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Meckel Gruber Syndrome- A Case Report

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ABSTRACT
This case report is about a neonate delivered at our centre, with multiple anomalies – encephalocele, micro opthalmia, flat nasal bridge, cleft lip and palate, polydactyly, ambiguous genitalia, clinically suggestive of a rare syndrome- meckel gruber syndrome.

INTRODUCTION

Meckel-Gruber syndrome [ALSO CALLED DYSENCEPHALIA SPLANCHNOCYSTICA] is rare constellation of anomalies, autosomal recessive in inheritance, due to mutations in the MKS1 and MKS2 genes. INCIDENCE- 0.07- 0.7 / 10,000 BIRTHS.

CASE

- 1 day old baby [sex undetermined] was shifted from OT to NICU in view of
- severe birth asphyxia [APGAR less than 3 at 5 min]

MOTHER'S NOTES

Name : Afrin Mohd Jishan Siddhiqui
MRD no: 2111216
DOA: 7.2.17 at 8.16 am
Weight: 54kg
G3P1L1A1
MA : 28
MS: 6 YEARS [THIRD DEGREE CONSANGUINITY]
G3P1L1A1
LMP: 10.5.16
EDD : 17.2.17
REGISTERED UNDER DR RGD
BLOOD GROUP : AB NEGATIVE
HIV/HBSAG/VDRL : NEGATIVE
C/O LEAKING PV SINCE 4 HOURS BEFORE ADMISSION
NO H/O PIH APH
PPH
GDM
TB
DRUG / ALLERGY

- 2 DOSES OF INJ TT RECEIVED
- G1 :2 YEARS/ MCH/LSCS IVO PLACENTA PREVIA/ AT JJH / 2.5KG BW/ 30.9.14/ A AND W
- G2 : MTP [25.10.15]
- G3 : PP
- ANC USG:
- 10.10.16 – SLIUG/ 21.2 WEEKS/ OCCIPITAL ENCEPHALOCELE WITH HYDOCEPHALUS WITH B/L HYDROURETERS AND HYDRONEPHROSIS.
- 17.11.16- 24.5 WEEKS/ SAME

- Mother was admitted on 7th feb and taken up for emerg lscs on the same day indication being prev lscs with breech with prom with multiple anamolies with rh negative status with sct

Lscs was done under spinal anaesthesia and baby was delivered on 7.2.17 at 10 am.

Baby details :

DOB: 7.2.17

TOB: 10 AM

SEX: ambiguous genitalia [undetermined]

BW: 2.4 kg

Baby did not cry after birth

- No spontaneous respiration was present, bag and mask ventilation was started.
- After 30 sec, due to lack of response, baby was intubated and shifted to NICU on bag and tube ventilation.
- In NICU, baby was put on simv mode
- Fio2- 70% pip 14 peep 6
- On examination:
- Baby was hypothermic
- Hr- 126/ min

Rr- on venti

- No spontaneous activity
- Peripheral cyanosis +
- Pallor+
- Hc – 39 cm [encephalocele]
- Cc 32 cm
- Length- 51 cm
- Head to toe examination :
- Occipital encephalocele +
- Micro opthalmia +
- Flat nasal bridge

Low set ears

- Cleft lip and palate [b/l]
- Short neck
- Polydactyly in all four limbs
- Ambiguous genitalia
- Systemic examination
- Cvs : s1s2 hear , no murmur
- Rs : air entry greatly reduced bilaterally
- Pa: soft, no organomegaly

INVESTIGATIONS:

- Cbc was s/o
- Hb 17.1
- Wbc 6700
- Platelets 287000
- RFT:
- Creat 1.8
- CXR : s/o b/l homogenous opacification
- Abg :
- Ph 7.26
- po2- 87
- Pco234
- Hco3 9.6

DISCUSSION

- AUTOSOMAL RECESSIVE
- MKS1 AND MKS2 GENES INVOLVED
- INCIDENCE- 0.07- 0.7 / 10,000 BIRTHS
- ALSO CALLED DYSENCEPHALIA SPLANCHNOCYSTICA
- NAMED AFTER JOHANN MECKEL AND GEORGR GRUBER.
- DYSPLASTIC KIDNEYS PRESENT IN 95% CASES
- OCCIPITAL ENCEPHALOCELE PRESENT IN 60-80% CASES.
- POLYDACTYLY IS PRESENT IN 55-75 CASES
- THE ABOVE THREE FORM A TRIAD AND 2/3 IF PRESENT MAKE THE DIAGNOSIS SOLID.
- BOWING OR SHORTENING OF LIMBS ALSO COMMON
- OTHER FEATURES :
- MICO OPTHALMIA / ANOPTHALMIA
- LOW SET EARS
- CLEFT LIP/ PALATE
- MICOGNATHIA
- SHORT NECK
- PULMON AMBIGUOUS GENITALIA
- CARDIAC DISORDERS LIKE VSD, ASD, TOD, COARCTATION OF AORTA
- NEURAL TUBE DEFECTS
- HEPATIC DEVELOPMENTAL DEFECTS
- MORTALITY : 100%

- CAN BE DETECTED ANTENATALLY ON USG.
- GENETIC COUNSELLING AND ANC DIAGNOSIS AND TERMINATION ARE THE ONLY TREATMENT OPTIONS.