Prenatal transient hydrops and vascular aortic ring in a 16p12.2 microdeletion associated with autism spectrum disorder

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Introduction
Prenatal abnormal ultrasound findings of 16p12.2 microdeletion are rare. We describe here a case of prenatal fetus with vascular aortic ring followed by a transient hydrops and microarray diagnosed 16p12.2 microdeletion.

Case presentation
A 39 y.o. G4P3003 with two children with severe autism, diagnosed with fetal vascular aortic ring during fetal anatomic survey. Declined invasive genetic testing and pursued cfDNA- resulting as low risk.

At 25w0d a follow up US revealed a new finding of fetal echogenic bowel and opted for collection of umbilical cord blood collection at delivery for karyotype and chromosomal microarray. CF carrier screening and IgG/IgM serology for CMV and toxoplasmosis were negative.

A repeat US at 30w0d showed mild ascites in addition to mild pericardial effusion with normal MCA and UA Dopplers and appropriate fetal growth and BMZ course for lung maturity was administered secondary to concern for worsening of fetal hydrops. Reassuring fetal status allowed conservative management.

At 31w5d FU US showed resolution of both pericardial effusion and ascites and subsequent bi-weekly US confirmed this resolution. At 35w6d she presented at labor and delivery with decreased fetal movements and NST confirmed acute fetal distress. An urgent LTCS delivered a male infant with BW 3990gm. Neonatal demise occurred on day 2 post delivery. Patient declined autopsy. Cord blood microarray resulted in a 479 Kb deletion at chromosome 16p12.2 which is associated with developmental/intellectual disabilities, speech and communication delay, autism, cardiac defects.

Conclusion
We demonstrate a confirmed case of triploidy by CVS diagnosis based upon early delayed growth while meeting other developmental milestones. To our knowledge, this the first case of displaying such an early discordancy in triploidy. Consideration of triploidy when early growth delay offers early counseling and genetic testing.