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Cystic lymphangioma of the axilla and chest wall. Jackson, DN MD, Brown L, MS, CGC, Fetal Diagnostic and Imaging Center, St. Vincent's Health Center, Department of Maternal Fetal Medicine and Fetal Imaging, Billings, MT

Fetal cystic lymphangiomas are benign tumors arising from the lymph vessels. They have been demonstrated in a number of different anatomic locations, including the nuchal region, axilla, mediastinum, chest wall, retroperitoneal area, abdominal viscera, groin and pelvis. The defect is most often found in the nuchal region. In these cases, the etiology is related to inadequate drainage of the lymphatic vessel into the venous system. Nuchal lymphangiomas are often associated with chromosome abnormalities. Cystic lymphangiomas at other locations are thought to have a distinct etiology, most likely as a result of insufficient anastomoses with larger lymphatic channels. The association with chromosome anomalies in these cases is less distinct. We describe 2D and 3D ultrasound findings associated with the prenatal finding of cystic lymphangioma in two patients. The first patient is a 29-year-old G4P2 female referred for an ultrasound due to marginal sinus bleeding. A follow-up scan at 23 weeks identified a multiloculated cystic mass measuring 7.2 x 6.8cm in its largest dimensions. The patient was followed at 3-week intervals with 2D and 3D imaging performed at each visit. At the time of delivery, the mass measured 8.2x7.9cm. Surgical excision was performed on day 2. The second patient is a 23-year-old G1P0 female referred at 35 wks for an ultrasound secondary to the finding of a multiloculated mass on the right chest wall, 4 cm below the axilla. The mass measured 2x3cm. The right arm appeared to move freely. Both 2D and 3D imaging were performed. At birth, the mass measured 1x1cm. Surgical excision has not been performed. Medical management is currently observational. Both patients were offered an amniocentesis. Neither patient opted for this procedure. Additional birth defects or genetic anomalies were not detected in either patient. The 3D ultrasound allowed for a unique imaging opportunity of a rare congenital malformation

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Congenital mesoblastic nephroma: perinatal, surgical and genetic features and associated 2D and 3D imaging. Jackson, DN MD, Haag, M, PHD*, Brown L, MS, CGC, Keel-Thompson, K RDMS, Phillips, SM, Tunby, M, Beischel, LS, Bennett, TL**.Fetal Diagnostic and Imaging Center, St. Vincent's Health Center, Department of Maternal Fetal Medicine and Fetal Imaging, Billings, MT, Shodair Hospital Department of Medical Genetics, Helena, MT**Department of Pathology, St. Vincent's Health Center, Billings, MT.

Mesoblastic nephroma is a mesenchymal neoplasm that arises from the metanephric blastema. This mass represents 80% of solid renal masses in neonates. We present a 23yo G3P2 Native American female initially referred at 24 weeks due to an enlarged uterus. On 2D and 3D ultrasound exam, a male fetus was noted to have a 4cmx5cm solid tumor in the right retroperitoneal space and polyhydramnios. By 28 wks, the mass had increased to 8x6cm with more extreme polyhydramnios. The mass originated in the renal fossa and extended to the level of the diaphragm superiorly and across the midline. The mass replaced the fetal right kidney. The left kidney appeared normal. Premature labor initiated hospitalization. A series of drainage amniocentesis were performed due to extreme polyhydramnios and preterm labor. Chromosome analysis on the fluid showed a normal male karyotype, 46, XY. Multiple 2D and 3D images were obtained on this patient. She delivered at 33w1d by cesarean section due to sudden labor and dilitation. Surgical resection was performed on day 5. The mass measured 10x10cm at birth. The diagnosis of mesoblastic nephroma, with classic morphology was confirmed by histopathology. Cytogenetic studies on cells from short term cultures showed a hyperdiploid male karyotype with extra copies of chromosomes 7,8,11, 16, and 17. Trisomy 16 was observed as the sole abnormality in two cells from one isolated area. Nonrandom chromosome abnormalities of trisomy 11, 8 and 7 have been reported for several cases of mesoblastic nephroma. Trisomy 11 is most commonly associated with cellular or mixed histology, unlike this specimen. Parental origin of the extra copy of 11 and the relationship to renal tumor etiology is of interest and molecular studies are in progress for this case. The combination of three-dimensional ultrasound, pathology and genetic studies has facilitated diagnosis, treatment and further investigation of this rare prenatal presentation of a congenital renal tumor.

