The eL18-4 PureWave linear array transducer in the evaluation of tuberous sclerosis

**eL18-4 PureWave linear array transducer**

**Category**
Tuberous sclerosis

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**Overview**
Many technological advancements have been made in ultrasound over the years and especially within the last decade. Imaging with high resolution transducers allows a very comprehensive evaluation of fetal anatomy at a level today that had not been possible in the past.

**Patient history**
A 19-year-old gravida 3, para 2 pregnant female was referred to our center with a history of echogenic masses within the fetal heart. The patient related an estimated date of delivery of 1/20/2019, which projected to a gestational age of 27 weeks and 5 days at the time of evaluation. Ultrasound performed at the referring center at 23 weeks and 4 days revealed a hyperechoic area measuring 2.0 x 1.6 x 2.0 cm seen in the region of the right chest, as well as another smaller hyperechoic area seen in the left ventricle of the heart, new findings not identified on the prior anatomy scan. She was referred to the Fetal Heart Program and the Center for Fetal Diagnosis and Treatment at the Children’s Hospital of Philadelphia for further evaluation and counseling regarding diagnosis, prognosis and management options.

**Protocol**
A complete detailed examination of the fetal anatomy was performed utilizing the Philips EPIQ 7G system and multiple transducers including the C5-1, C9-2 and eL18-4.

**Findings**
The detailed fetal anatomic survey noted a male fetus in a cephalic presentation with an anterior placenta, free of the region of the internal cervical os. The amniotic fluid index was normal, measuring 19.4 cm with a deepest pocket of 6.2 cm. Fetal biometry estimated the average gestational age approximately 1 day behind expected, with an estimated fetal weight of 1128 g which was normal at the 40th percentile. Multiple cardiac masses were identified, all of which were uniformly echogenic and solid. The largest mass arose from the right atrial wall and measured 2.5 x 2.2 x 2.3 cm (Figure 1).

Multiple echogenic subcortical nodules were also visualized, the largest of which measured 1.0 x 0.9 cm, located along the left frontoparietal lobe. The high-resolution images acquired with the eL18-4 transducer demonstrated clusters of tiny cysts within the lower pole of the left kidney and the interpolar region of the right kidney. The kidneys otherwise appeared unremarkable with normal size and cortical medullary differentiation. A normal cycling bladder was identified.

**Figure 1** Transverse image of the fetal chest using the C9-2 transducer – solid echogenic masses are seen within the heart, identified as rhabdomyomas.
Conclusion

Rhabdomyomas are the most common prenatal and neonatal cardiac tumors and are present in about 80% of fetuses with tuberous sclerosis. This is an autosomal dominant disorder characterized by abnormal cell proliferation and tumor growth in multiple organ systems, which creates various symptoms according to the location. The association of rhabdomyomas with cortical or subependymal nodules (tubers) in the brain, or rarely, renal lesions, permits the definitive diagnosis of tuberous sclerosis complex withcertainty. These findings can greatly assist with prenatal counseling; however, cortical tubers are often not discernible on ultrasound, and therefore fetal MRI hasbeen reported as more sensitive for the detection of these lesions.

Rhabdomyomas can grow over the course of the pregnancy, but often regress after birth. Primary issues prenatally are the possibility of obstruction of the outflow tracts, arrhythmias, fetal hydrops and cardiac shock. Cortical tubers postnatally may be associated with an increased risk of seizures, learning disabilities and developmental neurological disorders such as autism. Renal tumors, most commonly angiomyolipomas or nodular hamartomas, can lead to significant compromise of renal function, requiring transplantation.

Linear array transducers offer high-resolution anatomic assessment, but traditionally have been limited to superficial structures only, such as small parts and vascular examinations. By using the eL18-4 PureWave linear array transducer, we were able to identify tiny cystic clusters within the lower pole of the left kidney (Figure 2a) as well as the interpolar region of the right kidney. The eL18-4 transducer also more clearly defined the smaller cortical nodules in the right hemisphere as seen in Figure 3a, compared to the C9-2 transducer (Figure 3b).

The renal cysts were not defined with the C9-2 curved array transducer used earlier in the examination (Figure 2b) or with the C5-1 transducer (Figure 2c). In this case, the ability to penetrate the tissues in a third trimester patient to differentiate the retroperitoneal structures with such fine detail is simply without precedent. The renal findings, combined with the cardiac masses and cortical nodules, indicated a definitive diagnosis of fetal tuberous sclerosis, which assisted with prenatal counseling of this patient.

References


Results from case studies are not predictive of results in other cases. Results in other cases may vary.