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Objectives

Renal abnormalities comprise approximately 20% of congenital abnormalities and affect 3-4% of the population. Most fetal renal abnormalities are diagnosed during the second or third trimester. We present the first known case of an HNF1b mutation detected as an abdominal cyst during the first trimester anatomy ultrasound. This cyst later developed into a multicystic dysplastic kidney (MCDK). Additionally, we review the literature and describe three similar MCDK cases detected in the first trimester.

Study Design

We describe and compare our patient's case to three other cases reported in PubMed of MCDK diagnosed in the first-trimester US.

Case Description

A 36-year-old G5P4014 at 13 weeks with LR cfDNA presented to an early anatomical scan with a right abdominal cystic mass. Otherwise, her early scan was unremarkable, including NB and NT. She has one son with bilateral cystic dysplasia. The fetal anatomical survey at 20 weeks revealed right MCDK. Declined genetic amniocentesis. Delivered at term - IOL VD AGA baby girl . A cord blood for hereditary cystic kidney diseases Panel revealed heterozygosity for a ~ 1.3 Mb deletion of 17q12, including the HNF1B, which is associated with congenital cystic disease, early onset DM, and mental health disorder. At the age of 13 months, diagnosed with renal failure and convulsive disorder, likely focal epilepsy.

Results

All four known cases of MCDK diagnosed through first-trimester ultrasound share similarities in their detection timing, final diagnosis, and outcomes. However, our case is unique because it involves an HNFb1-associated MCDK, diagnosed later.

|                     | Our Case                  | Tudorache et al.              | Wong et al.           | Hata et al.     |
|---------------------|---------------------------|-------------------------------|-----------------------|-----------------|
| Age at US dx        | 12w1d                     | 11w3d                         | 13 wks                | 11wks           |
| Final dx            | Unilateral MCDK           | Unilateral MCDK               | Unilateral MCDK       | Unilateral MCDK |
| Outcome             | Term Delivery             | TOP<br>MCDK confirmed autopsy | Term Delivery         | Term delivery   |
| Genetic testing     | HNF1B mutation            | Normal karyotype              | Unknown               | Unkown          |
| Family hx           | Mother and maternal uncle | None                          | Unknown               | Unknown         |
| Other abnormalities | None                      | Contralateral pelviectasis    | Transient hydroureter | None            |

Table 1 Case Series Description

Conclusions

First trimester ultrasounds have advanced exponentially over the last two decades and are now used as an early screening tool for anatomical and genetic abnormalities. Patients are now privy to earlier knowledge and opportunities for additional testing, management, and planning a course of action throughout the pregnancy. This rare case, among a few others, can provide MFM experts with new insights into first-trimester abdominal cysts. We conclude that renal abnormalities, including HNF1b-associated MCDK, can be diagnosed as early as 12 weeks and should be added to the differential diagnosis of first-trimester abdominal masses and cysts, and early genetic evaluation should be considered.

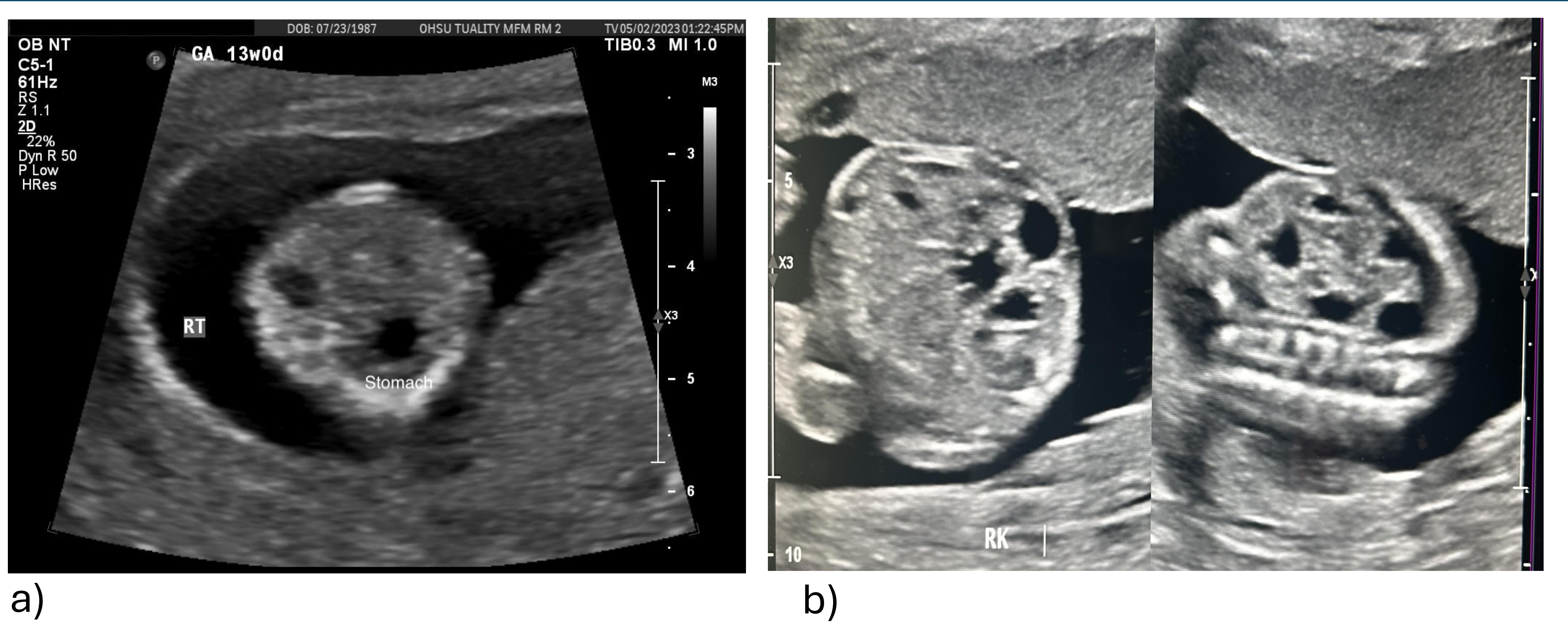


Figure 1a) Right abdominal cyst at 12w1d 1b) Right MCDK at 20w

For questions:  
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