The Importance of Fetal Gender Determination

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INTRODUCTION

Disorders of sex development (DSD), formerly termed ambiguous genitalia, are a group of conditions characterized by abnormal external genitalia. These abnormalities have the potential for negative emotional and social implications for parents. DSD abnormalities sufficient to prompt evaluation occur in approximately one in 4500 live births.1 The most common cause of DSD is congenital adrenal hyperplasia (CAH).2 In the United States of America (USA), the prevalence of CAH is lower in African-Americans than in Caucasians (1 in 42,000 versus 1 in 15,500, respectively).3

Prenatal diagnosis of DSD can prepare the parents to prepare for the diagnosis of DSD and provide an opportunity to perform more investigations to confirm or exclude other fetal abnormalities. Current guidelines were reviewed to determine current recommendations for gender determination during prenatal sonogram. During the mid-trimester routine scan, if the fetus is found to be at an increased risk of DSD, the mother should be offered genetic counseling and amniocentesis.

CASE

A 39-year-old G3P0sab1Lw1 intranatal pregnancy at 31 3/7 weeks’ gestation was referred to maternal fetal medicine (MFM) for consultation for a pregnancy complicated by suspected fetal skeletal dysplasia and intrauterine growth restriction based on outside sonograms. The pregnancy was also complicated by advanced maternal age and a history of pulmonary embolism managed with prophylactic anticoagulation. The patient had declined prenatal diagnosis and declined sonographic assessment for fetal gender earlier in the pregnancy.

The sonogram in MFM unit showed a fetus whose biometry was small for the assigned gestational age with an overall mean estimated fetal weight well below the 3rd percentile. A skeletal dysplasia was not suspected. The umbilical artery Doppler was elevated with a systolic to diastolic ration of 7.35. The amniotic fluid volume was within normal limits. Given the sonographic findings, the patient was admitted to the hospital for fetal monitoring, betamethasone therapy, and magnesium sulfate administration for cerebral palsy prophylaxis with the expectation of preterm delivery. Cesarean delivery was performed at 31 6/7 weeks’ gestation for non-reassuring fetal testing and abnormal follow-up umbilical cord Doppler.

The baby was found to be small for gestational age with a weight of 1120 grams. Appgar Scores were 8 and 9 in 5 and 10 minutes respectively. Most notably, the neonate was found to have abnormal genitalia and was diagnosed with a DSD. (Figures 1, 2, 3). The parents expressed emotional distress having to deal with the uncertainty in the baby’s gender identity. Both obstetrical care providers and neonatal care providers were as stunned as the parents. Both groups of providers were less than emotionally helpful to the parents and family immediately following delivery, taking a much more medical approach to the situation. The patient was discharged home on postpartum day 4 without gender identification. The parents were unable to announce gender to family and friends which was extremely distressing.

DISCUSSION

Executive summary statement of a Joint Eunice Kennedy Shriver National Institute of Child Health and Human Development, Society for Maternal-Fetal Medicine, American Institute of Ultrasound in Medicine, American College of Obstetricians and Gynecologists, American College of Radiology, Society for Pediatric Radiology, and Society of Radiologists in Ultrasound Fetal Imaging Workshop, reported that fetal genitalia screening should be a part of standard fetal ultrasound examination only in multiple gestations and when medically indicated.4 The International Society of Ultrasound in Obstetrics and Gynecology in 2010 practice guidelines for routine mid-trimester fetal ultrasound scan states that characterization of external genitalia to determine fetal gender is not considered mandatory in the context of a mid-trimester routine scan. Reporting of gender should be considered only with parental consent and in the context of local practices.5 The Society of Obstetricians and Gynecologists in Canada in a 2007 policy statement stated that review of the fetal perineum, including sex determination, is considered part of the complete obstetric ultrasound.6 However, if no fetal abnormalities are identified but determination is inconclusive, the examination should not be prolonged or repeated solely to determine fetal gender.7 The Royal College of Obstetricians and Gynecologists in London, United Kingdom does not include review of the genital organs as a part of the baseline fetal anomaly scan.8

Not surprisingly, the prenatal screening for many fetal disorders and abnormalities has become commonplace and standard of care. Currently, pregnant woman in the USA are offered prenatal screening through a combination of maternal serum screening, non-invasive prenatal testing for any reason and correlating the gender with screening results.9 Mid-trimester detection of DSD would prepare physicians and parents on the postnatal management course of working up common causes of disorders of sex development.

TEACHING POINTS

• Characterization of external genitalia to determine fetal sex should be mandatory in certain populations in the context of a mid-trimester routine scan, these populations are: Pregnant women who underwent invasive or non-invasive prenatal testing for any reason and correlating the gender with screening results.

REFERENCES