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Case Report

Beckwith-Wiedemann Syndrome- A Rare Case Report

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ABSTRACT

Introduction: Beckwith–Wiedemann syndrome (BWS) is a pediatric overgrowth disorder presents with classical features of exomphalos, macroglossia, and gigantism. Estimated incidence rate of Beckwith wiedemann syndrome is 1 in 13,700 in population. The incidence of BWS is equal in males and females.
Presentation of Case: An 18 months female child was brought by parents with hypertrophy in the right upper and lower extremity since birth. Patient was delivered normally with no congenital malformations like exomphalos, macroglossia but she had frequent episodes of hypoglycemia. Ultrasonography (USG) of abdomen to rule out organomegaly and intraabdominal malignancy done which showing no abnormality.
Discussion: BWS patient have increase chances of congenital abnormalities and medical complications, including abdominal wall defects, organomegaly, renal anomalies and cardiac malformations. 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. Patients with Beckwith-Wiedemann syndrome (BWS) may require frequent feedings or diazoxide.
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. Phenotypic variability is more with BWS and thus certain diagnostic criteria are not fit for every patient.

Key words: Beckwith-Wiedemann syndrome; Macroglossia; Wilms tumour; Exomphalos.

INTRODUCTION

Beckwith–Wiedemann syndrome (BWS) is a pediatric overgrowth disorder involving an increase chance of tumor development. The clinical presentation is highly variable. Some cases lack the classical features of exomphalos, macroglossia, and gigantism as described by Beckwith and Wiedemann. [1,2] BWS is a panethnic disorder with an estimated incidence of 1 in 13,700. This figure is likely an underestimate as milder phenotypes may not be detected. The incidence is equal in males and females with the notable exception of monozygotic twins that show a dramatic female preponderance.

The major imprinted gene cluster, occurring on human chromosome 11p15.5, that has been implicated in the imprinting disorder Beckwith-Wiedemann syndrome (BWS) and in a variety of human cancers including Wilms' tumor. [3,4] Individuals with BWS may grow at an increased rate during the latter half of pregnancy and in the

first few years of life. Adult heights are generally in the normal range. [5,6] We report a 18 month old female child as a case of Beckwith Wiedemann Syndrome.

CASE REPORT

An 18 months female child was brought by parents with hypertrophy in the right upper and lower extremity since birth.(As shown in Figure 1 & 2) Patient was delivered normally. No congenital malformations noticed at birth like exomphalos, macroglossia. But patient had history of frequent episodes of hypoglycemia and for that she was admitted also. On examination there were no signs of cellulitis. On examination of oral cavity there were no macroglossia. (as shown Figure 3) We had investigated her for blood sugar estimation and ultrasonography(USG) of abdomen to rule out organomegaly and intraabdominal malignancy. But on USG no abnormality detected. Her blood sugar level again showed to lower level (55 gm/dL). Based on the findings we reach to diagnosis of BWS. It is a rare presentation of BWS as there is a lack of the hallmark features of exomphalos, macroglossia and Wilms tumour as described by Beckwith and Wiedemann.

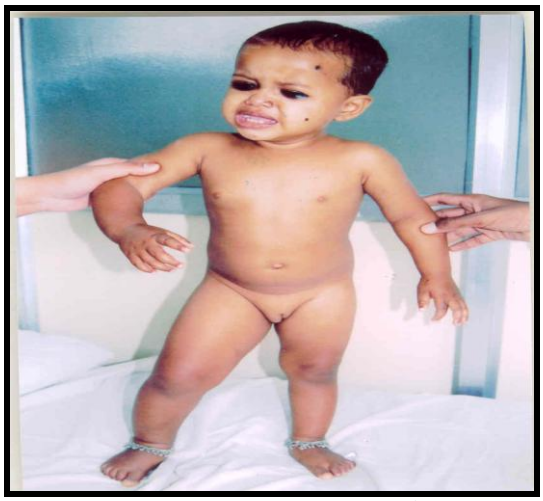

Figure 1: Photograph showing anterior view of a child having Beckwith Wiedemann Syndrome.

