A RARE CASE OF BECKWITH-WIEDEMANN SYNDROME (BWS) WITH HEPATOBLASTOMA

INTRODUCTION:

The baby with BWS usually 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 abdominal mass. Also, some premature newborns with BWS might not have macroglossia.

CASE:

The patients with Beckwith wideman syndrome usually come to notice after findings of hemihypertrophy macroglossia and exomphalos. Here we are reporting a case 5-month female child came with complain of abdominal distension and palpable abdominal lump. Ultrasound and CT findings suggested of abdomino-pelvic mass lesion which was further worked up for biopsy and diagnosed as hepatoblastoma with raised serum AFP level >1000 IU/ml (0-10.0). On retrograde examination no evidence of omphalocele was seen but the child had gross macroglossia since birth and hemihypertrophy of right lower limb with organomegaly found in ultrasound examination. Macroglossia, a large tongue, is a very common (>90%) (4) and prominent feature of BWS. Infants with BWS and macroglossia typically cannot fully close their mouth in front of their large tongue, causing it to protrude out. This features concluded us to the diagnosis of Beckwith wiedeman syndrome.

Beckwith-Wiedemann Syndrome is an overgrowth disorder characterized by macrosomia, macroglossia, organomegaly and developmental abnormalities (in particular abdominal wall defects with exomphalos). Its incidence is estimated to be 1 per 13,700 live births (1). BWS patients are prone to the development of embryonal tumors (most commonly Wilm’s tumor or nephroblastoma and hepatoblastoma). BWS is a multigenetic disorder caused by dysregulation of gene expression in the imprinted 11p15 chromosomal region. The management of patients with BWS involves the surgical cure of exomphalos and monitoring of hypoglycemia in the neonatal period. It also involves the treatment of macroglossia and the screening for embryonal tumor. In case cases reported with hepatoblastoma, overall survival was high in patients with BWS and hepatoblastoma, especially given lower stage at presentation and when treated with surgery and chemotherapy (3).
DISCUSSION:
The baby with BWS usually 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. Another definition presented by Elliot et al. includes the presence of either three major features (anterior abdominal wall defect, macroglossia, or pre/postnatal overgrowth) or two major plus three minor findings (ear pits, nevus flammeus, neonatal hypoglycemia, nephromegaly, or hemihyperplasia).

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.

Here in our case patient has already developed a hepatoblastoma and presented with the secondary effects of mass lesion. Retrospective study of the case turned out to be the case of Beckwith Wiedeman syndrome with characteristic features of hemihypertrophy, macroglossia, organomegaly with hepatoblastoma. 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.

Hepatoblastoma requires surgical resection and followed by chemotherapy and regular followup and monitoring with serum AFP level.

CONCLUSION:
Radiologist should be alert while dealing with organomegaly to think in the line of Beckwith-Wiedemann syndrome; macroglossia could be a clue to the diagnosis. Presence of abdominal mass raises its chance of occurrence. In our case, hepatoblastoma was associated which is still rare to be found. Children with BWS have higher of getting cancers during early childhood and should undergo cancer screening during this period.

REFERENCES: