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Case Report

Beckwith Wiedemann Syndrome: A Case Report

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Abstract:

The baby with Beckwith Wiedemann Syndrome (BWS) usualy presents with exomphalos, macroglossia and gigantism. In addition, some children with BWS have other findings including: nevus flammeus, prominent occiput, midface hypoplasia, hemihypertrophy, genitourinary anomalies (enlarged kidneys), cardiac anomalies, musculoskeletal abnormalities, and hearing loss. Incidence of one in 13,700 live birth. As children with BWS are at increased risk of childhood cancer, they should follow up strictly for cancer screening. We are reporting this type of a case who has got Exomphalos, Macroglossia, Gigantism and Hemihypertrophy. We have corrected the exomphalos and advised him for follow up for cancer screening.

Key words: Beckwith Wiedemann Syndrome.

Introduction:

Beckwith Wiedemann Syndrome (BWS) has an estimated incidence of one in 13,700; about 300 children with BWS are born each year in the United States[1]. The exact incidence of BWS is unknown in our country because of the marked variability in the syndrome's presentation and difficulties with diagnosis. The number of reported infants born with BWS is most likely low because many are born with BWS, but have clinical features that are less prominent and therefore missed. BWS has been documented in a variety of ethnic groups and occurs equally in males and females.

Children conceived through In vitro fertilization have a three to four fold increased chance of developing BWS. It is thought that this is due to genes being turned on or off by the IVF procedures[2,3].

The baby with BWS usualy presents with the combination of congenital abdominal wall defects as hernia (exomphalos), large tongues (macroglossia), and large bodies and/or long limbs (gigantism). In addition, some children with BWS have other findings including: nevus flammeus, prominent occiput, midface hypoplasia, hemihypertrophy, genitourinary anomalies (enlarged kidneys), cardiac anomalies, musculoskeletal abnormalities, and hearing loss. Also, some premature newborns with BWS do not have macroglossia until closer to their anticipated delivery date[4].

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Another definition presented by Elliot et al. includes the presence of either three major features (anterior abdominal wall defect, macroglossia, or prepostnatal overgrowth) or two major plus three minor findings (ear pits, nevus flammeus, neonatal hypoglycemia, nephromegaly, or hemihyperplasia)[5].

While most children with BWS do not develop cancer, children with BWS do have a significantly increased risk of cancer. Children with BWS are most at risk during early childhood and should receive cancer screening during this time[6].

Case Report:

A 9-month old female patient weighing 15 kg was admitted in Pediatric Surgery ward of Faridpur Medical College with the complaints of a swelling over the umbilical region since birth measuring about 6x5 cm which is reducible in lying condition. And the swelling is increased on crying. A large protruded tongue since birth. For these reasons breast feeding nor bottle feeding was possible. Right upper limb and lower limbs are larger than left one both in length & mid thigh circumference (Fig.1).

Examination revealed that there was ventral hernia at the umbilical region. Abdomen is soft and non tender. There is a soft swelling over the umbilical region which is reducible. On musculoskeletal examination her right upper limb and lower limbs are larger than those of left. Other systemic examination reveals no apparent abnormality.

Discussion:

Abdominal wall defects are common in newborns with BWS and may require surgical treatment. Newborns with an omphalocele typically require surgery to place the abdominal contents back into the abdomen in order to prevent serious infection or shock.

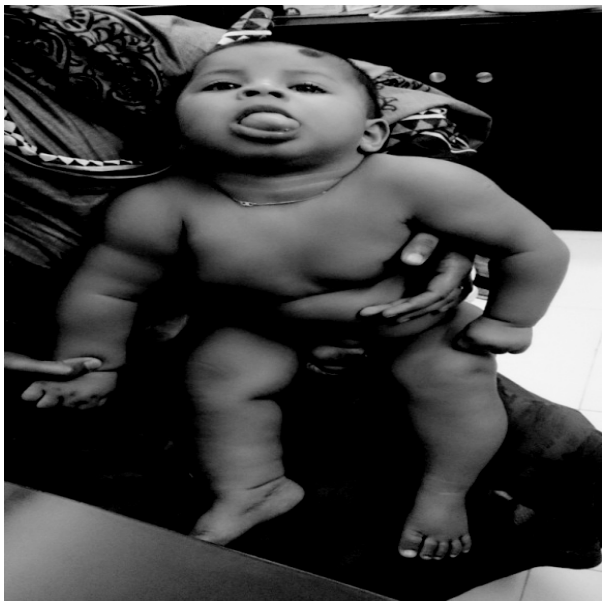

Fig-1: Exomphalos, Macroglossia and Hemihypertrophy are present in this patient

Neonatal hypoglycemia, low blood glucose in the first month of life, occurs in about half of children with BWS.[7] Most of these hypoglycemic newborns are asymptomatic and have a normal blood glucose level within days. However, untreated persistent hypoglycemia can lead to permanent brain damage. Hypoglycemia in newborns with BWS should be managed according to standard protocols for treating neonatal hypoglycemia. Usually this hypoglycemia can easily be treated with more frequent feedings or medical doses of glucose. Macroglossia, a large tongue, is a very common (>90%) and prominent feature of BWS[8]. Infants with BWS and macroglossia typically cannot fully close their mouth in front of their large tongue, causing it to protrude out. Macroglossia in BWS becomes less noticeable with age and often requires no treatment; but it does cause problems for some children with BWS. In severe cases, macroglossia can cause respiratory, feeding, and speech difficulties. The best time to perform surgery for a large tongue is not known. Some surgeons recommend performing the surgery between 3 and 6 months of age. Surgery for macroglossia involves removing a small part of the tongue so that it fits within the mouth to allow for proper jaw and tooth development[5,9].

Hemihypertrophy (hemihyperplasia) is an abnormal asymmetry between the left and right sides of the body occurring when one part of the body grows faster than normal. Isolated hemihypertrophy is associated with a higher risk for cancer[6,7]. As a result, children with hemihypertrophy should follow the general cancer screening protocol for BWS. Hemihypertrophy can also cause various orthopedic problems, so children with significant limb hemihyperplasia should be evaluated and followed by an orthopedic surgeon.

Most children (>80%) with BWS do not develop cancer; however, children with BWS are much more likely (600 times more) than other children to develop certain childhood cancers, particularly Wilms' tumor, pancreatoblastoma and hepatoblastoma, neuroblastoma, and rhabdomyosarcoma. Early diagnosis allows physicians to treat the cancer when it is at an early stage. In addition, there is less toxic treatment[8,9]. Given the importance of early diagnosis, all children with BWS should receive cancer screening.

Conclusion:

Children with BWS usually do very well and grow up to become the heights expected based on their parents' heights. As children with BWS are at increased risk of childhood cancer, they should follow up strictly for cancer screening. An abdominal ultrasound every 3 months 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. Families and physicians should determine screening schedules for specific patients, especially the age at which to discontinue screening, based upon their own evaluation of the risk-benefit ratio.

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