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Rare Syndrome of Thrombocytopenia Absent Radius Diagnosed Prenatally in a Female Fetus

Brittany Noel Robles*, Benham Enayataval and Daniel Faustin

Wyckoff Heights Medical Center, NY, USA

*Corresponding author: Brittany Noel Robles, Wyckoff Heights Medical Center, NY, USA, Tel: +1 212-241-6696; E-mail: BrittanyNoelRobles@gmail.com

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Abstract

Thrombocytopenia-absent radius (TAR) syndrome is a rare congenital condition which presents with thrombocytopenia and associated absence of the radius bone. The development of thrombocytopenia-absent radius syndrome has not been clearly identified, however there are several etiologies that have been proposed. Here we present a case of a 33-year-old African-American female, G6P3023 with no significant past medical, surgical, or obstetric history that was seen in our women's health clinic after finding out she was pregnant via a home pregnancy test. She had routine first trimester prenatal testing which was unremarkable. At the time of the twenty-week fetal anatomy scan, bilateral deformities of the fetal forearms with upper extremity shortening was noted. There was visualization of one upper extremity long bone bilaterally and the hands and fingers were internally rotated. The patient proceeded with amniocentesis which resulted in a female fetus with thrombocytopenia absent radius. She opted to continue the pregnancy and delivered via cesarean section at thirty-eight weeks at which she went into labor.

Keywords: Congenital Anomaly; Thrombocytopenia; Absent Radius; 1q21.1 Microdeletion; RBM8A Gene; Radial Aplasia; Hypomegakaryocytic; Amegakaryocytic Thrombocytopenia; Thrombocytopenia-Absent Radii Syndrome; TAR Syndrome; Hypoplastic Megakaryocytes; Rare Disorder; Prosthesis; Prosthetic Syndrome Tetraphocomelia-Thrombocytopenia; Autosomal Recessive; Cmpl Signalling Pathway; Sonogram; Anatomy Scan; Short Forearm; Shortened Ulna; Radial Deviation; 5-Digit Hand; Present Thumb.

radius bone bilaterally, with bilateral thumbs usually present. The thrombocytopenia that occurs in thrombocytopenia absent radius syndrome is often transient and is defined as less than 50 platelets/nL, usually due to an underproduction of platelets.

Here we present a case of a 33-year-old African-American female, G6P3023 with last menstrual period on February 24, 2019 with no significant past medical or surgical history that was seen in our women's health clinic after finding out she was pregnant via a home pregnancy test. Prenatal labs were sent and urine pregnancy test was positive at that time. A pap smear was performed and found to be normal, a beta human chorionic gonadotropin (bHCG) serum level was performed and found to be 4088. Urinalysis and urine culture were sent and found to be negative. The patient expressed that this was an unplanned pregnancy however it was desired. She had no other complaints at that time. Her obstetric history included three normal spontaneous vaginal deliveries, one spontaneous abortion in the first trimester, and one elective termination of pregnancy.

She was given miscarriage precautions as well as prenatal vitamins and scheduled for follow up in two weeks. Upon return to our clinic additional prenatal labs were drawn and found to be unremarkable. She had no complaints at that time and was referred for a nuchal translucency sonogram at our antepartum testing unit. Upon ultrasound testing, the fetus was found to be 12 weeks and 0 days based on the crown rump length (CRL), giving her an estimated due date of December 02, 2019. The fetal heart rate was 153 beats per minute (bpm) and the nuchal translucency (NT) was found to be 0.87mm. Her cervix was within normal limits and she was told to follow up for 20-week anatomy scan.

The patient returned to our clinic at 16-weeks' gestation at which time the quad screen was performed and the results were unremarkable.

