Antenatal Presentation of Wilms' Tumor

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Tumors discovered antenatally are rare and the prognosis is generally poor. We present a case of a fetal left renal mass, initially discovered via routine prenatal ultrasound. The mother was an otherwise healthy gravida 4 para one 30-year-old female with 2 previous miscarriages, reportedly secondary to septate uterus. Further imaging and subsequent genetic testing was consistent with an antenatal Wilms' tumor with a mutation in the *NPHP1* gene. The newborn received chemotherapy and had no evidence of recurrence at 3 months follow-up. UROLOGY 134: 225–227, 2019. © 2019 Elsevier Inc.

Antenatal tumors are rare and have been reported to have a prevalence of fewer than 10 per 100,000 live births.[1] Congenital mesoblastic nephroma is the most common neonatal renal mass, with Wilms' tumors rarely reported antenatally.[2-4] We present a case of an antenatally-detected fetal left renal mass, initially discovered via routine prenatal ultrasound.

CASE REPORT

At 29 weeks estimated gestational age (EGA), routine prenatal ultrasound testing showed a 3.4 cm solid mass arising from the lower pole of the left kidney, associated with mild hydronephrosis. Given its antenatal presence, this was felt to be most likely a congenital mesoblastic nephroma (CMN) as CMN accounts for nearly 2-thirds of prenatal renal tumors.[5] The right kidney was noted to be normal without masses or hydronephrosis. A pelvic magnetic resonance imaging (MRI) performed at 32 weeks EGA showed a 4.2 cm left renal mass (Fig. 1). The mass continued to grow on subsequent imaging studies, reaching up to 6.5 cm on final ultrasound obtained at 33 weeks EGA. Due to nonreassuring fetal status, the baby was delivered at 34 weeks via emergency Cesarean section. She was intubated after delivery for respiratory distress but was stabilized and extubated later that day. On examination, she was noted to have a palpable mass in the left flank and began making wet diapers shortly after birth.

On day of life 1, an abdominal MRI obtained for further evaluation showed a 7.5 cm left renal mass with no renal vein extension (Fig. 2). No metastases were seen on the MRI.

On day of life #4, she underwent an uncomplicated open left nephrectomy and periaortic lymphadenectomy via a subcostal incision. She tolerated the procedure well. She was transferred back to the neonatal intensive care unit postoperatively and extubated on postoperative day #4. Final pathology was consistent with favorable histology Wilms' tumor without anaplastic features. Capsular disruption and renal sinus lymphovascular invasion were present, however, consistent with Children's Oncology Group stage III Wilms' tumor. Given the unexpected pathology findings, the genetics team was consulted. An ophthalmologic examination and serial intracranial ultrasounds were normal. Serial echocardiograms showed a small patent ductus arteriosus, but no other anatomic anomalies. Newborn screening laboratories for primary congenital hypothyroidism, congenital adrenal hypoplasia, organic acid disorders, amino acid disorders, and lysosomal storage disorders were negative. *WT1* testing was normal, but a chromosomal microarray showed interstitial loss on the long arm of chromosome 2 at q12.1q13, which

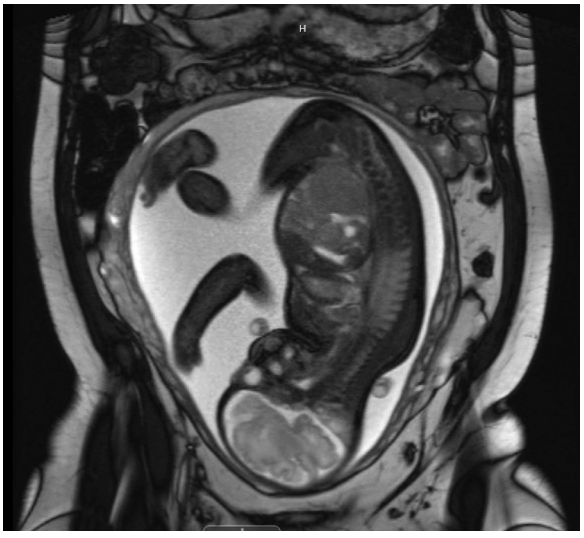

Figure 1. Antenatal pelvic MRI at 32 weeks estimated gestational age showing large left fetal renal mass. MRI, magnetic resonance imaging.

