Case Report

A Case of Beckwith-Wiedemann Syndrome with Polyhydramnios

Suatu Kasus Sindrom Beckwith-Wiedemann dengan Polihidramnion

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Abstract

Objective: To report a rare case of Beckwith-Wiedemann Syndrome with polyhydramnios.

Methods: Reporting a case of Beckwith-Wiedemann syndrome with polyhydramnios.

Results: Case of Mrs. Y, 27 years old woman, $G_2P_1A_0L_1$ preterm pregnancy (30-31 weeks) with polyhidramnios. From ultrasound found renomegaly, bilateral hyperechogenic polycystic kidney, and the karyotype result was 46,XX. Caesarean section was performed due to fetal distress. A female baby was born by caesarean section with birth weight of 1300 grams, 37 centimeters of body length, and APGAR score of 6/8. The congenital anomalies found were hepatomegaly, renomegaly, bilateral hyperechogenic renal polycystic, low set ears. The baby was died in NICU on day care 5th, with suspected of sepsis.

Conclusion: Prenatal diagnosis of Beckwith-Wiedemann syndrome on fetus with polyhydramnios.

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Keywords: amniocentesis, beckwith-wiedemann syndrome, polyhydramnios, prenatal diagnostic, USG

Abstrak

***Tujuan**: Melaporkan kasus sindrom Beckwith-Wiedemann pada polihidramnion.*

***Metode**: Melaporkan kasus sindrom Beckwith-Wiedemann dengan polihidramnion.*

***Hasil**: Kasus Ny. Y usia 27 tahun, $G_2P_1A_0H_1$ gravid preterm (30 - 31) minggu dengan polihidramnion. Pada pemeriksaan ultrasonografi dijumpai renomegali, hiperekogenisitasginjal, polikistik ginjal bilateral, hasil analisa kromosomnya 46,XX. Dilakukan seksio sesarea atas indikasi fetal distres. Lahir bayi perempuan, BBL 1300 gram, PBL 37 sentimeter, Apgar Skor 6/8. Terdapat kelainan kongenital: hepatomegali, renomegali, polikistik ginjal bilateral hiperekogenik, dan low set ear. Bayi meninggal pada hari kelima rawatan di NICU dengan dugaan sepsis.*

***Kesimpulan**: Diagnosis prenatal Beckwith-Wiedemann syndrome dengan polihidramnion.*

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***Kata kunci**: amniosintesis, beckwith-wiedemann syndrome, polihidramnion, USG*

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INTRODUCTION

Polyhydramnios is defined as AFI ≥ 25 centimeters or maximum vertical pocket (MVP) ≥ 8 centimeters at AFI <25 centimeters.[1] The incidence of polyhydramnios is approximately 1-2% in pregnancy. The etiologies of polyhydramnios include fetal anomalies, maternal diabetes, and idiopathic. It is estimated that 50% of cases are idiopathic. Polyhydramnios is known to be associated with an increased risk of poor pregnancy outcomes such as preterm labor, aneuploidy, sectio cesarean, and perinatal death. Congenital disorder that accompanies the disorder of the central nervous system, gastrointestinal tract, heart, and genitourinary tract.[2,3]

The incidence of abnormalities of the urinary tract that is 1-4 in 1000 pregnancies, and 15-20% of congenital abnormalities when prenatal diagnosis.[4,5] Bilateral enlarged hyperechogenic fetal kidneys are considered as diagnostic dilemma. If accompanied with oligo-or anhidramnios, the prognosis is usually very ugly.[6] Polyhydramnios accompanied with bilateral enlarged hyperechogic fetal kidneys renomegaly may be caused by some disorders including Beckwith-Wiedemann syndrome, Perlman Syndrome, Adult Polycystic Kidney Disease, Infantile Polycystic Kidney Disease, and Trisomy 13.[6] We present a case of patient with Beckwith-Wiedemann syndrome.

CASE

A 27-year-old femalewas referred to Dr. M. Djamil Padang Hospital on October 26, 2015 due to shortness of breath. She was diagnosed with $G_2P_1A_0H_1$ preterm pregnancy 30-31 weeks, polyhydramnios, intrauterine single live fetus. Ultrasound examination revealed fetal biparietal diameter of 75 milimeters, humerus length of 49 milimeters, femur length of 60 milimeters, abdominal circumference of 258 milimeters, estimated fetal weight of 1200 to 1300 grams, amniotic fluid index (AFI) 35.5 centimeters. Ultrasonography examination demonstrated bilateral enlarged polycystic hyperechogenickidneys.

