BILATERAL BENIGN HAEMORRHAGIC ADRENAL CYSTS IN BECKWITH-WIEDEMANN SYNDROME: A CASE REPORT

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Submission date: 21-Feb-2025 12:29PM (UTC+0700)
Submission ID: 2576797551
File name: IJAR-50347.docx (994.24K)
Word count: 1710
Character count: 10619

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ABSTRACT:

Beckwith-Wiedemann syndrome (BWS) is a genetic proliferative disorder, often associated with hyperplasia of various organs, including the adrenal glands.(1)

Benign hemorrhagic adrenal cysts are a relatively common form of adrenal mass observed in patients with this syndrome, although they are typically asymptomatic. However, their presence requires careful monitoring due to the potential risks of complications and progression.(2)

We report a case of bilateral adrenal cystic masses detected during a routine third-trimester prenatal ultrasound, associated with macrosomia and macroglossia at birth.

The diagnosis of Beckwith-Wiedemann syndrome was suspected in our patient. Prenatal and postnatal ultrasounds, magnetic resonance imaging (MRI), and biological assessments failed to establish the origin of these adrenal masses.

Differential diagnoses considered included bilateral cystic neuroblastoma, bilateral cystic lymphangioma, bilateral cystic adrenal cortical adenoma, and duplication of the renal-ureteral system.

A laparotomy was performed two months after birth due to the large size of the tumor and suspicion of its tumorigenic origin. Histopathological examination revealed a cystic adrenal hematoma, with no signs of malignancy.

The diagnosis of bilateral benign hemorrhagic macrocystic adrenal component associated with Beckwith-Wiedemann syndrome was confirmed.

A multidisciplinary approach, including close radiological and biological monitoring, was implemented.

INTRODUCTION:

Beckwith-Wiedemann syndrome (BWS) is a rare congenital disorder with low prevalence, estimated to occur in 1 out of 13,500 live births. Despite its rarity, it is one of the most common and well-documented congenital somatic overgrowth syndromes.(3)

This genetic syndrome results from alterations in gene expression located on the p15.5 region of chromosome 11. The genes primarily involved are IGF2, a fetal growth factor, and H19, a tumor suppressor gene. These alterations usually occur sporadically in 85% of cases, hereditary in 15%, or due to chromosomal abnormalities in about 1% of cases.(4)

The clinical expression of Beckwith-Wiedemann syndrome varies considerably from one patient to another. Diagnosis is based on the presence of at least two major criteria (such as macrosomia, macroglossia, and omphalocele), as well as one minor criterion, which may include neonatal hypoglycemia, hemihyperplasia, umbilical hernia, embryonal tumors, anterior ear creases, posterior helical fistulas, port-wine stains (or other vascular malformations), renal anomalies, abdominal visceromegaly, fetal adrenal cytomegaly (pathognomonic), cardiac malformations, or cleft palate.(5)
Positive molecular analysis confirms the diagnosis, but its absence does not exclude it.(4)
Beckwith-Wiedemann syndrome is a genetic proliferative disorder associated with growth anomalies and an increased predisposition to embryonal tumors. (6)It also causes hyperplasia of various organs, including the adrenal glands. Benign hemorrhagic adrenal cysts are a frequent complication in BWS patients, although they are usually asymptomatic and may resolve spontaneously. However, the large size and bilaterality of these lesions warrant exclusion of malignant tumors before regular clinical and radiological monitoring to prevent potential complications or abnormal progression. Imaging plays a crucial role in early detection and follow-up of these cysts, ensuring optimal management of the syndrome. (7)

CASE REPORT:

