CASE REPORT

Beckwith-Wiedemann Syndrome- A Rare Case Report

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Abstract

Beckwith-Wiedemann syndrome is a pediatric overgrowth disorder with classical features of macroglossia, exomphalocele and gigantism. Estimated incidence rate of BWS is 1in 13,700 in population. The incidence of BWS is equal in both the sexes. We got a rare case of Beckwith-Wiedemann syndrome in our NICU. The diagnosis is made by clinical findings suggestive of BWS. Baby born with a macroglossia and abdominal wall defects (exomphalocele), ear anomalies, renal abnormalities seen under ultrasound and neonatal hypoglycemia. Additional supportive findings were polyhydramnios and LGA baby, cardiomegaly, hemangioma over face, enlarged placenta and characteristic facies and infraorbital creases.

Key Words

Beckwith-Wiedemann syndrome, Macroglossia, Wilms tumour, Exomphalos

Introduction

BWS is a congenital overgrowth disorder. Beckwith Wiedemann Syndrome (BWS) represents a genetic syndrome of low prevalence and diverse clinical expression. The syndrome was first described by Beckwith (1) in 1963 and Wiedemann (2) in 1964. The syndrome is characterized by omphalocele, macroglossia, gigantism, neonatal hypoglycemia, hemihypertrophy, hepatosplenomegaly, nephromegaly, cardiac anomalies, adrenal cytomegaly, pancreatic islet cell hyperplasia, facial nevus flammeus, and ear lobe creases (2).

Case Report

A male newborn, product of normal vaginal delivery following uneventful 38 weeks normal pregnancy to a 27-year Gravida three mother who is not known to have any medical illness. He had an Apgar score of 8/10 at 1 and 10/10 at 5 min.

Physical examination revealed an active baby with the following anthropometric measurements: body weight 3.7 kg: length 51cm, HC-35cm. The baby had large tongue protruding out of the mouth *(Fig. 1)* and about 5x4 cm omphalocele with loops of intestine inside it. Overall physical examination showed unusual linear creases and pits over both ears. He had an ejection systolic murmur with grade of 2/6 over the lower left sternal border. Genitourinary evaluation showed both side retractile testis but otherwise normal male genitalia. The baby had no other abnormality and his general condition was stable.

Figure 1: Hemangiomata on Right Eyelid, Infraorbital Crease and Macroglossia

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His blood glucose levels were monitored regularly for the first 24 hours in NICU and had 2 episodes of asymptomatic hypoglycemia and were managed as per standard protocols.

Chest X-ray showed cardiac enlargement *(Fig. 2)*. Abdominal Sonography revealed that right kidney is enlarged in size than left kidney. Echo showed a tiny perimembranous VSD. His thyroid function tests (TFT) were normal.

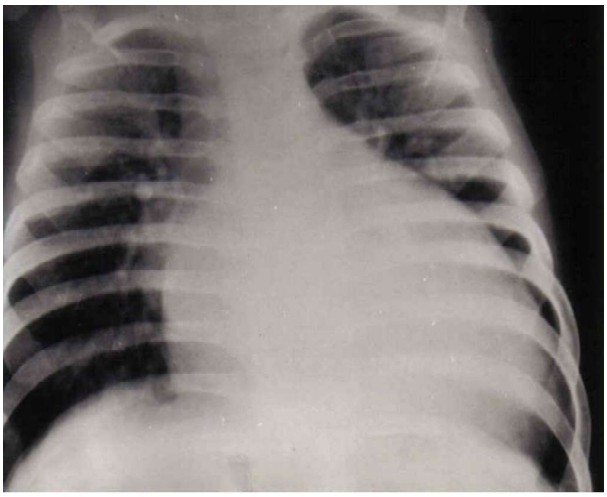

Figure 2: Chest X-Ray Showed Cardiac Enlargement

Pediatric Surgical consultation was done for the omphalocele, which was operated on the third day of life and managed postoperatively as per protocols. Baby was discharged in good condition and advised regular follow up for renal USG.

Discussion

BWS patient have increase chances of congenital abnormalities and medical complications, including abdominal wall defect, organomegaly, renal anomalies and cardiac malformations

There is a high risk (about 10%) of development of embryonal neoplasms, particularly Wilms tumor in the child with BWS, especially those with hemihypertrophy (3). Wilms tumor is the most common cancer in children with Beckwith-Wiedemann syndrome. It occurs in about 5-7% of all children with Beckwith-Wiedemann syndrome. Majority develop Wilms tumor prior to 4 years of age; however, children with Beckwith Wiedemann syndrome can develop Wilms tumor when they are as old as 7-8 years. By age 8 years, 95% of all Wilms tumor cases have been diagnosed (4).

Estimated incidence rate of BWS is 1in 13,700 in population. Although 85% of cases are sporadic there is also an inherited form, which has autosomal dominant inheritance with imprinting on the short arm of chromosome11(5). Imprinting has been associated with structural modifications of DNA near the gene, such as methylation or lack of acetylation. Several 11p genes are imprinted, including p57 (a cation independent cyclase), IGF-2 (the gene for insulin like growth factor-2 [IGF-2]), the gene for insulin, and H19 (6).

Unilateral or bilateral renal anomalies may include primary malformations, renal medullary dysplasia, nephrocalcinosis, and nephrolithiasis (7,8). Cardiac malformations are found in about 20% of children with BWS; approximately half manifest with cardiomegaly that resolves spontaneously (7).

Conclusion

Beckwith-Wiedemann syndrome is a rare type of congenital disorder. Early diagnosis and detection of intra abdominal malignancy should be prompt for better outcome.

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