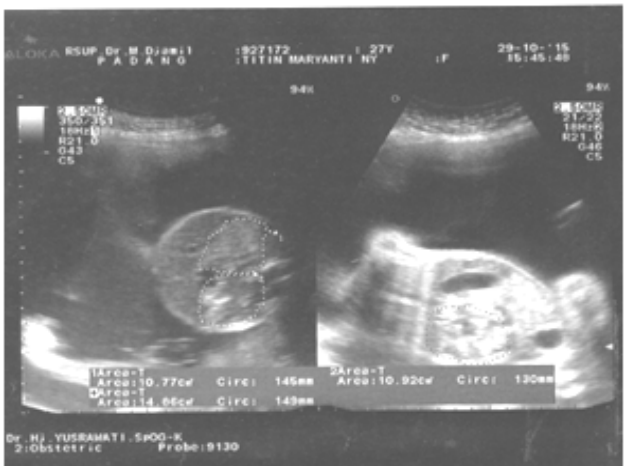


Figure 1. Polyhydramnios. Renomegaly, Polycystic Kidney Bilateral

Amnioreduction and amniotic fluid analysis were performed. The analysis revealed a karyotypes of 46,XX. No major structural abnormality was found (Figure 2).

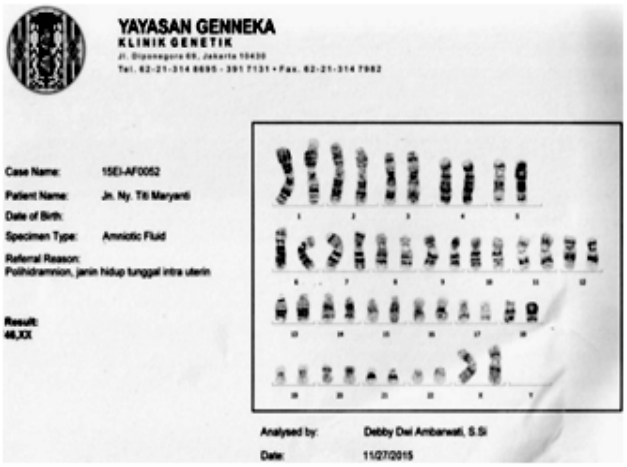
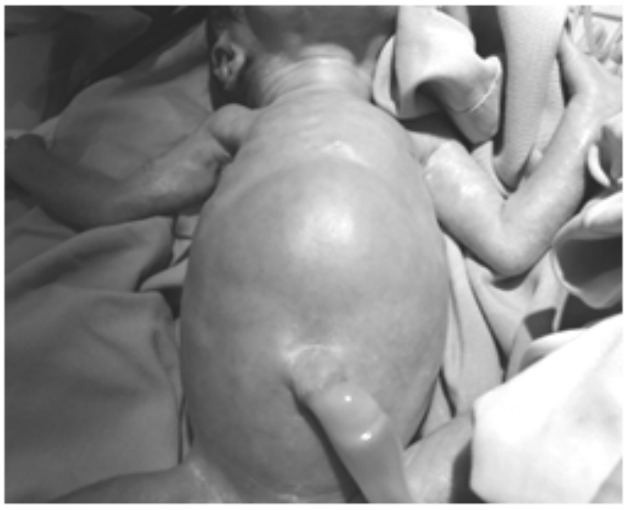
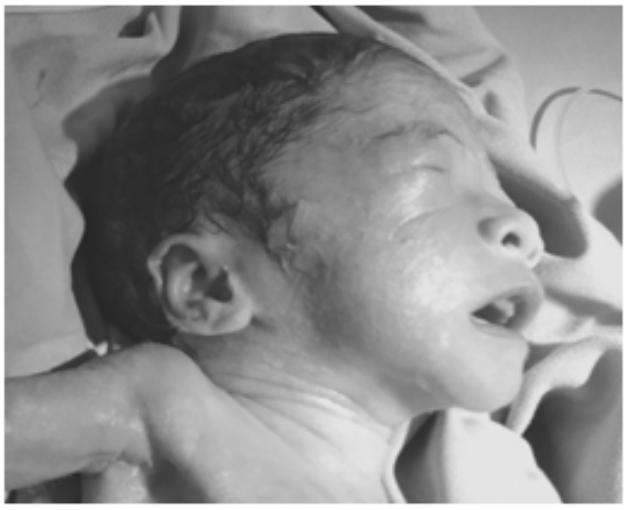


Figure 2. The Results of Chromosome Analysis

During treatment, a non reassuring NST (nonstresstest) was observed. Subsequently, cesarean section was performed. A baby girl was born with birth weight and body length of 1300 grams and 37 centimeters, respectively. Her Apgar score was 6/8. Low set ears, anterior ear creases, hepatomegaly, and renomegaly were present. The placenta was 14 x 13 x 3.5 centimeters, weighing 350 grams, the cord length was 40 centimeters, paracentral insertion, no abnormality found in the



Hepatomegaly and renomegaly



Anterior ear lobe creases (+)

Figure 3. Hepatomegaly, Renomegaly and Ear Lobe Creases of the Baby.

placenta and umbilical cord. Baby was cared in the NICU due to respiratory distress which was caused by hyaline membrane disease.