We report the case of a male newborn admitted at 10 days of age for evaluation of an abdominal mass detected during a third-trimester prenatal ultrasound (figure1), along with macrosomia and macroglossia at birth.
The newborn was born to a non-consanguineous couple, with a 26-year-old mother who had a gravidity of 2 and parity of 2. She was not diabetic and had a normal body mass index. Our patient has a healthy older brother. The birth was at term, vaginal, after an uncomplicated pregnancy. The baby adapted well to extrauterine life, with an APGAR score of 7 at 5 minutes and 10 at 10 minutes. At birth, his weight was 5600 g, and his length was 58 cm, indicating macrosomia (+2 SD, 98th percentile). His head circumference was 48 cm. (figure 3)
Clinical examination revealed staturo-ponderal advancement, macroglossia with tongue protrusion, and a reducible umbilical hernia. A diastasis of the rectus muscles and auricular anomalies (sulci in the ear lobes) were also noted, along with "ear creases." Glycemic monitoring was normal. (figure 2)
Skin examination revealed a frontal port-wine stain. Abdominal examination showed a very distended abdomen with palpation revealing a large, bilateral, firm, non-tender mass, as well as bilateral lumbar contact.
Blood pressure was normal. Biological tests, including blood and urine analysis, were within normal limits, as were blood glucose, catecholamine metabolites (VMA, HVA), alpha-fetoprotein (AFP), and β-hCG (human chorionic gonadotropin) levels. Corticoadrenal hormone levels were also normal, with no signs of adrenal insufficiency.

Due to the clinical signs, the diagnosis of Beckwith-Wiedemann syndrome (BWS) was suspected, and molecular analysis was requested for confirmation.
Three differential diagnoses were discussed: neuroblastoma, cystic lymphangioma, and cystic adrenal cortical adenoma.
Abdominal ultrasound revealed two large, solid-cystic bilateral masses in the adrenal regions, closely associated with the kidneys. The masses measured 94 x 58.5 mm on the right and 66 x 49 mm on the left, suggestive of bilateral neuroblastoma.
Magnetic resonance imaging (MRI) showed well-demarcated, lobulated bilateral lesions with hypodense cystic areas, thin walls, and homogeneous tissue parts after contrast injection. The lesions measured 67 x 64 x 90 mm on the right and 62 x 63 x 90 mm on the left, suggesting bilateral cystic neuroblastomas.
A few days later, cytogenetic study results were positive, confirming the presence of Beckwith-Wiedemann syndrome.
In the context of Beckwith-Wiedemann syndrome (BWS), three possible diagnoses were considered: bilateral cystic neuroblastoma, bilateral cystic lymphangioma, or bilateral cystic adrenal cortical adenoma.

Three weeks after birth, a laparotomy was performed due to the large size of the masses and suspicion of their tumor origin.
Complete excision of the masses was successfully carried out, and histopathological examination confirmed the presence of a benign hemorrhagic cyst with no signs of malignancy. Postoperative follow-up was uneventful.
The final diagnosis was that of a benign bilateral hemorrhagic macrocystic component of the adrenal glands, associated with Beckwith-Wiedemann syndrome.
The patient was discharged without specific treatment and is currently being monitored by a multidisciplinary team for embryonal tumor screening, including alpha-fetoprotein (AFP) marker measurements and abdominal ultrasounds, with initial results being negative.

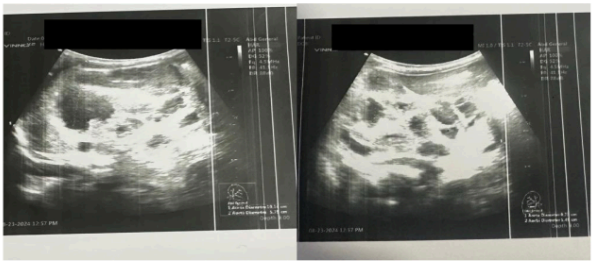


Figure 1: Antenatal abdominal ultrasound image showing a bilateral abdominal cystic mass (32 weeks of gestation).

