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Beckwith-Wiedemann Syndrome: A Case Report

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ABSTRACT

Background & aim: Beckwith Wiedemann Syndrome (BWS) is an infrequent inborn malformation that presents with exomphalos, macroglossia and gigantism. In addition, some children with BWS have other features including nevus flammeus, prominent occiput, midface hypoplasia, hemihypertrophy, genitourinary anomalies, heart anomalies, musculoskeletal aberrations, and auditory loss. In this case report a newborn with BWS presenting with macroglossia is reported.

Case report: A 26 years old gravida 3 para 2 married women admitted at hospital when she was in labour at 38 weeks' gestational age (GA). During admission all vital signs were normal as well as fetal status were noted to be normal. She attended three ANC visits, no any abnormality that was detected during prenatal. No history of genetic abnormality in her family, she was treated malaria during pregnancy at 19 weeks GA. In previous obstetrics history she got PPH during her previous pregnancy in the last two years.

The women had normal labour. She had spontaneous vaginal delivery of a female baby with 3.3 kg with Apgar score of 7 in 1 minute and 9 in 5 minutes. On postnatal examination the baby was detected to have an abnormal big tongue a case which is known as BWS. The case management was too complicated following its rarity and later the baby died due to complications of hypoglycemia.

Conclusion: Revelation of the etiological mechanisms and use of a laboratory procedure to detect alterations in these disorders may be useful for management of these rare malformations and genetic counseling of the families.

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Introduction

The Beckwith-Wiedemann syndrome (BWS) is a human imprinting disorder that leads to overgrowth and a predisposition to cancer (1). More or less some cases may lack the hallmark features of exomphalos, macroglossia, and gigantism as originally described by Beckwith and Wiedemann (2). This figure is likely an under estimate, as minor phenotypes may not be made certain (3). Male to female ratio is equal (4) fetuses with distinguished exemption of monozygotic twins that show a dramatic female fetuses predominance due to delayed development of female compared to male embryos (5).

The precise frequency of occurrence of BWS in Tanzania is unknown due to inconsistency in the syndrome presentation, difficulties with diagnosis and cases under reporting. Statistics of documented newborns with BWS is low most likely because many newborns may be being born with clinical features that are not much prominent and therefore are underlooked.

About 80% of patients with BWS have a known molecular defect in the 11p15 region, which includes imprinted genes that regulate fetal and postnatal growth, most commonly due to aberrant DNA methylation (6). Normally, the paternal allele is methylated at IC1 and the maternal allele is methylated at IC2 (3).

BWS is always diagnosed during neonatal period or in early infancy stage and has clinical features that vary in severity (6). The clinical presentation is highly variable and could include macroglossia, lateralized overgrowth, abdominal wall defects, enlarged abdominal structures, and an elevated danger for

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embryonal tumors developing during early childhood (7).

BWS is characterized by an enlarged and thick tongue that may have fissures and ulcers, may cause language alterations, difficulties for feeding and swallowing, sialorrhea and recurrent infections of the upper airway or even its obstruction (8). There are no absolute requisites for the clinical diagnosis of BWS since diagnosing rare pediatric syndromes remains challenging (8). Often the diagnosis may be done by a pediatrician in collaboration with geneticist (9).

Its clinical evaluation must include a complete clinical chart with careful physical exploration and a pedigree of that may identify the presence or absence of a hereditary associated syndrome. This case is unique due to its rarity and complexity in management.

Case report

A 26 years old woman, Gravida 3 Para 2 with one living child presented at our hospital being in labour and was admitted at 16:00 hours.

Antenatal history: She attended antenatal clinic three times. First booking was at 13 weeks of gestation. She received three doses of Intermittent Preventive Treatment of malaria during Pregnancy (IPTp).

Previous obstetric history: In the past pregnancies she had postpartum haemorrhage (PPH) due to retained placenta. There was no history of assisted reproduction.

Figure 1. Clinical picture of a baby with Beckwith–Wiedemann Syndrome

Contraceptives history: This woman has a history of using Depo Provera as a method of family planning.

Medical history: The woman had history of being treated for malaria with Alu during the second trimester when she was at 19 weeks gestational age and was fully recovered.

Family and genetic history: The woman was married living with her husband and there was no family history of any congenital malformation history among the relatives of neither her nor her husband.

Physical assessment during antenatal care was done, which revealed that she was overweight. Other variables were normal.

Obstetrics examination was done and the following were the findings: single tone baby at gestational age of 38 weeks, cephalic presentation, FHR=140 bpm, and moderate contractions. During per vaginal examination the cervix was thin effaced, and dilated 6cm.