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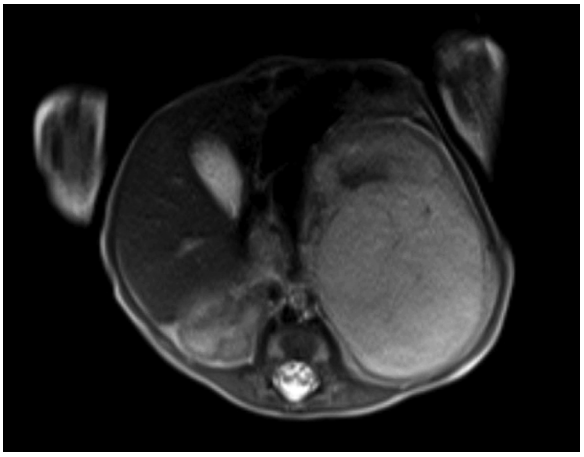

Figure 2. On day of life 1, abdominal MRI showing large left renal mass with anterior displacement of kidney, axial view. MRI, magnetic resonance imaging.

spans the *NPHP1* gene. Deletions of this gene have been associated with nephronophthisis but not Wilms tumor.[6]

On postoperative day #14, chemotherapy was initiated with vincristine, dactinomycin, and doxorubicin (Regimen DD4A). After discussion with radiation oncology, the decision was made to avoid radiation therapy given the patient's age. She completed 4 weeks of chemotherapy while inpatient and was discharged home 6 weeks status post nephrectomy in good condition. She was readmitted 4 days after initial discharge for anemia that responded to transfusion and again for nausea and vomiting 2 weeks later that resolved with conservative measures. Follow-up imaging obtained 3 months following completion of the chemotherapy regimen showed no evidence of disease (*Fig. 3*).

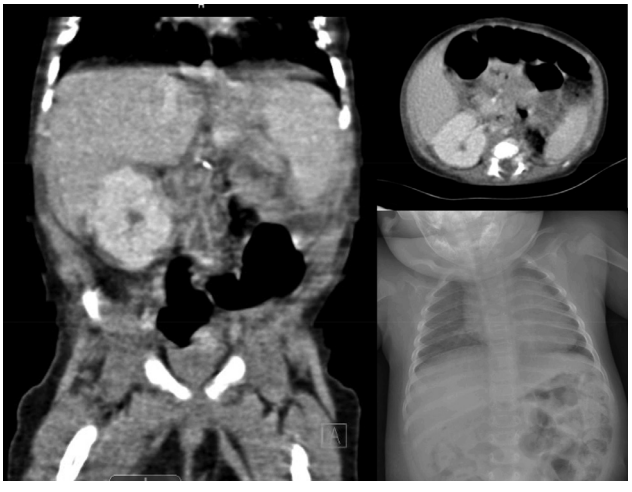

Figure 3. Postoperative abdominal CT showing no evidence of disease on coronal, left panel, and axial views, top right panel. Chest radiograph 3 months following completion of chemotherapy also showing no evidence of disease, right lower panel. CT, computed tomography.

DISCUSSION

The prevalence of antenatal renal tumors is extremely rare, around 7 in 100,000 live births.[1] Moreover, antenatal renal tumors are most commonly CMN while other tumors like Wilms' tumors, neuroblastoma, and teratomas are less common. The detection of Wilms' tumors with prenatal MRI and ultrasound has been the focus of prior case reports and studies.[4,7,8] Differentiating CMN from Wilms' tumors with prenatal ultrasound is difficult with very few reports demonstrating histologic confirmation of Wilms' tumor antenatally.[7]

While antenatal Wilms' tumors have been previously reported, the possible role of the *NPHP1* gene in the development in Wilms' tumor has not been previously reported. The role of *NPHP1* in the development of nephronophthisis has been clearly delineated, but no prior case report has shown a possible link with an isolated Wilms' tumor.[6] In this case, there were no other congenital abnormalities discovered on further work-up to indicate Denys-Drash syndrome WAGR syndrome, or Beckwith-Wiedemann syndrome, as well as the lack of mutation in *WT1*. The extensive genetic work-up failed to show evidence for primary congenital hypothyroidism, congenital adrenal hypoplasia, organic acid disorders, amino acid disorders, or lysosomal storage disorders to explain other associations with Wilms' tumor. Genetic predisposition to Wilms' tumors are known to be associated with numerous genetic mutations,[9] and it may be possible that *NPHP1* is another predisposing genetic variant.

CONCLUSION

Antenatally detected renal tumors are rare and may require additional work-up to assess the tumor genetics. CMN is the most common variety but Wilms' tumors do occur rarely and can respond to surgical resection and chemotherapy. Further research is needed to elucidate the possible genetic pathogenesis of Wilms' tumors from the *NPHP1* gene.

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