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CURIOUS CASE OF A TEASING TONGUE: A CASE REPORT ON BECKWITH WIEDEMANN SYNDROME(BWS).

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ABSTRACT

Beckwith Wiedemann Syndrome(BWS) is an overgrowth disorder with an incidence of 1 in 13,700 live birth. It is a multigenetic disorder caused by dysregulation of gene expression in the imprinted 11p15 chromosomal region. It is characterized by macrosomia, macroglossia, organomegaly and developmental abnormalities in infants. We present a case report of a fourday old full term male neonate, who presented to our hospital with respiratory distress, macroglossia and abdominal distension who on further investigation was diagnosed with Beckwith Wiedemann Syndrome.

KEYWORDS: Macrosomia, Visceromegaly, Hemihyperplasia, Overgrowth disorder.

INTRODUCTION

Beckwith Wiedemann Syndrome was presented independently by Beckwith (1963) and Wiedmann (1964). Beckwith Wiedemann Syndrome is a congenital overgrowth disorder.[1,2,3] It has an incidence of 1/13,700 live births, equal male to female ratio.[4] It is a multigenetic disorder caused by dysregulation of gene expression owing to mutation in the dominant 11p15.5. Neonates with BWS present usually with the combination of congenital abdominal wall defects as hernia (exomphalos), large tongues (macroglossia), and large bodies and/or long limbs (gigantism). In addition, some may have other findings like nevus flammeus, prominent occiput, midface hypoplasia, hemihypertrophy, genitourinary anomalies (enlarged kidneys), cardiac anomalies, musculoskeletal abnormalities, and hearing loss. Its challenging in diagnosing BWS owing to its low prevalence with clinical and genetic heterogeneity.

Case Presentation

A four day old, full term, male neonate, appropriate for gestational age, with a birth weight of 3.3kg, second order by birth was brought by parents in view of protruding tongue, increased respiratory activity and abdominal distention since birth. On examination the neonate had respiratory distress, poor peripheral pulses, delayed capillary refill time, icterus upto thigh and purulent discharge at umbilical stump. The neonate also had macroglossia (protruding tongue), distended abdomen, omphalocele with purulent discharge, hepatomegaly, bilateral palpable kidneys and a systolic murmur on auscultation.

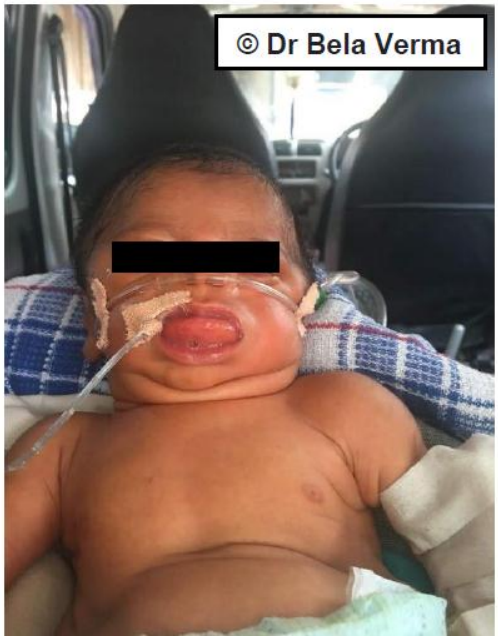

Figure 1.

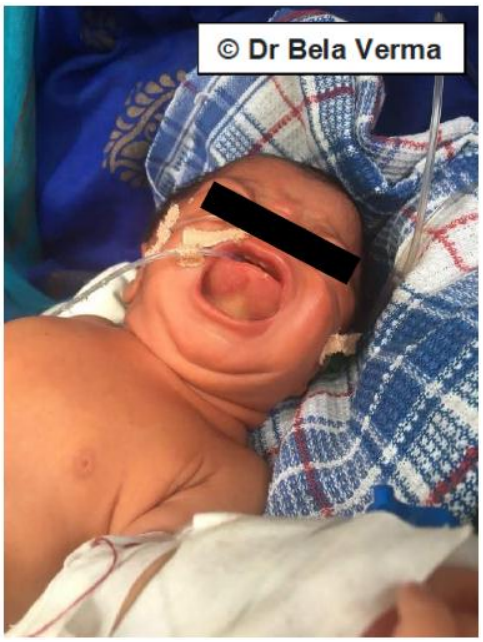

Figure 2.

Figure 1 and 2: The four day old neonate with the large protruding tongue (macroglossia).

