Three-dimensional ultrasound of massive macroglossia in a fetus with Beckwith-Wiedemann syndrome

Abstract: We present the prenatal ultrasound findings of massive macroglossia in a fetus with prenatally diagnosed Beckwith-Wiedemann syndrome. Three-dimensional surface mode ultrasound was utilized for enhanced visualization of the macroglossia.

Keywords: Beckwith-Wiedemann syndrome; macroglossia; prenatal diagnosis; three-dimensional ultrasound.

Introduction

Beckwith-Wiedemann syndrome (BWS) is an imprinting growth disorder that is associated with macroglossia, abdominal wall defect, and macrosomia. We present a case of BWS that was prenatally diagnosed based on two-dimensional (2D) and three-dimensional (3D) ultrasound depicting massive fetal macroglossia.

Case report

A healthy 32-year-old pregnant African woman, gravida 3, para 2, was referred to our tertiary antenatal unit at 20 weeks of gestation for a second opinion. The fetal anatomy ultrasound examination was abnormal, showing macroglossia, an omphalocele, enlarged kidneys, placentomegaly, polyhydramnios, and a suspected ear lobe groove (Figure 1A). The sonographic findings were suggestive for BWS. An amniocentesis was performed, which revealed a normal cytogenetic karyotype (46 XY). Molecular tests in chromosome 11p15.5 showed hypermethylation at gene H19 and hypomethylation at gene KCNQ1OT1, confirming BWS.

Serial prenatal ultrasound examinations revealed a large for gestational age fetus (estimated fetal weight and abdominal circumference exceeding the 95th percentile) with a permanently opened mouth, a protruding tongue (Figures 2 and 3A), and increasing amounts of amniotic fluid. At 30 weeks of gestation, an amnioreduction was performed as a result of maternal discomfort. At 35 weeks, an urgent cesarean delivery was performed because of fetal bradycardia and vaginal bleeding after preterm rupture of membranes. The macrosomic male infant was born weighing 3700 g (>97th percentile), and had Apgar scores of 6 and 8 at 1 min and 5 min, respectively. Cord blood arterial pH was 7.05 due to partial placental abruption. The placenta was enlarged weighing 1700 g (three times the expected weight). The newborn had massive macroglossia (Figure 3B) and showed distinctive ear creases on the lobes (Figure 1B).

Suggesting that airway management of the newborn would be difficult, a pediatric anesthesiologist was present at the delivery room and fiberoptic intubation available. After initial face mask ventilation, a laryngeal mask airway was inserted without difficulty. The newborn was immediately transferred to the neonatal intensive care unit and uncomplicated nasotracheally reintubated. Complete resolution of the omphalocele was achieved by means of external compression. At the 20th day of life, the abdominal wall defect was closed in one step. The postnatal course was complicated by hypoglycemia, severe respiratory distress syndrome, pulmonary hypertension requiring transient nitric oxide ventilation and bilateral intraventricular hemorrhage grade II-III°, and ultimately
A B

Figure 1  Infants with Beckwith-Wiedemann syndrome frequently have anterior linear ear lobe creases. Three-dimensional surface mode at 30 weeks (A) and the postnatal findings (B).

Fatal colonic necrotizing enterocolitis 32 days after birth. A postmortem examination was performed and confirmed visceromegaly, intraventricular hemorrhage II°, and perforated enterocolitis.

Discussion

BWS is a rare congenital overgrowth disorder with a reported incidence of approximately 1 in 13,500 births [8]. The etiology of BWS is complex. About 85% of affected individuals experience a single occurrence and 15% demonstrate an autosomal dominant mode of inheritance.
with incomplete penetrance. Macroglossia, defined as a resting tongue that extends beyond the alveolar ridge or teeth [22], is the most common finding in children with BWS [4, 16]. Surgery for macroglossia is often considered but should not be performed before 6 months of age [11].

Children with BWS also have an increased risk for certain embryonal tumors, most commonly Wilms’ tumor but also hepatoblastoma, neuroblastoma, and adrenocortical carcinoma. The different molecular subgroups of BWS are correlated with different risks of developing specific types of tumor [2]. The abnormal methylation variant found in the present case carried a 30% risk for embryonal tumor [17]. Therefore, the care plan for BWS children includes a vigilant tumor screening protocol involving abdominal ultrasound and measurement of serum α-fetoprotein every 3 months until the age of 8 years [20].

The majority of BWS cases are diagnosed after birth. The literature describes approximately 20 cases that were diagnosed prenatally, and there is no consensus on pre- and postnatal diagnostic criteria for BWS [23]. Prenatal 3D ultrasound observation of macroglossia in fetuses with BWS has been previously reported in two case reports. Storm et al. described the successful prenatal diagnosis of the protruding tongue with the assistance of 3D ultrasound [19]. In 2011, Eckmann-Scholz et al. published the prenatal findings using 3D sonography [3].

Most cases of BWS develop well, both physically and mentally, however, approximately 20% die during the perinatal period due to airway compromise, severe hypoglycemia, prematurity, and cardiomyopathy. It is possible that this rate could be reduced by improved prenatal diagnosis to ensure arrangements for delivery in a tertiary care center to enable better neonatal management of hypoglycemia and potential airway obstruction.

Upper airway obstruction and difficulties in endotracheal intubation as a result of an enlarged tongue has been reported from several authors in children with BWS [9, 10, 21]. Although the intubation with laryngoscope has been successfully used in children with macroglossia [5, 18], there is high chance of difficult or failed intubation and multiple laryngoscopy attempts may result in bleeding and edema of the upper airway. Goldman et al. reported the unproblematic airway control with the laryngeal mask in an infant with BWS [6] and its use has been advocated in children with potentially difficult airways [1, 7]. Assuming a massive macroglossia as prenatally visualized by 3D ultrasound in our case, we emphasized to avoid excessive airway instrumentation, minimizing the risk of trauma and airway obstruction by using a laryngeal mask as first choice to manage neonatal respiratory distress. The option of the “ex utero intrapartum treatment” procedure was prenatally discussed in our team. This technique allows access to the fetal airway in a controlled and secure manner, while the baby is under uteroplacental support. The procedure is an ideal delivery strategy for fetuses with prenatally diagnosed oropharyngeal tumors, for example, but so far, it has not been reported in newborns with macroglossia [12, 14]. Although an obstructed airway was anticipated, we did not have difficulty in airway management, however, the necessary preparations and precautions were taken.

Necrotizing enterocolitis is a known complication of gastroschisis but is rarely seen after omphalocele repair [15]. It has been reported as short-term morbidity in cases of giant omphaleoceles [13].

The present report describes the ultrasound findings of massive macroglossia in a prenatally diagnosed BWS using 3D surface mode, which allowed clear depiction of the fetal face and appropriate preparation for potential neonatal respiratory distress.

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References


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