Earlobe creases | *Ear creases* | *Macroglossia*

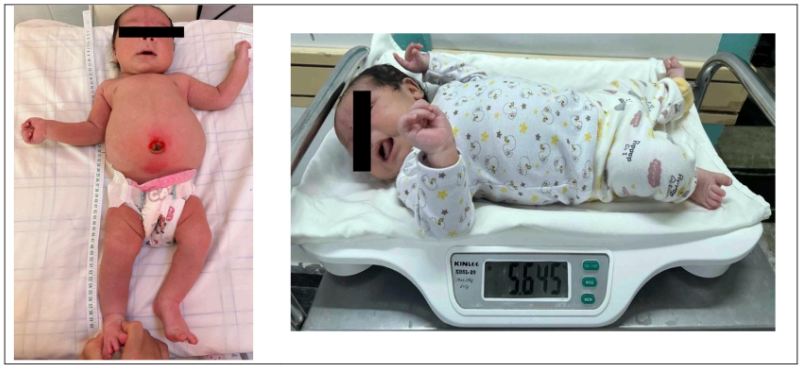
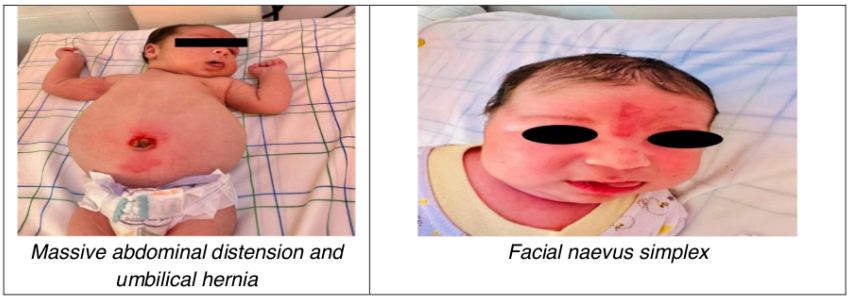


FIGURE 2: A photograph showing tall stature and macrosomia in the patient



Massive abdominal distension and umbilical hernia

Facial naevus simplex

FIGURE 3: Different signs of Beckwith-Wiedemann syndrome found in our patient.

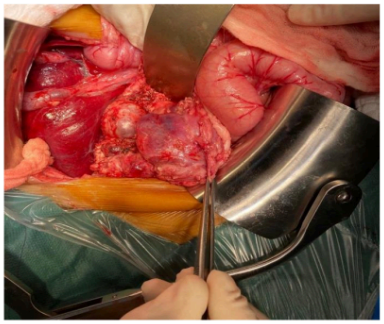


Figure 4: Operative photograph, showing the cystic mass.

DISCUSSION:

Beckwith-Wiedemann syndrome (BWS) is the most common congenital overgrowth syndrome. Diagnosis is most often made after birth. It predisposes to a high risk of embryonal tumors.(8)

Among the common clinical manifestations of this syndrome are adrenal anomalies, including benign bilateral hemorrhagic cysts, which, although usually asymptomatic, can lead to complications such as rupture, infection, or be associated with other

organ anomalies like hypertension (HTN), abdominal pain, or adrenal insufficiency. (9)

These cysts are often discovered during imaging studies in the first few months of life and typically tend to resolve spontaneously, resulting in a favorable prognosis. However, rigorous follow-up is needed to exclude malignant progression, although this is rare.(10)

In our study, a benign bilateral hemorrhagic adrenal cyst was diagnosed after surgery, with complete excision and histopathological analysis revealing no malignancy.

Bilateral hemorrhagic cysts in Beckwith-Wiedemann syndrome (BWS) are generally benign, but their association with other anomalies, such as macrosomia, macroglossia, and omphalocele, necessitates careful monitoring. The large size and bilaterality of the lesions in this population at risk for malignant tumors, combined with limited imaging information, may occasionally lead to surgical exploration. This approach allows confirmation of the diagnosis and exclusion of malignancy.(11)

Management of BWS involves regular monitoring of tumor markers, such as alpha-fetoprotein (AFP), along with periodic abdominal ultrasounds to detect any abnormal progression.(12)

The results of this study and other research underscore the importance of multidisciplinary follow-up and early screening to assess for the absence of tumor or endocrine complications associated with the syndrome.(13)

CONCLUSION:

In conclusion, although bilateral hemorrhagic cysts associated with Beckwith-Wiedemann syndrome are generally benign, their careful monitoring remains crucial due to their potential to cause clinical complications. Early and multidisciplinary follow-up is essential for optimal management and prevention of any adverse progression.

Figures :

Figure 2: Antenatal abdominal ultrasound image showing a bilateral abdominal cystic mass (32 weeks of gestation).

