

# CASE REPORT

## CASE REPORT OF BECKWITH-WIEDEMANN SYNDROME

Ravikanth K<sup>1</sup>, G. Somaiah<sup>2</sup>, G. Anusha<sup>3</sup>, D. V. Umesh Reddy<sup>4</sup>, R. Naveen Reddy<sup>5</sup>

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**ABSTRACT:** In 1963 Beckwith presented a report on the first patient with extreme cytomegaly of adrenal cortex, hyperplasia of kidneys as well as pancreas and Leydig cell hyperplasia. Wiedemann completed description of the new syndrome by adding umbilical hernia and macroglossia. The diagnosis is made based on the clinical signs of omphalocele.<sup>[7]</sup> or some other umbilical deformity, macroglossia.<sup>[8]</sup> congenital asymmetry, visceromegaly (liver, pancreas, and kidneys). Newborn with macrosomia, macroglossia first differential diagnosis are infant of diabetic mother, congenital hypothyroidism etc. IDM can be identified easily with history of maternal diabetes. So congenital hypothyroidism is next common diagnosis and will be treated as hypothyroidism. If TSH, T3 and T4 are not clearly indicative of congenital hypothyroidism next consider the Beckwith Wiedemann syndrome in differential diagnosis of large for gestational age.

**KEYWORDS:** Macroglossia, Beckwith Wiedemann syndrome, glossectomy.

**INTRODUCTION:** Beckwith-Wiedemann Syndrome (BWS)<sup>[9]</sup> is an overgrowth, multigenic disorder caused by dysregulation of the expression of imprinted genes in the 11p15 chromosomal region.<sup>[11,12]</sup> It is characterized by Omphalocele.<sup>[7]</sup>, Gigantism, macroglossia, microcephaly and visceromegaly.<sup>[1]</sup> In an attempt to standardize the classification of BWS,<sup>[9]</sup> De Baun et al. have defined a child as having BWS if the child has at least two of the five common features (macroglossia, macrosomia, midline abdominal wall defects, ear creases/ear pits, neonatal hypoglycemia.<sup>[2]</sup>

Another definition presented by Elliot et al. includes the presence of either three major features (anterior abdominal wall defect, macroglossia,<sup>[8]</sup> or prepostnatal overgrowth) or two major plus three minor findings (ear creases and pits, facial nevus flammeus, neonatal hypoglycemia,<sup>[13]</sup> nephromegaly,<sup>[10]</sup> or Hemihyperplasia, embryonal tumors, polyhydramnios).<sup>[3]</sup>

**CASE REPORT:** A 4years-old female child presented to Pediatric outpatient of Mamata Medical College with the chief complaints of abdominal distension for the last 3 months, mass per abdomen for 4 days. She was apparently normal 3 months back when the parents noticed abdominal distension which was gradually increasing. Instinctly child's father noticed mass in the abdomen 4 days back on palpation. There was no history of constipation.

History of loss of appetite and weight loss present. This child is a product of non-consanguineous marriage, full term, delivered by cesarian section because of a very bad obstetric history. Child of 10<sup>th</sup> pregnancy, 1<sup>st</sup> live child. Birth weight 4.35kgs, motor milestones delayed. On examination she was well built, large tongue with mouth open all the time (Figure 1). Her height 104cms expected 100cms. weight 20kgs expected 16kgs. MAC 18cms.

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**Figure 1: Siblings with large tongue and coarse features**



**Figure 2: CECT Abdomen showing large heterogenous mass lesion from left kidney**



**Figure 3: Plain X-Ray KUB showing enlarged left kidney**

**Systemic Examination:** Per abdomen examination revealed a smooth, non-tender, large mass with ill-defined borders occupying most of the left lumbar and hypochondriac regions. Hepatomegaly present, diastasis recti present. Respiratory, cardiovascular & central nervous systems appeared normal. Routine investigations CBC, CUE, chest X ray were normal except platelet count 6lakhs/cumm; X-Ray KUB plain suggesting enlarged left kidney (Figure 3). Abdominal ultrasonogram showing: 1. Mild hepatomegaly 2. Large heterogenous mass lesion (12.1 X 8.5 X 7.8cms) involving left lumbar & iliac fossa regions 3. Left mild hydronephrosis. IVU & CECT Abdomen were suggesting Nephroblastoma (figure 2). Chromosomal Analysis by Karyotyping showed normal female karyotype-46, XX. The above described clinical features and investigations are suggestive of Beckwith-Wiedemann syndrome. Due to financial constraints, genetic and molecular studies could not be performed.

**DISCUSSION:** BWS<sup>[9]</sup> incidence is 1 in 13, 700 about 300 children with BWS<sup>[9]</sup> are born each year in the United States.<sup>[4]</sup> However, the exact incidence of BWS is unknown because of the marked variability in the syndrome presentation and difficulties with diagnosis.>85% cases of BWS are

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sporadic, remaining <15% cases of BWS are familial. BWS<sup>[9]</sup> can be caused by a range of different genetic defects. These include cytogenetic abnormalities, genetic abnormalities [11p 15<sup>[11, 12]</sup> paternal uniparental disomy (UPD), mutations in the CDKN1C gene], epigenetic abnormalities [H19 gene, KCNQ10T1 gene & microdeletions within IC1 or IC2. 7.5 to 10% of BWS patients will develop tumor.

Wilm's tumor is the most common embryonal tumor in patients with BWS followed by hepatoblastoma.<sup>[5]</sup> children with BWS<sup>[9]</sup> are also at increased risk of developing adrenal cortical carcinoma, neuroblastoma, and rhabdomyosarcoma. Wilms tumor and hepatoblastoma can usually be cured if diagnosed early. All children with BWS<sup>[9]</sup> should receive cancer screening. An abdominal ultrasound every 3 months<sup>[6]</sup> until at least eight years of age is recommended and a blood test to measure alpha-fetoprotein (AFP) every 6 weeks until at least four years of age. Prognosis is good in these cases.

Our case was not diagnosed as a case of BWS<sup>[9]</sup> till 4 years of age and hence regular screening was not carried out. Child presented with wilms tumor. She was kept on chemotherapy for wilms tumor.

As child was having large tongue with open mouth and macrosomia child was misdiagnosed as congenital hypothyroidism and was kept on thyroxine from 10<sup>th</sup> day of life in a private hospital.

In conclusion, early recognition of BWS<sup>[9]</sup> is important because of its associated risk of malignancy. Prognosis for long term survival is good if these children are identified early and appropriately screened for malignancy

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### AUTHORS:

1. Ravikanth K.
2. G. Somaiah
3. G. Anusha
4. D. V. Umesh Reddy
5. R. Naveen Reddy

### PARTICULARS OF CONTRIBUTORS:

1. Post Graduate Student, Department of Paediatrics, Mamata Medical College & General Hospital, Khammam, Andhra Pradesh.
2. Professor of Paediatrics & Guide, Department of Paediatrics, Mamata Medical College & General Hospital, Khammam, Andhra Pradesh.
3. Post Graduate Student, Department of Paediatrics, Mamata Medical College & General Hospital, Khammam, Andhra Pradesh.
4. Post Graduate Student, Department of Paediatrics, Mamata Medical College & General Hospital, Khammam, Andhra Pradesh.

5. Post Graduate Student, Department of Paediatrics, Mamata Medical College & General Hospital, Khammam, Andhra Pradesh.

### NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Ravikanth K,  
Mamata Medical College & General Hospital,  
Khammam, Andhra Pradesh.  
Email: kanthmedico@gmail.com

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