At 20 weeks and 3 days the patient had the anatomy scan performed. The fetus was noted to be in the breech presentation, fetal heart rate was 143bpm and the placenta was noted to be anterior/low lying. Fetal biometry revealed the following: biparietal diameter (BPD) 56th percentile, head circumference (HC) 40th percentile, abdominal circumference (AC) 32nd percentile, and femur length (FL) 15th percentile,

Case Report

Thrombocytopenia-absent radius (TAR) syndrome is a rare congenital condition which presents with megakaryocytic thrombocytopenia and associated aplasia of the radius bone [1]. TAR typically presents with truncated upper extremities and a radial deviation of both hands due to the absence of the

which calculated an estimated fetal weight in the 35th percentile. Bilateral deformities of the fetal forearms with upper extremity shortening was noted at that time. There was visualization of one upper long bone extremity bilaterally and the hands and fingers were found to be internally rotated bilaterally (**Figure 1-3**). The humeri bones were measured at the 36th percentile and the ulnae were measured at less than the 5th percentile.

The patient denied any history of congenital abnormality in her family or her partners' family. The patient also denied consanguinity in her union. The patient was counseled on confirmatory testing and opted to have an amniocentesis. Amniocentesis was performed with ultrasonic guidance and the specimen was sent for fetal karyotype, Fluorescence in Situ Hybridization (FISH) and microarray analysis. Fetal echocardiogram was also requested to rule out any cardiac abnormalities.

She presented at the antepartum testing unit two weeks later for another sonogram at which point she was 22 weeks and 3 days. On sonography, the fetus was found to be in the cephalic presentation, with a heart rate of 147bpm. Shortened forearms and absent radii were noted bilaterally. The results of the amniocentesis and microarray were consistent with a female fetus with thrombocytopenia absent radius syndrome. These findings were discussed with the patient as well as management options. The patient elected for continuation of the pregnancy. She was counseled on the need for primary low transverse cesarean section at 39 weeks to avoid any birth trauma to which she was in agreement.

When the patient was 38 weeks and 5 days she presented to the hospital in labor. She was taken to the operating room for primary low transverse cesarean section. The neonate was delivered atraumatically with the pediatrics team present. APGARS at birth were 9 and 9 at 1 and 5 minutes respectively. The neonate was brought to the Neonatal Intensive Care Unit (NICU) for monitoring. Imaging modality using x-ray was performed of the bilateral upper extremities which revealed bilateral absence of the radial bone. The right hand was grossly normal, however brachydactyly was noted on the left hand. She had an echocardiogram as well as a renal ultrasound performed, both of which were normal. There were no abnormalities of the lower extremities noted. The neonate was mildly thrombocytopenic with platelets ranging from 80,000 to 130,000/microliter. She did not require any sort of blood or platelet transfusion nor any orthopedic intervention. She was discharged home on day of life 5 in good condition.



Figure 1: Deformity of the fetal forearm with upper extremity shortening. Visualization of one upper extremity long bone.



Figure 2: Deformity of the fetal forearm with upper extremity shortening. Visualization of one upper extremity long bone.



Figure 3: Bilateral deformities of the fetal forearms with upper extremity shortening. Visualization of one upper extremity long bone bilaterally with the hands and fingers internally rotated bilaterally.

Discussion

Thrombocytopenia absent radius can cause a plethora of problems during the first few years of a child's life including but not limited to cow's milk allergy, significant symptomatic bleeding, platelet transfusions, and developmental abnormalities. Avoidance of cow's milk is recommended to reduce the severity of gastroenteritis and also to prevent the worsening of thrombocytopenia [2]. Bleeding can be quite common during childhood due to coagulation dysfunction. In patients without extreme thrombocytopenia (platelet count $>10/nL$), platelet transfusion can be avoided to decrease the risks of alloimmunization and infection. Platelet count should be performed any time evidence suggesting increased bleeding tendency such as bruising or petechiae is found [2]. If

platelet transfusion does need to be performed for thrombocytopenia, it is recommended that a central venous catheter be used as an alternative to repeat venipunctures. Teenage and adult women may experience menorrhagia because of this. Motor development is often delayed due to the underlying absent radii and shortened upper extremities.

References

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