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

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

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### SUMMARY

Beckwith-Wiedemann syndrome is the most common overgrowth malformation syndrome. The classical features include macrosomia, macroglossia, omphalocele and ear lobe anomalies. Among the associated adrenal anomalies, foetal cortical cytomegaly, outer cortical haemorrhage and unilateral benign cysts are well described. A term neonate was admitted with typical features of the syndrome. Radiological evaluation revealed a rare association of bilateral benign hamorrhagic adrenal cysts. Serial sonography confirmed hamorrhage into benign cysts and ruled out neoplasms. Only one similar case has been documented in the literature previously.

### INTRODUCTION

Overgrowth syndromes are a group of disorders associated with excessive somatic growth and organomegaly. Among them, the best described is the Beckwith-Wiedemann syndrome (BWS), with an incidence of 1/13,700 births (1). The most consistent features of this syndrome are macrosomia, macroglossia, and omphalocele (2). In addition, several rare associations are also well known. A neonate with the typical features of BWS who had bilateral cystic haemorrhagic adrenal masses is reported.

### CASE REPORT

A 16-year-old primigravida delivered a female baby with a birth weight of 4350 gm (>90<sup>th</sup> percentile) at 37 weeks of gestation. The antenatal period was uneventful and antenatal ultrasonogram was not done. Baby was severely asphyxiated at birth and required immediate endotracheal intubation and positive pressure ventilation. Following resuscitation she was admitted in the neonatal intensive care unit.

#### Figure 1

*Neonate with macrosomia, macroglossia, distended abdomen with flank fullness and edematous umbilicus*

#### Figure 2

*CT scan abdomen showing enlarged left kidney (K) and bilateral multiple loculated adrenal cysts (C) with haemorrhage*

Clinical examination revealed macrosomia, macroglossia, linear fissures over both ear lobules, thick and edematous umbilical cord, distended abdomen with hepatosplenomegaly and bilaterally enlarged kidneys (Figure 1). There was no hemihypertrophy or vertebral anomalies. Serial monitoring of blood sugar revealed persistent hypoglycaemia. Serum insulin was 8.4 mIU/ml (normal range 6-27 mIU/ml). Haematocrit value was 68%. Ultrasonography of abdomen showed hepatosplenomegaly, nephromegaly and multiple large cysts in both adrenals, a few of which showed sediments in the dependent part. CT scan of abdomen showed bilateral nephromegaly and bilateral loculated suprarenal cystic masses (Figure 2). Some of the cysts showed hyperdense layering at the dependent parts, signifying haemorrhage within the cysts. After stabilization and initiation of supervised feeding, the baby was discharged and is under regular clinical and radiological follow up. A repeat sonographic evaluation two weeks after discharge revealed no change in the size of the cystic masses but showed regressive changes suggestive of haemorrhage.

## DISCUSSION

Beckwith-Wiedemann Syndrome (BWS) was first reported as a distinct clinical entity independently by Beckwith and Wiedemann in late 1960s. To date, about 200 such cases have been reported (3). This disorder seems to be caused by excess availability of insulin-like growth factor-2 encoded by a gene located at 11p15.5. The occurrence is usually sporadic.

The initial designation of the disorder as Exomphalos-Macroglossia-Gigantism (EMG) syndrome was based on the classical triad of external features. Later, abnormality of ear creases was identified as a fourth unique external marker. All the four characteristics were identified in the present case as well.

Hyperplasia and dysplasia of various abdominal organs are well described in BWS. These include nephromegaly, renal medullary dysplasia, polycystic kidney, pancreatic hyperplasia and various adrenal anomalies (2-4). Foetal adrenocortical cytomegaly is a constant feature of this syndrome (3,5). Cystic adrenal masses were reported for the first time in association with BWS in 1991 by Walton and colleagues (6). In the same year, bilateral haemorrhagic adrenal macrocysts were reported by McCauley *et al.* as a new pseudotumorous lesion in BWS (7). Subsequently, a few workers have confirmed this association. However, all these cases had unilateral adrenal cysts. In 1997, the first report of bilateral cystic adrenal masses in a neonate with incomplete form of BWS was made by Akata *et al.* from Turkey (8).

The infant reported here is the second documented instance of bilateral adrenal cysts in BWS. The infant showed all the classical features of complete BWS and in addition, had features of haemorrhage into the cysts. This form of adrenal haemorrhage can be differentiated from idiopathic neonatal adrenal haemorrhage by its location in the permanent (outer) cortex of the adrenal gland, in contrast to bleeding within the foetal (inner) adrenal cortex in the latter (7).

Because neonatal adrenal cysts cannot be clearly distinguished from malignant lesions like congenital cystic neuroblastoma, close radiological observation or surgical confirmation is needed in most cases (9). In the present case, hyperdense layering in the dependent parts suggested haemorrhage into the cysts. Subsequent

radiological evaluation revealed regressive changes confirming the diagnosis of benign haemorrhagic cysts and surgical intervention was thus deferred.

Other associated features such as polycythemia and hypoglycaemia were also encountered in our patient and were managed appropriately. Her large tongue demanded suitable interventions to maintain airway patency and to ameliorate feeding difficulties.

The presence of classical external markers renders early clinical diagnosis of BWS relatively easy. However, radiological evaluation is important for delineating associated lesions and prognostication in such neonates. Considering the relative rarity of this clinical entity, a high index of suspicion is warranted to identify hitherto unreported associations.

## REFERENCES

1. Sotos J. F. Overgrowth: Genetic syndromes and other disorders associated with overgrowth. *Clinical Pediat.* 1997; **36**:157.
2. Elliott, M, Bayly, R, Cole, T., Temple, I. K, and Maher, E.R. Clinical features and natural history of Beckwith-Wiedemann syndrome: Presentation of 74 new cases. *Clinical Genetics.* 1994; **46**:168-174.
3. Jones K.L. Beckwith-Wiedemann syndrome: In Smith's recognisable Patterns of Human Congenital Malformation. 164-167.
4. DeBaun, M.R, Siegel, M., and Choyke PL. Nephromegaly in infancy and early childhood: A risk factor for Wilm's tumor in Beckwith-Wiedemann syndrome. *J. Pediat.* 1998; **132**:401-404.
5. Martinez-y-Martinez R, Martinez-Carboney R, Ocampo-Campos R. Wiedemann-Beckwith syndrome: Clinical, cytogenetical and radiological observations in 39 new cases. *Genetic Counseling.* 1992; **3**:67-76.
6. Walton, G. R, Peng B.C, Berdon W.E., *et al.* Cystic adrenal masses in the neonate associated with hemihypertrophy and the relation to the Beckwith-Wiedemann syndrome. *J. Urology.* 1991; **146**:580-582.
7. McCauley, R.G., Beckwith, J.B., Elias, E.R., *et al.* Benign hemorrhagic adrenocortical macrocysts in Beckwith-Wiedemann syndrome. *Amer. J. Roentgenology.* 1991; **157**:549-552.
8. Akata, D, Haliloglu, M., Ozmen, M.N. and Akhan, O. Bilateral cystic adrenal masses in the neonate associated with the incomplete form of Beckwith-Wiedemann syndrome. *Pediat. Radiology.* 1997; **27**:1-2.
9. Hendry G.M.A. Cystic neuroblastoma of the adrenal gland: a potential source of error in ultrasonic diagnosis. *Pediat. Radiology.* 1982; **12**:204-206.