing. The identification of 45,X as the cause of bilateral multicystic dysplastic kidneys is unusual. Establishing the diagnosis of a chromosome abnormality as the cause of the fetal anomalies has important implications for prognosis and recurrence risks. It also provides closure to the patient as a diagnosis has been established. Although the patient was counseled that prognosis for fetal survival due to the kidney anomalies and the anhydramnios is extremely poor, the patient was interested in continuing the pregnancy and her decision was supported.

References

1. Gravholt CH, Andersen NH, Conway GS, et al; International Turner Syndrome Consensus Group. Clinical practice guidelines for the care of girls and women with Turner syndrome: proceedings from the 2016 Cincinnati International Turner Syndrome Meeting. *Eur J Endocrinol*. 2017;177(3):G1-G70. doi:10.1530/EJE-17-0430. PMID:28705803
2. Gravholt CH, Viuff M, Just J, et al. The changing face of Turner syndrome. *Endocr Rev*. 2023;12;44(1):33-69. doi:10.1210/endrev/bnac016. PMID:35695701
3. Bronshtein M, Zimmer EZ, Blazer S. A characteristic cluster of fetal sonographic markers that are predictive of fetal Turner syndrome in early pregnancy. *Am J Obstet Gynecol*. 2003;188(4):1016-1020. doi:10.1067/mob.2003.230. PMID:12712103
4. Deshpande C, Hennekam RC. Genetic syndromes and prenatally detected renal anomalies. *Semin Fetal Neonatal Med*. 2008;13(3):171-180. doi:10.1016/j.siny.2007.10.006. Epub 2007 Dec 26. PMID:18162447
5. Fanos V, Schena S, Dal Moro A, Portuese A, Antoniazzi F. Multicystic kidney dysplasia and Turner syndrome: two cases and a literature review. *Pediatr Nephrol.* 2000;14:754-757. doi:10.1007/PL00013430

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