DISCUSSION

In our case, the patient, Mrs. T, 27 years old, diagnosed with polyhydramnios at 30-31 weeks of pregnancy. Ultrasonographic examination revealed AFI of 35.5 centimeters (severe polyhydramnios), renomegaly, hyperechogenic and bilateral polycystic kidneys.

The analysis demonstrated a karyotypes of 46,XX. Congenital abnormalities may be present in these patient closer towards Beckwith-Wiedemann syndrome (BWS), which is an abnormality in the short arm of chromosome 11, 11p15 which causes over activity of gene IGF-2 and / or the absence of copying active CDKN1C (inhibitor of cell proliferation).

For the diagnosis of BWS, there should be three major findings or two major and one minor finding. The findings are listed in Table 1.[7] In this case, the major findings that we found were anterior ear lobe creases (folds in front of the ear lobe), visceromegaly (renomegaly, hepatomegaly), and the minor finding is polyhydramnios.

Chitty LS et al suggested the differential diagnosis in case of enlargement hyperechogenic kidney with normal amniotic fluid or increased, which is shown in Table 2.

Table 1. Major and Minor Findings of Beckwith-Wiedemann Syndrome

Major findings
Abdominal wall defect: omphslocele (exomphalos) or umbilical hernia
Macroglossia
Macrosomia (traditionally defined as height and weight > 97th percentile)
Anterior ear lobe creases and/or posterior helical pits (bilateral or unilateral)
Visceromegaly of intra-abdominal organ(s) (e.g. liver, kidney(s), spleen, pancreas, and adrenal glands)
Embryonal tumor in childhood.
Hemihyperplasia.
Cytomegaly of adrenal fetal cortex, usually diffuse and bilateral.
Renal abnormalities, including medullary dysplasia and later development of medullary sponge kidney (MSK).
Positive family history of BWS.
Cleft palate.
Minor findings
Pregnancy related findings of polyhydramnios, enlarge placenta and or thickened umbilical cors, premature onset of labor and delivery.
Neonatal hypoglycemia.
Nevus flammeus
Cardiomegaly/structural cardiac anomalies/cardiomyopathy
Characteristic facies
Diastasis recti
Advanced bone age

Table 2. Overview of the Prenatal Diagnosis of Fetal Echogenic Kidney Enlargement.[6]

	Beckwith*	Perlman	APCKD	IPCKD	Trisomy 13
Renomegaly	+	+	+	+	+
Increased echogenicity	+	+	+	+	+
Renal cysts	±	-	+	-	-
Mild hydronephrosis	-	±	-	-	-
Polyhydramnios	+	+	-	-	-
Oligohydramnios	-	-	rare	common	-
Generalized organomegaly	+	+	-	-	-
Macrosomia	+	+	-	-	-
Macroglossia	+	-	-	-	-
Hemihypertrophy	+	-	-	-	-
Omphalocele	+	-	-	-	-
Other anomalies	-		-	-	+
Consanguinity	-	+	-	+	-
Family history in parent/grandparent	±	-	+	-	-
Family history in sibling	±	+	-	+	-

**Beckwith=Beckwith-Wiedemann syndrome; APCKD=adult polycystic kidney disease; IPCKD=infantile polycystic kidney disease*

Amnioreduction was performed due to breathlessness and abnormal maternal chromosomes examination. During treatment, fetal distress was observed, thereby cesarean section was performed. According to the literature genetic examination of Beckwith-Wiedemann syndrome is very complex, causing difficulties in molecular diagnosis, and diagnosis is based on clinical findings.[8]

Patient was discharged on the 3rd day post-operative with good condition, while the baby died on day 5th of treatment due to sepsis. Severe polyhydramnios is associated with unfavorable outcomes, including as low Apgar score, fetal death, fetal distress, and neonatal death.[9] Weksberg R et al suggested that infant death could arise from complications such as hypoglycemia, prematurity, cardiomyopathy, macroglossia, or tumor.[7]

CONCLUSION

Beckwith-Wiedemann syndrome is a rare disorder caused by a defect on chromosome 11p15 mold. Prenatal diagnosis using ultrasound is necessary to detect abnormal findings.

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