

Beckwith–Wiedemann syndrome (BWS): A case report and literature review

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Introduction

Beckwith–Wiedemann Syndrome (BWS), also known as the EMG (Exomphalos, Macroglossia, Gigantism) syndrome was recognised independently by Beckwith in 1963 and Wiedemann in 1964 and is now a well established entity having been reported in more than two hundred individuals^{1,2,3}. It constitutes a wide spectrum of clinicopathologic entity with varied combinations of congenital and time dependent abnormalities that often make diagnosis and management tasking^{3,4}. There is paucity of report in the literature on this entity from the developing world. We present a case recently seen at the University College Hospital (UCH) Ibadan, Nigeria in order to create further awareness and highlight peculiarity of management as may be applicable in a setting as ours.

Keywords: *Exomphalous, Macroglossia, Visceromegaly, Conservative management, Close follow-up.*

Case Summary

A 17-hour-old male baby (UCH # 966465) was referred from a private hospital to the Children Emergency Ward of the UCH Ibadan with an umbilical defect and a large protruding tongue both noticed at birth. He was delivered at home by spontaneous vaginal delivery to a 29-year-old multiparous petty trader whose other children had no history of congenital abnormality. The pregnancy, labour and delivery were uneventful. Physical examination revealed a plenthoric male infant with a large protruding tongue. His birth weight was 4.05kg. The

occipito-frontal circumference was 34cm with an occipital prominence. He had low set ears with bilateral clefts of the pinnae. There was a 6cm diameter defect centrally in the abdomen, which was covered with an intact membrane, with the umbilical cord arising from its apex and loops of intestine were visible under the membrane. There was an undescended left testicle palpable in the left groin. There was subtle left sided hemihypertrophy with a mid-arm circumference of 10cm on the right and 11cm on the left while the mid-thigh circumference measured 17cm on the right and 18cm on the left.

A diagnosis of Beckwith–Wiedemann syndrome was made. A nasogastric tube was passed for continuous gastric decompression. He was commenced on intravenous fluid containing dextrose, saline and maintenance electrolytes. He was also commenced on prophylaxis parenteral cefuroxime and metronidazole and was given anti-tetanus prophylaxis. The sac was painted with gentian violet and a full leg plaster of paris (POP) cast with a bridge was applied to immobilise the lower limbs in order to prevent rupture of the sac inadvertently by the baby's legs. The packed cell volume estimation was 71%, serum electrolytes and urea estimation was normal but glucose-6-phosphate dehydrogenase estimation was deficient. The random blood sugar estimation was 37mg/dL. Hypoglycaemia was corrected with intravenous 50% dextrose solution. The serial blood glucose estimation was normal thereafter. An ultrasound scan of the abdomen showed no gross abnormality. The omphalocoele healed with good epithelialisation thus converting it to an umbilical hernia after 4 weeks of non-operative treatment. He was discharged home and presently being followed up at the surgical and children's outpatient clinics regularly.

Discussion

The incidence of BWS has been reported as 1 in 13,700 live births⁴ and most cases are sporadic. The aetiology is unknown but familial cases suggestive of autosomal dominance, autosomal recessive, polygenic inheritance and delayed mutation have all been recorded¹. Elevated concentrations of the plasma growth hormone and somatomedin have been detected at birth, however, suggesting an abnormality in the production of foetal growth regulating peptides^{1,5}. A prenatal diagnosis of BWS is possible in pregnancy with uterine sizes incompatible with dates and by the use of serial ultrasound monitoring of the uterus of the pregnant woman.

Beck–Wiedeman Syndrome is now a clearly delineated clinical entity that exhibits a myriad of features including exomphalos, macroglossia, gigantism (visceral or somatic), neonatal hypoglycaemia, cryptorchidism, ear anomalies, craniofacial dysmorphism etc. These features manifest in varying non-specific combinations and even the absence of one or two major manifestations does not invalidate the diagnosis of BWS. The hitherto diagnostic difficulty is easily overcome if one has a high index of suspicion. The immediate challenge therefore is in the management of the omphalocoele and the macroglossia that may



Fig. 1 Clinical photograph of the patient with BWS

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pose serious threat to the survival of the baby. The omphalocele can rupture, bleed, get obstructed if containing bowel loops or become a potential source of sepsis. An enlarged tongue may interfere with the neonate's respiration and feeding, and also with speech in infancy and early childhood.

Early surgical closure of the umbilical defects (small and medium sized) is usually advocated. This is achieved by a single procedure of excising the sac and closing the defect primarily or through a staged procedure of covering a large defect with a mobilised skin flap with or without a silastic silo thus converting the defect into a ventral hernia that is due for repair later. The reduction of the contents of a large omphalocele into a small abdominal cavity by early surgery causes respiratory and circulatory embarrassment that necessitates perioperative neonatal intensive care unit admission. In the developed world where specialised neonatal surgical intensive therapy units are available, early surgical management of omphalocele major is the rule. This is hardly the case in most developing countries, where such specialised facilities may be lacking or ill-equipped. Instead of early surgical reduction, conservative measures that are aimed at causing epithelialisation of the sac and thereby converting it to a ventral hernia are adopted.

As illustrated in our case, daily topical application of gentian violet on the sac hardens the sac and causes it to granulate and gradually epithelialise within 4 weeks. Honey is a good alternative. It is cheap and locally available but it causes pain in wounds. The use of mafenide, 2% aqueous mercurochrome and povidone-iodine (betadine) is discouraged because of their systemic effects following absorption by the sac. One major disadvantage of this initial conservative approach, though rare, is inadvertent rupture of the sac, which then requires surgical intervention. To prevent this, the baby's legs are put in backslab P.O.P with a crossbar in between to prevent rubbing the knees and the legs against the soft sac. Other drawbacks of conservative management include inability to explore the abdomen and detect other possible congenital intra-abdominal abnormalities, prolonged hospital admission of the baby, which consequently tells on the family's schedule and purse, and potential toxic effects of the escharotics.

Macroglossia, like many of the other physical abnormalities of BWS tends to regress with increasing age. However indications for partial glossectomy include recurrent airway obstruction, poor speech development, cosmesis and prevention of development of mandibular prognathism. Maldescended testes should be fixed as soon as possible to reduce the possible complications of subfertility, trauma, torsion and malignant transformation^{7,8,9}. When these patients need operative surgery, careful preoperative evaluation, perioperative monitoring and suitable choice of anaesthetic technique are required for a successful outcome⁹. The oncogenic potentials in patients with hemihypertrophy in BWS are high especially if these patients have

visceromegaly at birth^{3,4,6}. The most common malignancy is Wilm's tumour with an estimated risk of 7.5% increasing to 10% if hemihypertrophy is present. Other associated tumours are hepatoblastoma, neuroblastoma, adrenocortical carcinoma and soft tissue sarcoma which can arise in the limbs, tongue or urinary bladder¹⁴.

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