VOLUME - 12, ISSUE - 11, NOVEMBER - 2023 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

General Surgery

BECKWITH WEIDEMANN SYNDROME- A DIAGNOSTIC DILEMMA

Aditya Upadhyayula	Intern Esic Medical College, Sanathnagar Hyderabad- 500038
Yogi Nagender M	Consultant Department Of Pediatric Surgery Apollo Hospitals, Jubilee Hills Hyderabad- 500090
Srinivas Upadhyayula	Consultant Department Of Anaesthesiology Apollo Hospitals, Jubilee Hills Hyderabad- 500090

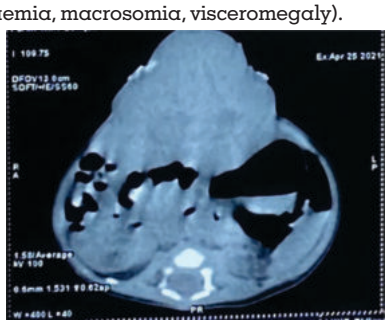
ABSTRACT Beckwith Wiedemann Syndrome (BWS) is a congenital condition characterised by overgrowth of different body parts which is usually manifested at birth. It is a rare condition where there may be hemi hyperplasia, omphalocele or other abdominal wall defects, hypoglycaemia in neonatal period, macroglossia, intra-abdominal visceromegaly, ear skin creases or pits, and renal abnormalities (Wilms tumor). They have high risk to develop tumours; especially Wilms tumour, hepatoblastoma, rhabdomyosarcoma. Degree of clinical manifestations vary from person to person as some may have all features while some may have only one of the many symptoms.

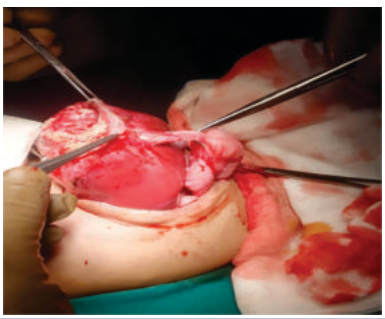
KEYWORDS :

CASE HISTORY

The given patient is a 2-month-old male with a birth weight of 2.39kgs and a present weight of 3.8kgs with a history of a late preterm delivery at 34+1 weeks with a history of omphalocele major. He also presented with mild colpocephaly, ASD/PDA, left dysplastic kidney, and indirect hyperbilirubinemia (G6PD deficient) requiring phototherapy. His BW- 2.39kg BL-43cm HC-32cm. now presented for further management of Omphalocele.

Clinically child had microcephaly, low set ears, flat nose, significant macroglossia more on left side, micrognathia, large omphalocele. Thyroid function normal. Blood urea and serum creatinine were normal. USG abdomen showed large membrane covered omphalocele, large dysplastic left kidney and normal right kidney, bilateral inguinal hernia and left undescended testis. CT scan showed gastroschisis with herniation of liver and large multicystic left kidney. 2D Echo showed PDA 2mm with L-R shunt, ostium secundum ASD 5mm. Dynamic Scinti-renography showed normal right kidney and large non-functioning left kidney. Child was clinically diagnosed as a case of BWS as fits 3 criteria (macroglossia, hypoglycaemia, macrosomia, visceromegaly).





DISCUSSION

BWS is diagnosed clinically, but genetic testing is better advised in suspected familial cases. Management includes initially management of complications as per usual protocols of isolated presentations of these conditions and then follow up. Management of complications includes as follows:

- Abdominal wall defects- omphalocele require emergency surgery to place the abdominal contents back into the abdomen, umbilical hernia wait and watch up to 2to 4 years and then surgery if not resolve its own, Diastasis recti usually requires no treatment.
- Neonatal hypoglycaemia- to be managed according to usual protocol for hypoglycaemia in neonates. Rarely (carcinoma, melanoma, rhabdomyosarcoma, and mesoblastic nephroma are present. Given the importance of early diagnosis, all children with BWS should receive cancer screening. USG Head shows evidence of mild colpocephaly and no other pathology identified. USG abdomen every 3 months until at least eight years of age and blood alpha-fetoprotein (AFP) estimation every 6 weeks until at least four years of age is recommended by some authorities. In general, the prognosis is very good. Children with BWS usually do very well and grow up to become the heights expected based on their parents' heights. While children with BWS are at increased risk of childhood cancer, most children with BWS do not develop cancer and the vast majority of children who do develop cancer can be treated successfully. Children with BWS for the most part had no significant delays when compared to their siblings. However, some children with BWS do have speech problems that could be related to macroglossia or hearing loss. Severe hypoglycaemia if occurs can otherwise cause developmental delay or seizures.

Conclusion BWS, if followed as per protocol have good prognosis.

Sources of Support: None
Conflicts of Interests: None.

REFERENCES

1. Beckwith, J. Bruce (1963). "Extreme cytomegaly of the adrenal fetal cortex, omphalocele, hyperplasia of kidneys and pancreas, and Leydig cell hyperplasia - another syndrome?". Annual Meeting of the Western Society for Pediatric Research. Los Angeles.
2. Wiedemann HR (September 1964). "Familial malformation complex with umbilical hernia and macroglossia - a "new syndrome"?". Journal de génétique humaine (in French). 13: 223–32.
3. Halliday J, Oke K, Breheny S, Algar E, J Amor D (September 2004).

"BeckwithWiedemann syndrome and IVF: a casecontrol study". American Journal of Human Genetics. 75 (3): 526–8. doi:10.1086/423902.
4. Gosden R, Trasler J, Lucifero D, Faddy M (2003). "Rare congenital disorders, imprinted genes, and assisted reproductive technology". Lancet. 361 (9373): 1975– 7. doi:10.1016/S0140-6736(03)13592-1
5. Li M. Molecular Genetics of Wiedemann Beckwith Syndrome (BWS) a case report and literature review . Am J Med Gen. 1998 Oct 2 ; 79(4) : 253-9. 4. Kang M. Inherited microdeletions that give rise to Beckwith–Wiedemann Syndrome. Clin Genet. 2005 Apr ; 67(4) : 299–300
6. Thorburn MJ, Wright ES, Miller CG, Smith-Read EH (April 1970). "Exomphalos-macroglossia-gigantism syndrome in Jamaican infants". American Journal of Diseases of Children. 119 (4): 316–21. doi:10.1001/archpedi.1970.02100050318006.
7. DeBaun MR, Niemitz EL, McNeil DE, Brandenburg SA, Lee MP, Feinberg AP (March 2002). "Epigenetic alterations of H19 and LIT1 distinguish patients with Beckwith-Wiedemann syndrome with cancer and birth defects". American Journal of Human Genetics. 70 (3): 604– 11. doi:10.1086/338934
8. DeBaun MR, Tucker MA (March 1998). "Risk of cancer during the first four years of life in children from The Beckwith-Wiedemann Syndrome Registry". The Journal of Pediatrics. 132 (3 Pt 1): 398–400. doi:10.1016/S0022-3476(98)70008-3