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A Rare Case of Beckwith Wiedemann Syndrome

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Abstract

Beckwith Wiedemann syndrome is rare congenital overgrowth syndrome. This syndrome usually presents in neonates with macroglossia, large birth weight, omphalocele, visceromegaly and hypoglycemia. However, there is little information available regarding the natural history in adults with BWS. Here we present a case of Beckwith-Wiedemann syndrome with its major manifestations but without hypoglycemia that was misdiagnosed in infancy with mucopolysaccharidosis and presented to us at 14 years of age with tall stature.

Keywords: Beckwith Wiedemann syndrome • Macroglossia • Omphalocele • Visceromegaly

Introduction

Beckwith Wiedemann Syndrome (BWS) was described independently by two investigators. In 1963, Beckwith presented 3 postmortem cases with macroglossia, omphalocele, cytomegaly of the fetal adrenal cortex, renal medullar dysplasia and visceromegaly. On the other hand, Wiedemann in 1964 reported 3 cases of siblings with similar clinical characteristics, adding diaphragm defects and hypoglycemia. It is a disease of low prevalence; however it represents the most common among the genetic overgrowth syndromes, with prevalence of 1:13,700 births [1]. Babies with BWS are mostly macrosomic at birth and either symmetric or asymmetric overgrowth continues throughout childhood. Characteristic features of BWS which help to clinch diagnosis include eye proptosis with periorbital fullness, capillary malformation (nevus flammeus), earlobe creases and pits, large mouth with large tongue (macroglossia), organomegaly and omphalocele [2].

Syndrome was initially described by Beckwith and Wiedemann in 19693 with three cardinal features; Exomphalos (omphalocele), Macroglossia and Gigantism (EMG). BWS is quite variable and EMG is seen in some but not all of the children. BWS experts including Dr. J Bruce Beckwith and Dr. Rossana Wecksberg published criteria in Gene Reviews in 20104.

Frequently, macroglossia is the sign that prompts the diagnosis in the newborn. However, there is little information available regarding the natural history in adults with BWS.

Case Presentation

14 year old boy, product of non-consanguineous marriage, first in birth order presented with concern of tall stature. Historically mother had polyhydramnios antenatally with need of therapeutic drainage of amniotic fluid once. Birth weight was 3.75 kgs (+1.1 SDS). There was history of anterior abdominal wall defect presented as umbilical hernia, which was repaired in first few months of life [3]. There was history of delayed speech. He also had bilateral cryptorchidism for which orcheidopaxy was done on right side and orcheidectomy was done on left side at age of seven years. Other siblings were normal with no significant family history. Examination revealed tall stature [Ht-183 cm (>97th percentile), SDS of +3.44 with height age of 20 years, arm span 182 cm, Upper Segment and Lower Segment ratio (US/LS) of 0.9 and weight was 44 kg (50th percentile). Systemic examination revealed macroglossia, low posterior hair line and infra orbital groove (Figure 1). In addition he had characteristic ear findings including horizontal ear crease and characteristic pits on pinnae of ears (Figure 2). Linear verrucous epidermal naevus, presenting as brownish wart like papules, was seen on the neck (Figure 3). Rest of the systemic examination was normal. Sexual maturity rate revealed; Stretched Penile Length (SPL)-10 cms, testicular volume 20 ml on right side, mustache line present, AH+, PH4. Investigations revealed; bone age 14 years, normal growth hormone suppression test, normal slit lamp examination of eyes and hearing evaluation. Hormonal profile revealed LH-2.15 IU/L, FSH-4.05 IU/L, serum testosterone 186.38 ng/dl, serum prolactin-6.61 IU/L, T3-0.89 ng/dl, T4-7.74 µg/ml, TSH-3.68 µIU/ml, 25(OH)D 17.83 ng/ml. USG abdomen and 2D-ECHO was normal [4].

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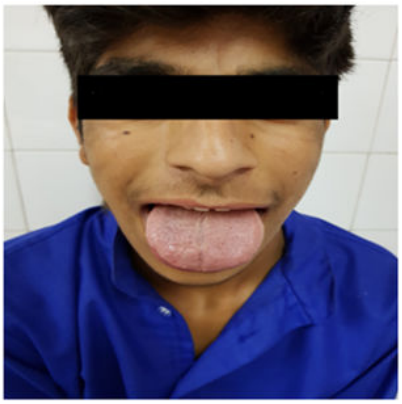

Figure 1. Showing macroglossia.

Figure 2. Showing characteristic ear pits.

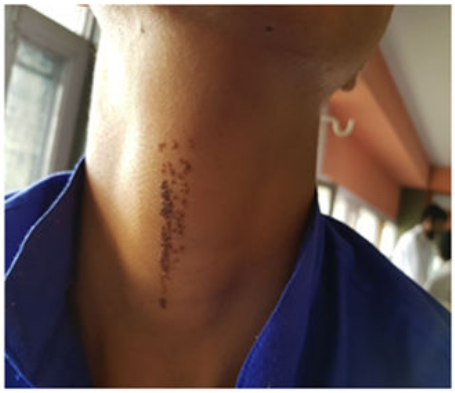

Figure 3. Showing linear epidermal naevus.

Discussion

Beckwith Wiedemann Syndrome (BWS) represents a genetic syndrome of low prevalence and diverse clinical expression. However, it is one of the most common overgrowth syndromes and presents macroglossia in almost all its forms. Facing a macroglossia of unknown cause, the clinician can suspect a BWS. The diagnosis of macroglossia is based on the morphology and protrusion; situation that carries functional, growth, psychological or feeding problems. There are a few medical concerns for adults with BWS, *i.e.,* renal medullary dysplasia and decreased fertility in males. These are rare complications and there is an association with a specific molecular abnormality. Suggested referrals include counseling for possible male infertility, offering referral for infertility assessment and semen evaluation as appropriate, echocardiography every 3 years to 5 years, renal ultra-sound once early in adulthood, renal function testing every 3 years to 5 years and evaluation of hearing every 2 years to 3 years [5,6].

Conclusion

These recommendations are based on a limited number of cases. Embryonal tumours occur in ~8% of children with BWSp149. The most common types of embryonal tumours are Wilms tumour (52% of all tumours), hepatoblastoma (14% of all tumours), neuroblastoma (10% of all tumours), rhabdomyosarcoma (5% of all tumours) and adrenal carcinoma (3% of all tumours). Although there are some differences in mean age at diagnosis between tumour types, the overall cancer risk is highest in the first 2 years of life and clinical experience suggests that the cancer risk then declines progressively before puberty, approaching the cancer risk of the general population. Currently, there is no evidence of an increased risk of malignant tumours in adulthood.

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