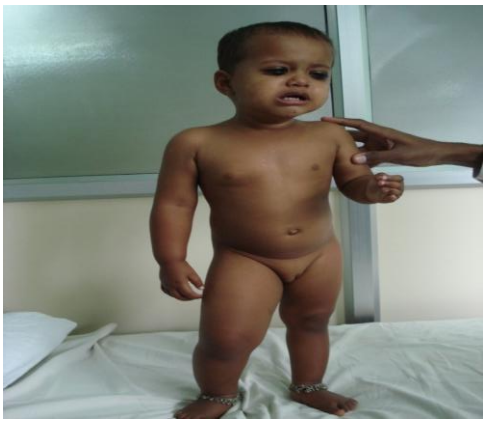

Figure 2: Photograph showing Right upper and lower limb hypertrophy.

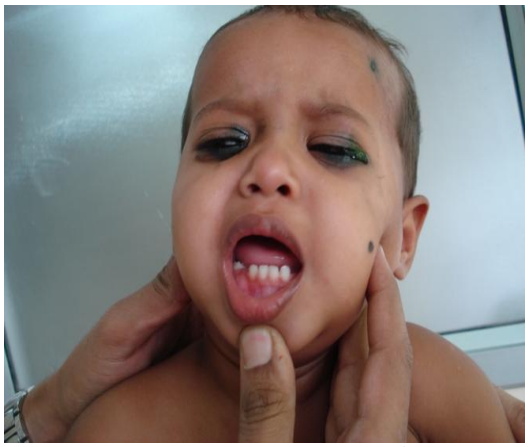

Figure 3: Photograph showing examination of oral cavity for macroglossia.

DISCUSSION

Individual person with BWS have an increased chances of congenital abnormalities and medical complications, including abdominal wall defects (omphalocele, umbilical hernia, and diastasis recti); organomegaly involving any single or combination of organs: liver, spleen, pancreas, kidneys, and adrenals. Fetal adrenocortical cytomegaly is a pathognomonic finding for BWS. Unilateral or bilateral renal anomalies may include primary malformations, renal medullary dysplasia, nephrocalcinosis, and nephrolithiasis. [7-9] Cardiac malformations are found in about 20% of children with BWS; approximately half manifest

cardiomegaly that resolves spontaneously. [5,10] Cardiomyopathy is rare.

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*. [11]

Beckwith-Wiedemann syndrome is a congenital disorder. 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. [12]

The cardinal features of Beckwith-Wiedemann syndrome include prenatal and postnatal overgrowth, [13] macroglossia, and anterior abdominal wall defects (exomphalos).

Variable findings include posterior helical indentations (pits of the external ear) and organ over growth, particularly hepatomegaly and nephromegaly.

Although mental retardation has been reported as a feature of Beckwith-Wiedemann syndrome, uncontrolled hypoglycemia during infancy may be a significant etiological factor.

Additional variable complications include organomegaly, hypoglycemia, hemihypertrophy, genitourinary abnormalities and in about 5-20% of children, embryonal tumors (most frequently Wilms tumor) and adrenal tumors such as adrenocortical neoplasias.

Patients with Beckwith-Wiedemann syndrome (BWS) may require frequent feedings or diazoxide to treat their hypoglycemia.

Embryonal tumors require appropriate oncologic treatment modalities, which often includes nephrectomy. Nephron-sparing partial nephrectomy is feasible if embryonal renal tumors are detected early. Macroglossia seldom requires resection to attain an independent airway. Macroglossia has been surgically reduced, with variable cosmetic outcomes. [14,15]

The BWS phenotype can present variably; for example, the diagnosis may be considered in a child presenting only with hemihyperplasia and nevus flammeus or possible ear creases, whereas the severe end of the spectrum may involve intrauterine, neonatal, or pediatric death. Death may be due to complications arising from hypoglycemia, prematurity, cardiomyo-pathy, macroglossia, or tumors.

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. Phenotypic variability is more with BWS and thus certain diagnostic criteria are not fit for every patient.

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