

Early Second Trimester Asymmetric Growth and Mosaic Triploid Liveborn

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Abstract

Background: Mosaic triploidy is uncommon in a liveborn. There have been no previous reports of prenatally diagnosed diploid/triploid mosaicism by amniocentesis. We present a case of prenatally diagnosed diploid/triploid mosaicism.

Case: A 26 year old G3P1 presented for ultrasound at 21 weeks 5 days following a positive quad screen. Asymmetrical growth restriction, abnormal hand positioning and suspected heart defect were noted. Amniocentesis revealed mosaic triploidy; 69, XXX (10)/46, XX (6). Infant was delivered at 37 weeks by repeat cesarean section. Neonatal cord blood karyotype confirmed mosaic triploidy; 69, XXX (2)/46, XX (18).

Conclusion: Prenatal diagnosis and subsequent outcome of a liveborn mosaic diploid/triploid infant has not been previously described. Our case demonstrates that diploid/triploid mosaicism may be diagnosed by amniocentesis and that the pregnancy may be managed to live birth near term.

Case Report

Introduction

Mosaic triploidy is uncommon in a liveborn and there have been no previous reports of prenatally diagnosed diploid/triploid mosaicism by amniocentesis. Our case demonstrates that early asymmetric fetal growth in the second trimester may be an early indicator of diploid/triploid mosaicism. Patients may be successfully managed with serial ultrasound and antenatal testing.

Case

A 26 year old Gravida 3, Para 1, presented to our clinic at 17 weeks 6 days for routine ultrasound. At that time, the fetal AC was measuring at the 7th percentile for gestational age. The remainder of the fetal anatomy survey was limited by gestational age and fetal position. A follow up ultrasound was performed at 21 weeks 5 days following a positive quad screen; 1/36 for trisomy 18. Ultrasound revealed asymmetrical growth restriction. Both the BPD and AC measured < 5th percentile with the AC measuring 4 weeks behind. Abnormal hand positioning was noted and a heart defect that was later diagnosed as an atrial septal defect by fetal echo was suspected. An amniocentesis revealed mosaic triploidy. FISH analysis was significant for 34 of 72 cells trisomic for chromosomes 13 and 21 while 42 of 82 cells were trisomic for chromosomes X and 18. Final karyotype on amniocytes confirmed mosaic triploidy; 69, XXX (10)/46, XX (6).

The difficulty in predicting the prognosis for mosaic triploidy was conveyed to our patient. Possible outcomes including fetal demise, neonatal death, motor delay and developmental delay (possibly profound) were discussed. After extensive counseling, the patient desired continuation of the pregnancy with full intervention for fetal indications. The patient was followed with serial growth ultrasound scans; IUGR was noted to be symmetrical at 34 weeks at which point the EFW was 1296 g. The patient was followed twice weekly with biophysical profile and non-stress testing.

A female infant weighing 1555 g (< 10%) at 36 weeks 6 days was delivered via repeat cesarean section. The neonate was 38 cm in length (< 10%) and had an FOC of 32 cm (25-50%). Apgars were 6 and 8 at 1 and 5 minutes respectively. Dysmorphic features were noted including a prominent occiput, prominent forehead, up-slanting palpebral fissures, hypertelorism, hypoplastic helices, low and posteriorly rotated ears, prominent nasal bridge with beaked shape, small mouth

with a very high palate arch, small chin, short neck, asymmetric chest with left ribs more anteriorly placed than the right, short rib cage/sternum, complete syndactyly of 3rd and 4th fingers bilaterally w/ decreased creasing pattern, syndactyly of 4th and 5th toes on right foot and a lower back midline hair tuft. A weak cry, positive grasp and decreased suck were also noted on exam. Bone survey revealed microcrania and hypoplasia of the distal phalanx of all fingers and toes with no bony fusions between any digits. Chest X-ray noted thin ribs. Echocardiogram revealed a secundum atrial septal defect with left to right shunting, dilated coronary sinus, small PDA with left to right shunting, trace tricuspid regurgitation, trace pulmonary insufficiency, and mild right atrial and ventricular dilation. Lumbar spine ultrasound revealed termination of the conus medullaris at level of L3, raising the possibility of a tethered cord. Renal, pelvic and brain ultrasounds revealed no abnormalities; however, kidneys were noted to be small for gestational age. Pathology on the placenta revealed congestion and hemorrhagic disc as well as widespread villous edema.

Neonatal cord blood karyotype confirmed mosaic triploidy; 69, XXX (2)/46, XX (18). The infant was discharged home to be followed by pediatric cardiology, genetics and neurology. At 9 months of age our patient was able to roll over completely and was able to sign "more". At 12 months of age, she was able to grasp objects and move them from hand to hand with fine motor skills still lacking. Due to persistent upper body hypotonia, our patient was able to lift her head and chest off the floor for a limited amount of time while lying on her stomach. At 12 months, she was unable to sit unassisted or crawl and was without speech. At 16 months she is able to say "mama," has 3 teeth, is getting close to crawling and is able to sit unassisted once placed in seated position. She has required no surgeries to date.

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Discussion

Triploidy is a rare, sporadic chromosomal abnormality that is uniformly lethal with most affected pregnancies ending in miscarriage [1,2]. Triploidy is generally characterized by a large placenta with hydatidiform changes, asymmetric growth retardation, syndactyly, congenital heart defects and brain abnormalities [1,2]. Mosaic triploidy is thought to be less common than full triploidy with only a small number of reported liveborn individuals [3]. In addition to being less common, the prognosis for mosaic triploidy is more difficult to predict with the majority of affected individuals expected to exhibit some degree of psychomotor retardation. Other characteristics commonly found in patients with mosaic triploidy include growth and mental retardation, hypotonia, clinodactyly, syndactyly, body and facial asymmetry, truncal obesity and skin pigmentation anomalies [4].

The prognosis for mosaic triploidy is difficult to predict due to the inability to identify the proportion of triploid cells in each cell type of an individual, specifically the brain [5]. Adding to the difficulty of diagnosis, mosaic triploidy is not always able to be confirmed by blood karyotype as it is often not present in non-fibroblastic cell lines. Approximately 70- 75% of time blood karyotype is normal and the diagnosis of mosaic triploidy has to be made by cultured fibroblasts [3,6]. If the triploid cell line is found in lymphocytes, it usually comprises <5% of cells [3]. In 2003, Flori et al. [7] described an individual who escaped diagnosis until one year of age by cultured skin fibroblasts as the karyotype by amniocentesis, cordocentesis and cultured lymphocytes at birth were

all normal. Using the search terms “mosaic, triploid and prenatal” in pubmed from 1963 to present, no prior case of human diploid/triploid mosaicism diagnosed by prenatal amniocentesis has been described. Our case demonstrates that diploid/triploid mosaicism may be diagnosed by amniocentesis and that the pregnancy may be managed to live birth near term. Long term prognosis remains uncertain.

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