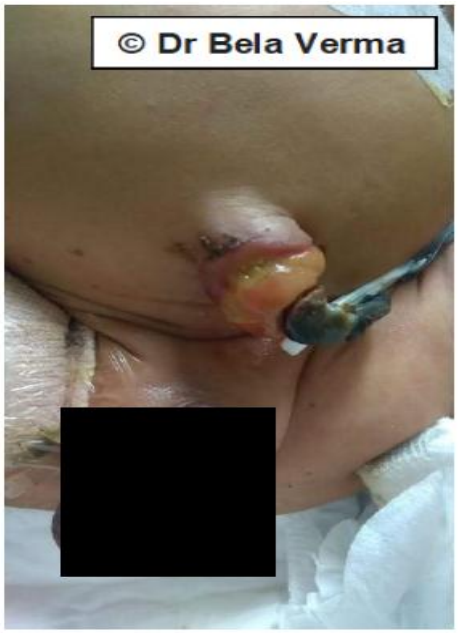

Figure 3: Soft tissue protrudingthrough the umbilical stump(Omphalocele).

On investigation significant findings included his HGT-41g/dl, Ultrasonography of abdomen showing hepatomegaly with bilateral nephromegaly, Chest X-Ray showing cardiomegaly and 2D ECHO showed a tiny PDA-2.5mm, small PFO with LR shunt with mild TR. Blood culture sensitivity had Candida Pelluculosa and Catheter tip culture showed MRSA. Random blood sugar levels were monitored on daily basis and maintained on high rates of glucose infusion therapy. With this clinical picture, a provisional diagnosis of Congenital Hypothyroidism and Beckwith Wiedemann Syndrome were considered. Thyroid function test of the neonate came within normal limits. In view of the hypoglycemia, critical blood samples were sent and it revealed an elevated S. Insulin (17.7mIU), normal S. Cortisol(6.70), increased Growth hormones (17.10) and an elevated A-fetoprotein level for the age. The clinical findings and the lab reports directed more towards Beckwith Wiedemann syndrome which was further confirmed by a positive genetic DNA methylation study showing EPIGENTIC/IMPRINTING DEFECT AT 11p15.5 locus.

Medical management of the neonate included treatment for hypoglycemia with intravenous glucose infusion, oxygen support, inotropes, intravenous antibiotics, antifungals, furosemide and phototherapy. The neonate however succumbed to septic shock.

DISCUSSION

Beckwith-Wiedemann syndrome (BWS) is a congenital overgrowth and cancer predisposition disorder. BWS is caused by changes on chromosome 11p15.5.[5,6] It is a multigenetic disorder caused by dysregulation of gene expression owing to mutation in the dominant 11p15.5 region proximal to genes for insulin: sur1 and igf2.[1,2] It includes geneduplication, loss of heterozygosity, loss of imprinting.

It has associated features like large for gestational age, macrosomia, macroglossia, organomegaly (liver, spleen, pancreas, kidneys, or adrenal glands) and abdominal wall defects (omphalocele, umbilical hernia, or diastasis recti). BWS may also be associated withlow blood sugar levels in the first few days of life or beyond leading to persistent low blood sugars, hyperinsulinism, ear pits, facial abnormalities, abnormal enlargement of one side or structure of the body resulting in asymmetric growth. Patients with BWS may have an enlarged tongue (macroglossia), which can cause difficulties in speaking, feeding, and breathing. They may also have prominent eyes with intraorbital hypoplasia, and/or a prominent occiput and facial nevus simplex.

A variety of renal abnormalities can occur including nephromegaly, renal medullary dysplasia, duplicated collecting system, medullary sponge kidney, diverticula and nephrocalcinosis which could impair kidney function.

They have an increased risk of developing certain childhood cancers like Wilms tumor, hepatoblastoma, neuroblastoma rhabdomyosarcoma and adrenal carcinoma.[6] The overall tumor risk is highest during the first two years of life. BWS is considered a clinical spectrum, in which affected individuals may have many of these features or may have only one or two clinical features. The phenotypic expression of Beckwith Wiedemann Syndrome is variable and diagnosis is still based on clinical signs.

Clinically BWS must be distinguished from other overgrowth disorder like Congenital Hypothyroidism and Simpson Golabi Behmai Syndrome. Cytogenetic analysis of 11p15 region (DNA Methylation Studies) and fluorescent in situ hybridization (FISH) can be used for recognizing epigenetic changes. Children conceived through IVF are more prone to BWS.

Management in BWS focuses on omphalocele repair, airway issues (a result of macroglossia) and neonatal hypoglycemia correction. Children with macroglossia may need to undergo tongue reduction surgery. Cancer risk is high until 8 years of age and regular surveillance with abdominal USG and measurement of a-fetoprotein is recommended every 3months until age of 8 years.[8] BWS children usually have normal intelligence and normal lifespan. By adolescence growth is normalized and risk of childhood cancers decreases.

Beckwith Wiedemann Syndrome is a genetic disorder and hence prenatal screening for pregnancies in the general population that identifies findings suggestive of a diagnosis of BWS may lead to the consideration of chromosome analysis, chromosomal microarray, and/or molecular genetic testing. Identification of the underlying genetic mechanism causing BWS permits better estimation of recurrence risk.

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Conflict of Interest: Nil.

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