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Case Report

Beckwith-Wiedemann Syndrome-An Uncommon Presentation in Two Consecutive Siblings in UCH Ibadan

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Abstract

BWS is a congenital overgrowth disorder. It is the most common of the overgrowth syndromes, occurring in 13,700 live births. Basic features are exomphalos, macroglossia, and gigantism. It is diagnosed using clinical features and genetic testing; in developing countries especially Nigeria, genetic testing is not within reach of the average citizen. We present two siblings who were born pre-term and had clinical features suggestive of Beckwith Wiedemann syndrome but died within two weeks of life from complications of prematurity.

Keywords: *Beckwith-Wiedemann Syndrome; Genetic Testing; Challenges*

Abbreviation

BWS- Beckwith Wiedemann Syndrome

Introduction

Beckwith-Wiedemann Syndrome (BWS) is a congenital overgrowth disorder. It is the most common of the overgrowth syndromes. Basic features are exomphalos, macroglossia and gigantism. Most cases are diagnosed after birth by clinical features and genetic testing. We report the cases of two consecutive siblings who were born with Beckwith-Wiedemann syndrome.

Case Report

The two siblings were born preterm (32 weeks gestation) at 1-year interval. There was a positive history of recurrent spontaneous abortions in the mother. She had routine antenatal drugs, no history of exposure to irradiation. In the second pregnancy, the ultrasound scan showed bilateral hydronephrosis with exomphalos; for which she was referred to the paediatric surgeon to schedule delivery, but the mother refused. There was no history of consanguinity; their father is a farmer who has a positive history of exposure to herbicides.

Clinical findings

Cardinal Features ++	Suggestive Features +	Sibling 1	Sibling 2
Macroglossia	Polyhydramnios	++	++
Exomphalos	Ear creases and/or pits	++	++,+
Lateralized overgrowth	Birthweight > 2SD	++,+	++
Hyperinsulinism-Hypoglycaemia > 1 week	Transient hypoglycaemia < 1 week	------	++
Renal Abnormalities	Umbilical Hernia	+	++,+
Visceromegaly	Prematurity	+	++,+
+ Family History	Advanced Bone Age	--------	++

Table 1

Cardinal features- 2 points per feature, Suggestive features- 1 point per feature.

Management was multidisciplinary with the involvement of neonatologists, paediatric endocrinologists and paediatric surgeons. They both had supportive management throughout admission, which include- phototherapy for the management of jaundice, Intravenous antibiotics for the treatment of infection, correction of recurrent hypoglycaemia with intravenous dextrose water and intravenous hydrocortisone. They both had dressing of the Omphalocele while waiting for primary closure of the anterior abdominal wall defect. They both died within two weeks of delivery from complications of prematurity.

Investigations

Full Blood Count and Differential	Normal	Normal
Electrolyte and Urea	Hypocalcaemia	Hypocalcaemia
Random Plasma Glucose	Normoglycaemia	Recurrent Hypoglycaemia
Thyroid Function Test	Normal	Hypothyroidism
Serum Bilirubin	Hyperbilirubinaemia	Hyperbilirubinaemia
Serum Protein	----------	Low
G6PD Status	Normal	Normal

Table 2

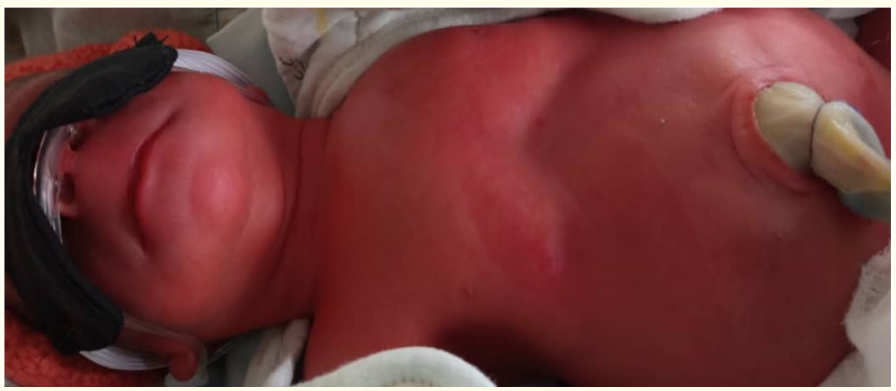

Figure 1

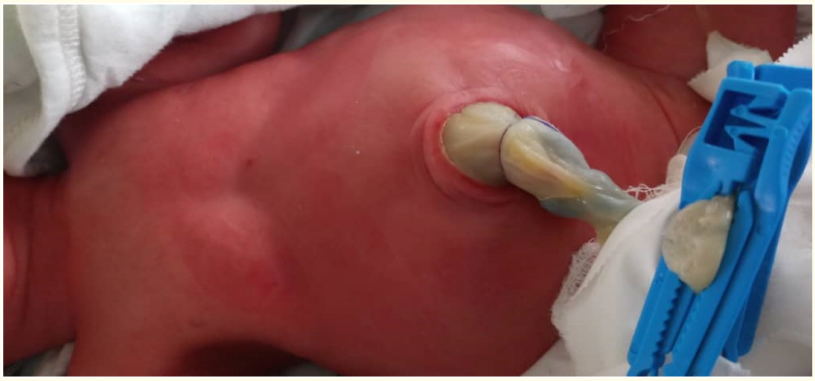

Figure 2

Discussion

Beckwith Wiedemann syndrome is the commonest overgrowth syndrome with an incidence of 1 in 13,700 live births [1]. Beckwith, an American paediatric pathologist, and Wiedemann, a German geneticist first described the anomaly in the mid-60s [2]. It is the most commonly seen of the documented overgrowth syndromes worldwide [3]. It has no racial or gender predilection [4]. The siblings in our report were of different gender. It occurs, due to deregulation of imprinted gene expression in the chromosome 11p15.5 region [5]. About 85% of documented cases are sporadic while the remaining 15% are familial [6]. Basic features as described by Beckwith and Wiedemann include exomphalos, macroglossia, and gigantism. Other findings include posterior auricular pits and organ overgrowth, commonly hepatomegaly and renomegaly [4]. In our report, most of the findings were present in both babies, which made them meet the clinical criteria for diagnosis of BWS. Berland., *et al.* discovered a familial presentation between 2 sisters and their male cousin [7]. Our report was on two consecutive siblings and there had been repeated spontaneous abortions in their mother. The familial nature possibly suggests the heterozygous maternally inherited variant that has high penetrance; seen in about 40% of familial cases and about 10% of cases without family history. While survival of > 80% has become the usual experience since increased awareness of the condition in the last few decades, early death as seen in these cases have been documented in literatures to occur from complications of prematurity, airway obstruction from macroglossia, hypoglycemia, cardiomyopathy, or tumors [8]. Both of the cases reported died of complications of prematurity within two weeks of life. Molecular confirmation of this syndrome is possible in about 90% of the cases [9]. This is however still very expensive in developing countries. Genetic counselling for parents is essential however will not be specific and will be unable to estimate recurrence risks since the underlying genetic mechanism has not been determined.

Conclusion

The cases presented highlights the difficulties in the management of patients with Beckwith Wiedemann syndrome. Prenatal testing and close surveillance of subsequent pregnancies is necessary to aid appropriate decision-making and optimise chances of survival of the newborn.

Conflicts of Interest

There is no conflict of interest.

Funding

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Ethical Approval

No approval was required.

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