At 21:00 hours the mother gave birth through spontaneous vaginal delivery and a female newborn of 3.3kg with Apgar score of 7 in the 1st minute and 9 in the 5th minute was born. The vital signs of the baby were stable as heart rate was 122, respiratory rate was 25 bpm, and temperature was 36.6°C. The blood pressure was not measured.

On postnatal physical examination the baby had an abnormal big tongue. The malformation was noted and the mother was counseled. Nasogastric tube was inserted to the newborn. Other systems were normal.

On day 2 the baby glucose was 2 mmol/L. She was given 2ml/Kg intravenous bolus of 10% dextrose after 6 hours the baby was given glucagon bolus 0.02 mg/kg., Dexamethasone 0.25 m/kg and Hydrocortisone.

On the same day during physical assessment the extensive physical assessment was done where by: in neurological the baby had altered conscious state and seizures; in respiratory assessment she had hypoventilation, apnea and cyanosis; in cardiovascular examination she had tachycardia (190 bpm) and in gastrointestinal examination she had poor feeding. Ultimately, the newborn's conditions continued to deteriorate, unfortunately the newborn died due to the conditions related to hypoglycemia.

Discussion

According to Hassan, more than ninety percent of a hyperglossia is very common and is a marked feature of BWS (1). Newborns who has BWS and hyperglossia classically cannot close their mouth fully in front of their huge tongue, causing the tongue to bulge out. This feature is similar to the current case report. The cause of enlargement of the tongue is due to hyperplasia of muscle fibres, making the tongue to increases in all three dimensions, but individual variation in the presentation of the tongue have been noted.

Currently, there are no specific techniques used regularly in the clinical diagnosis of macroglossia, since the diagnosis is based on subjective criteria like the extent of tongue protrusion, and the clinical signs and symptoms that occur secondary to the macroglossia. According to (1) large tongue in BWS becomes less obvious with age and may entail no management, nevertheless in some children the situation may be severe leading to problems like difficulty in respiration, feeding, and speech difficulties (9). According to (10), clinical specialties should collaborate in giving services to the affected child, the specialists should include speech and language therapist, surgeon, orthodontist, and clinical nurse specialist.

Several studies have shown that, the appropriate period for performing surgery in this condition is not well established, but specialists endorse operation between three and six months of age involving removal of apportion of the tongue so as to be accommodated within the mouth to permit proper development of jaw and tooth (11). Prognosis differs depending on the clinical presentation (1). In the current case report the newborn died due to hypoglycaemia, thus it is similar to the findings from a study by Weksberg and colleagues who stated that hypoglycemia has been documented in 30–50% of newborns with BWS, and this is likely caused by hyperplasia of islet cell and hyperinsulinemia (11).

This case report might sensitize readers and thus facilitate detection of similar or identical cases. In addition, it will add new information to scholars, generate hypotheses and accumulate scientific data about this kind of rare disorders and serve as education case.

The authors are underway in developing Genetic and Rare Diseases (GARD) Information Center and a registry is being established to coordinate research efforts into rare congenital malformations in Tanzania.

Diagnosing rare pediatric syndromes remains challenging because of limited resources and expertise. The medical records did not contain all relevant information about the case. Due to the rarity of the case it was difficult to find good search terms for our objectives as such we have not been able to review all relevant literatures.

Conclusion

In this case report, the prognosis was poor, the newborn died of hypoglycemia. For this reason, calls for follow-up and management of the patients with BWS should be done in an early and individualized way in order to reduce the complications and mortalities that may arise and provide available treatment. As the etiopathogenic causes vary, therefore it is necessary for appreciate for guiding interdisciplinary medical surveillance protocols, that is individualized to each patient and include timely family genetic counseling since BWS molecular subgroups are associated with dissimilar risks of recurrence.

Patients and their families should be offered counseling, education, support, and guidance since the moment of diagnosis. As part of the holistic approach to the management of patients diagnosed with BWS, genetic counseling which involves providing individuals and families with information on the nature, mode of inheritance, and implications of genetic disorders help the

family members to have autonomy and make informed medical and personal decisions.

Special ethical issues related to the confidentiality and privacy protection should be adhered to since the information about the individual, family history, carrier status, risk of genetic disease to self or offspring can be stigmatizing therefore confidentiality is paramount. Much sensitivity to the meaning of the communication and commitment to clarity and consistency should be considered.

This syndrome is rare and many few cases have been reported in the world English literatures, so no much information is available.

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Conflicts of Interest

The authors declare no conflicts of interest.

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