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Sonogram findings with Brachmann-de Lange syndrome. G.A. Jervis, C.S. Kalter, and B.G. Kousseff. University of South Florida Regional Genetics Program and Florida Perinatal Associates, Tampa, FL.

We present KK, a 32 year old woman, G4 P3, who had a sonogram at 21 weeks of gestation which revealed bilateral ulnar hypoplasia, bilateral radial aplasia, single ray distal digit, single umbilical artery, micrognathia and small right pleural effusion. No movements were noted in the elbows. The humeri appeared normal. The legs appeared normal. Fetal growth, fetal echocardiogram and amniotic fluid volume were normal. Family history was unremarkable for limb abnormalities. There was a maternal second cousin with Trisomy 13 and a paternal first cousin with Turner syndrome. The differential diagnosis based on the sonogram findings of upper limb abnormalities included chromosomal abnormalities, Roberts syndrome, Holt-Oram syndrome, Tabatznick syndrome, Weyers syndrome, TAR syndrome and Baller-Gerold syndrome. Amniocentesis at 33 weeks revealed normal female chromosomes and no evidence of premature separation of the centromeres, as seen in Roberts syndrome. RK was born by spontaneous vertex delivery at 37 weeks by dates; birth weight: 2300 g (2nd centile), length: 44.5 cm (2nd centile) and head circumference: 32.5 cm (2nd centile). Three-vessel cord was reported. RK had micromelia of the arms, bilateral single digital ray, underdeveloped forearms and flexion contractures at 30 degrees were impossible to passively correct. Legs were normal and toes were somewhat small. Ears were lowset. Synophrys was present. Short, upturned nose, long, thin philtrum and thin lips were noted. Short neck, low posterior hairline were present. Nipples were wide spaced and hypoplastic. The phenotype was consistent with Brachmann-de Lange syndrome (MIM# 122470). This is a well-delineated syndrome, which includes limb deformities. RK did not have prenatal growth retardation and she did not have lower limb deformities, which can include absent tibia and bifurcated femur. However, the upper limb deformities are consistent with Brachmann-de Lange syndrome. This case illustrates the variability seen in Brachmann-de Lange syndrome and emphasizes the importance to include this condition in the differential diagnosis when limb defects are seen on prenatal sonogram.

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Recurrence of triploidy in a woman with low level 45,X mosaicism. L.R. Johnson[1], R.I. Blough[2] and M.E. Miller[1]. [1]Children's Medical Center, Dayton, OH, [2]Children's Hospital Medical Center, Cincinnati, OH.

Triploidy (69 chromosomes) occurs in 1 to 3% of recognized pregnancies typically resulting in first trimester spontaneous abortion. The recurrence risk after one affected pregnancy is 1-1.5%. Triploidy can be diandric or digynic in origin. Diandric triploids result from dispermy or fertilization from a diploid sperm. Diandric triploids result in a partial hydatidiform mole. Digynic triploids may be due to a complete nondisjunctional event during oogenesis, retention of a polar body or fertilization of an ovulated primary oocyte. Digynic triploids may survive to the second trimester and develop as a growth retarded fetus with a small placenta. We report a woman with four first trimester spontaneous abortions of which three were triploid. The first miscarriage was not karyotyped, the second showed 69,XXX, the third 69,XXY and the fourth 69,XXX. Parent of origin studies were not performed, so it is unclear whether the triploid conceptions were diandric or digynic in origin. The proband also has a normal son. Chromosomal analysis on the proband showed 45,X[3], 46,XX[27]. Although nondisjunction is reported among offspring of Turner mosaics, we are not aware of any other reports of recurrent triploidy associated with low level sex chromosome mosaicism in the mother. The triploidy may be directly correlated with Turner mosaicism or may represent a random association.