Figure 2: A photograph showing tall stature and macrosomia in the patient

Figure 3: Different signs of Beckwith-Wiedemann syndrome found in our patient.

Figure 4: Operative photograph, showing the cystic mass.

Conflicts of Interest :

The authors declare no conflicts of interest.

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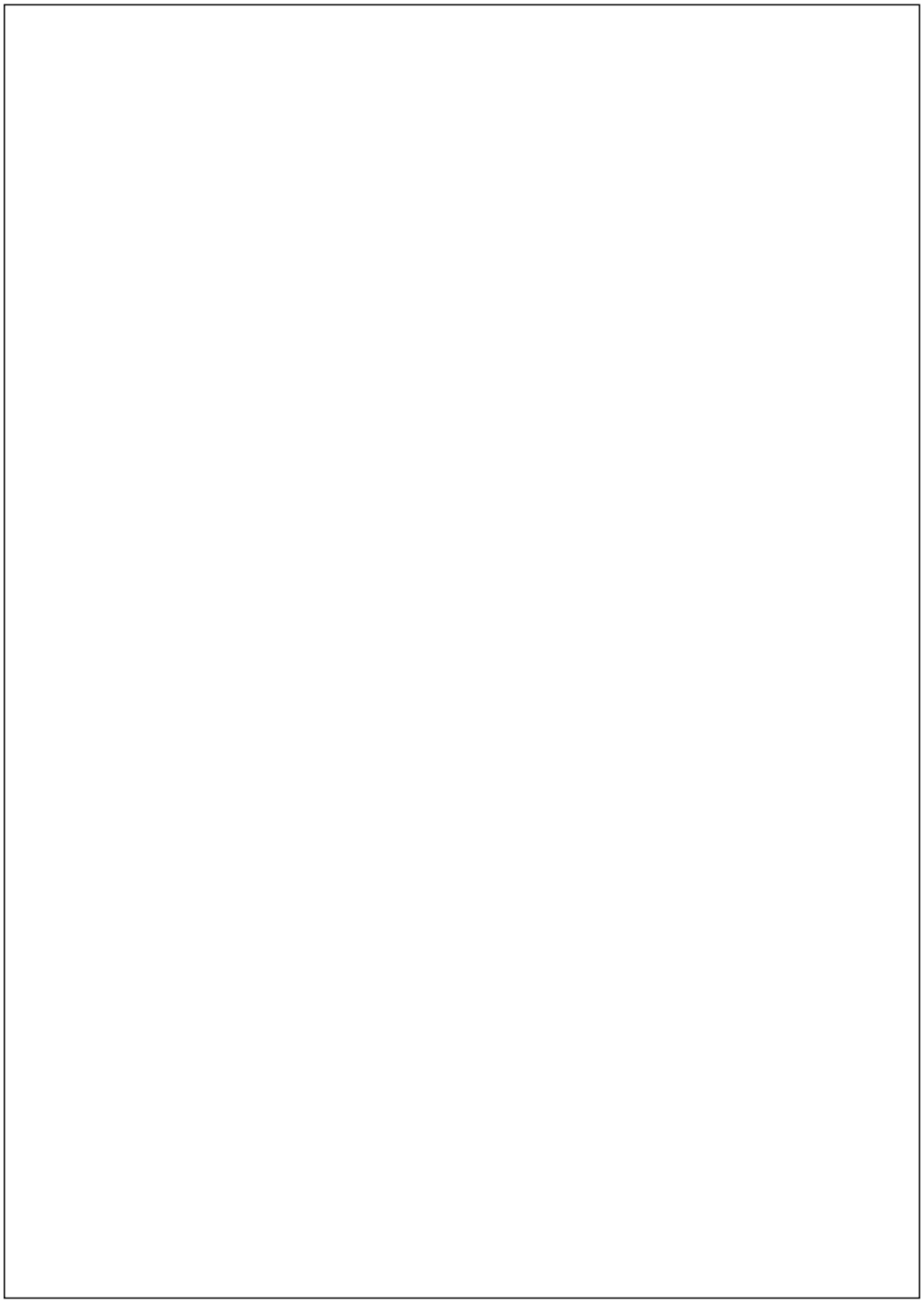

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