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Original Research Paper

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CASE REPORT OF BECKWITH-WIEDEMANN SYNDROME

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ABSTRACT **Introduction:-** Beckwith Wiedemann syndrome is rare, genetic condition. It is also known as exomphalos, macroglossia and gigantism syndrome. Incidence of Beckwith Wiedemann syndrome is 1:13700 births. Chromosome involved 11p15.5, it is also involved with an increased risk of cancer. **Case Report:-** 1 day old male child delivered at our hospital New civil Hospital Surat. Baby was delivered by LSCS due to breech presentation, delivered at 34 weeks of gestation. Resuscitation (Intubation) was required. Rbs on admission was 36 mg/dl iv. ANC USG suggestive of heptomegaly, nephromegly, macrosomia. Patient was kept on SIMV mode, antibiotic and GIR pint was started. Patient died due to sepsis and hypogycemia. **Discussion:-** It is a clinical diagnosis, characterised by omphalocele, gigantism, macroglossia microcephaly, visceromegaly, other features include lateral earlobe fissure, facial nevus flammeus, hemihypertrophy1. **Conclusion:-** Routine screening upto 8yr of age is required as they are at risk to develop cancer, diagnosis can be made by both antenatally as well as postnatally.

KEYWORDS : Beckwith Wiedemann syndrome, hemihypertrophy, gigantism, facial nevus flammus, macroglossia.

INTRODUCTION:-

Beckwith-Wiedemann Syndrome/ Overgrowth disorder/ multigenetic disorder/ imprinting disorder Characterised by Omphalocele, gigantism, macroglossia, microcephaly, visceromegaly. Other forms include lateral earlobe fissure, facial nevus flammeus hemihypertrophy.

According to De Bau et al [2] 2 out of 5 features must be present to call child as Beckwith-Wiedemann Syndrome (macroglossia, macrosomia, midline abdominal wall defect, ear crease, neonatal hypoglycemia) Elliat et al[3] :- either 3 major features and 2 major + 3 minor features must be present.

Major features include:- anterior abdominal wall defect, macroglossia or prepostnatal overgrowth.

Minor features include:- ear crease and pit, facial nevus flammeus, neonatak=1 hypoglycemia, nephromegaly or hemihyperplasia embryonal tumor, polyhydramnios.

Invitro-fertilization increases the risk by 3-4 times of developing Beckwith-Wiedemann Syndrome. Chromosome involved 11p15.5 Incidence of Beckwith-Wiedemann Syndrome 1:13,700 [1] births, equal in male and female.

Child with Beckwith-Wiedemann Syndrome are at risk of developing tumor (Wilms tumor, Hepatoblastoma, Adrenal carcinoma, gonadoblastoma, rhabdomyosarcoma) therefore until 8yrs of age regular follow-up with abdominal ultrasound is required.

Case Report :-

30yr old female $G_1P_1L_1A_0$, primi delivered male child at 32-34 wk gestation baby was born out of non-consanguinous marriage. Born out of primary infertility treatment. Pregnancy was a booked pregnancy, 3 antenatal visits were taken also TT injection was taken. Mother had GDM, hypertension, hypothyroidism and was on treatment for the same. ANC USG suggestive of macrosomia, hepatomegaly, nephromegaly, marginal and velamentous cord insertion, absent end diastolic flow, polyhydramnios. Baby delivered by LSCS. Birth weight was 2.5kg. Baby did not cry after birth so resuscitation [tactile stimulation, bag & mask, Intubation] was required.

Apgar score at 1min was 4/10 and at 5min was 7/10. On examination in NICU under warmer care temperature was normal, heart rate was 150/min, RR=54/min, RS=clear, CVS=no murmur, CNS=lethargic

PA: - Hepatomegaly present. Urine output & stool output present. Abdominal girth=34cm, which was static throughout NICU admission.

RBS was 36 mg/dl for which 2 ml/kg 10% IV glucose given repeat RBS 40 mg/dl so GIR6 (glucose infusion rate) started. Fenton chart plotted [suggested LGA baby]

On physical examination macroglossia, hemihypertrophy, macrosomia present. Baby was kept on SIMV mode, frequency 40, PEEP=5, Fio_2=100%, baby was started on GIR pint, antibiotic to cover sepsis, RT in-situ for feeding routine sampling like CBC, CRP, RFT with electrolyte, LFT with enzyme ABG and blood glucose level sent.

