**Case Report**

# A Case of Atypical Beckwith Weidemann Syndrome Spectrum with Uterine Hypertrophy, Hepatic and Renal Cysts and Myocardial Hypertrophy – A Case Report

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**Abstract :**

Beckwith Wiedemann Syndrome is a rare, genetic imprinting disorder involving the 11p15 chromosome, resulting in a wide variety of clinical features like macroglossia, anterior abdominal wall defects, hemihypertrophy and hypoglycaemia. Here we present a case of a new-born female with macrosomia, macroglossia,ear pits and generalised visceromegaly. Baby had no recording of hypoglycaemia. Ultrasound was suggestive of generalised visceromegaly including hypertrophic uterus. Simple cysts of liver and left kidney were noted. Baby was clinically stable and has hence been counselled for regular follow up for monitoring for development of tumours.

**Keywords:**

Beckwith Weidemann Syndrome, General Visceromegaly.

## Introduction

Beckwith Weidemann syndrome is a type of foetal overgrowth syndrome associated with increased predisposition to embryonal tumours. It is a spectrum of clinical features ranging from macroglossia, hypoglycaemia, omphalocele to other features such as visceromegaly, hemihypertrophy and predisposition to embryonal tumours especially of the liver and kidneys (1). International consensus on Beckwith Weidemann syndrome defined clinical diagnostic criteria based on cardinal and suggestive features and developed a point system for diagnosis of BWSp and further need of genetic testing (2). An increased risk of embryonal tumours warrants strict tumour surveillance in such cases.

## Case Presentation:

A female baby was born to a G2P1L1 mother at 38 weeks of gestation via lower segment caesarean section at Civil Hospital, Sangrur, Punjab, India. Baby cried immediately after birth but was referred to our centre in view of respiratory distress soon after birth and suspected multiple congenital anomalies. On admission the baby had a heart rate of 140beats per minute, respiratory rate of 68 breaths per minute, oxygen saturation of 98% on room air and a Downes score of 3/10. On examination, the baby was large for gestational age with a birth weight of 3.98kg (>90th centile) and had a head circumference of 34cm which was within normal limits. On general physical examination, macroglossia and ear pits were noticed. On per abdomen examination: abdomen was protuberant, hepatosplenomegaly and bilateral firm ballotable mass (?renal mass)was palpable. On respiratory system examination: minimal retractions were present. CNS examination revealed bilaterallower limb hypotonia. Upper limb tone, truncal tone and reflexes were within normal limits. Cardiovascular system examination was within normal limits. Baby was given oxygen support via nasal prongs for 1 day. Orogastric feeds were started by day 2 of life. Haematological investigations-

complete blood picture, coagulation profile, renal function tests and liver function tests were within normal limits. Septic screen done was positive for qualitative C-reactive protein, however I/T ratio and ANC were within normal limits. Thyroid function tests done were within normal limits. Close monitoring of blood glucose level was done;however, no reading of hypoglycaemia was noted. Ultrasound whole abdomen done was suggestive of generalised visceromegaly – hepatomegaly and nephromegaly. Liver measuring 5.9cm in size, normal in shape and echo texture, a simple hepatic cyst measuring 4.9 cm x2.9cm predominantly in segments IVa and VIII was noted. Spleen was 5.5cm in size, normal in shape and echo texture. Right Kidney measured 6.5 cm x3.3cm and the left kidney measured 6.5 cm x3.3cm in size. Left kidney had 2 anechoic lesions seen at upper pole, largest size 1.3cmx1.5cm suggestive of simple renal cysts. Uterus measured 1.3cmx2.2cmx3.4cm –appeared enlarged, normal in shape and echo texture. Right ovary measured 1.5 cm x1.5cm and appeared enlarged. 2D Echo done was suggestive of biventricular hypertrophy. As the baby was clinically stable and was tolerating orogastric feeds well, the baby was referredback to Civil Hospital, Sangrurfor establishment of breastfeeding. Father of the baby was counselled regarding the baby's condition and 3-monthly follow up visitshave been planned for the baby. Counselling regarding further genetic testing and risk of tumours was explained to the family in their own language.

Macroglossia

Macrosomia with protuberant abdomen

**Ear Pits**

**Discussion:**

BWSp is a growth regulation disorder presenting with a wide variety of clinical features with an incidence of 1 in 13700 live births (3). An international consensus statement describes the cardinal features and suggestive features. The cardinal features include macroglossia, omphalocele, lateralized overgrowth, multifocal, and/or bilateral Wilms tumour or nephroblastomatosis, and hyperinsulinism. Whereas "suggestive features" include being large for gestational age (birth weight > 2 SDs above mean), facial nevus simplex, polyhydramnios, placentomegaly, ear creases/pits, transient hypoglycaemia, nephromegaly, hepatomegaly, umbilical hernia, diastasis recti, and tumours including neuroblastoma, rhabdomyosarcoma, unilateral Wilms tumour, hepatoblastoma, adrenocortical carcinoma, and pheochromocytoma (2). According to the consensus criteria, this baby has one cardinal feature i.e., macroglossia and 3 suggestive features i.e., macrosomia, ear pits and nephromegaly and hepatomegaly. Based on classification provided by Duffy et al (2019), our baby was classified as atypical BWSp, as our patient had a clinical score of <6 with one cardinal feature.

Transient neonatal hypoglycaemia occurs in 30-60% of patients with BWSp (6). However, in our case hypoglycaemia was not recorded. In BWSp 28-

61% cases have some nephro-urological anomalies and 10% cases have reported cortical and medullary abnormalities (4). In our case ultrasound revealed renal cysts and nephromegaly. Nephromegaly predisposes her to the risk of developing Wilms tumour and hence has to be followed up closely for the same. (5) Generalised visceromegaly is part and parcel of BWSp, however in our case along with nephromegaly and hepatomegaly, uterine hypertrophy and possible ovarian hypertrophy were noted. Congenital heart disease is seen in 13-20% cases of BWSp (6). Our patients Echo done was suggestive of biventricular hypertrophy and cardiomegaly.

There is an 8% risk of developing embryonal tumours in patients with BWSp. The most common embryonal tumours associated it include Wilms tumour, hepatoblastoma and neuroblastoma. International consensus on tumour surveillance for the above is to follow up with 3 monthly abdominal ultrasounds till 7 years of age. (2) As our baby is predisposed to develop Wilmstumour and hepatoblastoma, she would be followed up for the same. An atypical feature of uterine hypertrophy may warrant follow up for development of uterine carcinoma/infertility.

**Conclusion:**

BWSp is a type of foetal overgrowth syndrome with a wide variety of clinical presentation. We have reported a case of atypical presentation of BWSp with no recording of hypoglycaemia, presence of simple hepatic and renal cysts and uterine hypertrophy. Regular follow up is warranted for tumour surveillance.

**References**


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