CXR done ABG suggestive of severe metabolic acidosis pH=7, PCo_2=26.5, PO_2=108, HCo_3=9.7 Bicarbonate correction given for same. Since baby was intubated so USG At pelvis, cranium was not done. Despite on GIR12 pint patient was not able to maintain blood glucose level, therefore Hydrocortisone was given. Patient condition was deteriorating day by day finally patient died due to hypoglycaemia and sepsis.

DISCUSSION:-

Beckwith-Wiedemann Syndrome also known as exomphalos, macroglossia and gigantism syndrome. Its is a clinical diagnosis. It can be diagnosed antenatally as well as postnatally, also genetically by molecular diagnosis. Antenatally diagnosed by ANC USG which is suggestive of excess liquor, increase in growth by 25-36wk, may or may not

show omphalocele. Postnatally diagnosis is based on large tongue, face, omphalocele, hypoglycemia, facial nevus flammeus, hepatomegaly, cardiac defect.

Isolated hemihypertrophy at risk of cancer[4,5] so periodic follow-up is necessary.

Abdominal USG and measurement of α-fetoprotein recommended every 3-monthly until the age of 8 years.

Since in our case patient died on 3rd day of life such follow-up was not possible. For abdominal wall defect treatment required is surgery. Tongue enlargement due to hypertrophy of muscle fibre leads to respiratory failure, feeding difficulty and speech difficulty, which can be managed by surgery.

Beckwith-Wiedemann Syndrome management require multidisciplinary team approach. Studies even show that artificial reproductive techniques show correlation with epigenetic syndrome like Beckwith-Wiedemann Syndrome and Angelmanns. Even in our case patient conceived by IVF. IVF may cause turning on or off gene[6,7]. Majority of the time Beckwith-Wiedemann Syndrome is caused sporadically, however 15% familial correlation also may be seen.

CONCLUSION:-

Beckwith-Wiedemann Syndrome is a Rare genetic disorder They are at risk to develop cancer so routine screening is required.

REFERENCES :-

1. Kliegman RM, St. Geme J. Nelson Textbook of Pediatrics E-Book. 21st ed. Elsevier; 2019.
2. DeBaun MR, Niemitz EL, McNeil DE, Brandenburg SA, Lee MP, Feinberg AP. Epigenetic Alterations of H19 and LIT1 Distinguish Patients with Beckwith-Wiedemann Syndrome with Cancer and Birth Defects. The American Journal of Human Genetics. 2002 Mar;70(3):604–11.
3. Elliott M, Bayly R, Cole T, Temple IK, Maher ER. Clinical features andnatural history of Beckwith-Wiedemann syndrome: presentation of 74 new cases.Clin Genet. 1994 Aug;46(2):168-74. doi: 10.1111/j.1399-0004.1994.tb04219.x.PMID: 7820926.
4. Chang AS, Moley KH, Wangler M, Feinberg AP, Debaun MR. "Association between Beckwith-Wiedemann syndrome and assisted reproductive technology: a case series of 19 patients". Fertility and Sterility 2005; 83 (2): 349354.
5. Choyke PL, Siegel MJ, Craft AW, Green DM, DeBaun MR. "Screening for Wilms tumor in children with BeckwithWiedemann syndrome or idiopathic hemihypertrophy". Medical and pediatric oncology 1999; 32 (3): 196-200.
6. DeBaun MR, Tucker MA. Risk of cancer during the first four years of life in children from The Beckwith-Wiedemann Syndrome Registry. J Pediatr. 1998 Mar;132(3 Pt 1):398-400. doi: 10.1016/s0022-3476(98)70008-3. PMID: 9544889.
7. Thorburn MJ, Wright ES, Miller CG, Smith-Read EH "Exomphalos-macroglossia-gigantism syndrome in Jamaican infants". American journal of diseases of children 1970; 119 (4): 316-21.