ORIGINAL RESEARCH PAPER

Neonatology

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A RARE CASE REPORT SUGGESTING MECKEL GRUBER SYNDROME IN ASSAM

Dr Yabom Jamoh* Department of Paediatrics, Fakhruddin Ali Ahmed Medical College and Hospital, Barpeta, Assam. *Corresponding Author

Dr Nazarul Islam Barbhyiyan Department of Paediatrics, Fakhruddin Ali Ahmed Medical College and Hospital, Barpeta, Assam.

Dr Sahidul Islam Department of Radiodiagnosis, Fakhruddin Ali Ahmed Medical College and Hospital, Barpeta, Assam.

ABSTRACT

Meckel Gruber Syndrome/Dysencephalia Splanchniococystica is a rare and lethal autosomal recessive disorder described by Friedrich Meckel and George B. Gruber with a worldwide incidence of 1 in 13,250 to 1,40,000 live birth characterized by occipital encephalocele, bilateral renal dysplasia and polydactyly. We report a rare case suggestive of Meckel Gruber Syndrome delivered live at 28 weeks gestation.

INTRODUCTION

Meckel Gruber Syndrome is a rare and lethal ciliopathic autosomal recessive disease and carries 25% recurrence in each pregnancy[1]. Since the time it was first reported by Meckel in 1822; only 200 cases have been reported till now[2]. It is characterized by multi-system development malformation and classical features of occipital enceph alocele, polydactyly and renal dysplasia. The criteria for diagnosis is the presence of any two of the three classical feature[3,4].

Our reported case is associated with polydactyly, occipital encephalocele and other associated features. This kind of ailment is infrequently reported in literature.

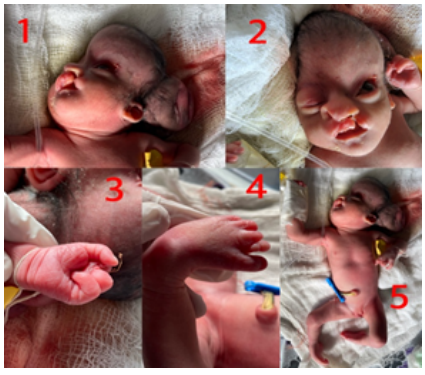

Figure 1-5: showing the major congenital anomalies associated with Meckel Gruber Syndrome

Case Report

A 27 yrs multigravida female delivered a female baby at 28 weeks gestation by normal vaginal delivery in Fakhruddin Ali Ahmed Medical College and Hospital, Barpeta, Assam.

The Baby did not require active intervention at birth with APGAR score of 8/10 at 1 and 5 mins of life. Baby was admitted immediately after birth in SNCU and given neonatal care as per institute protocol.

On examination

Baby weighed 2.5 kgs, length-43cm, Head circumference-22cm, Heart rate-132 bpm, Respiratory Rate-40 pm, Capillary refill time less than 3 secs, associated with major congenital anomalies- Occipital and frontal encephalocele(Fig1), cleft lip and palate(Fig2), syndactyly, fibrous bands in left hand and polydactyly in left foot(Fig3,4), micropthalmia, broad nasal root and hypertelorism(Fig2), bilateral congenital talipes equinovarus(Fig5). On Systemic examination, no obvious abnormality was detected.

Maternal History

Mother had a non-consanguineous marriage, third gravida. From her first pregnancy, she has a living female child. On her second pregnancy, a pair of twins was delivered prematurely at 24 weeks gestation; a male and a female, expired at one hour of life with no obvious, similar anomalies.

In all her pregnancies, she did not attend a single antenatal check-up.

There was no history of similar anomalies in the family.

Investigations

Ultrasound whole abdomen-bilateral kidneys showed increased echogenicity with prominent renal pyramids, however, no cystic lesions visible in bilateral kidneys. Liver showed coarse echotexture.

Xray Skull-defect seen in the parieto-occipital bony calvarium with herniation of intracranial content posteriorly suggestive of encephalocele.

Chest Radiograph-enlarged right border cardiac contour with predominant vasculature.

Ultrasound cranium-Parieto-occipital calvarium appears with a large heteroechoic brain parenchyma protruding through skull vault with marked ventriculomegaly.

Based on the clinical and radiological findings, a diagnosis of Meckel Gruber Syndrome was considered as per the criteria for diagnosis of MGS.

The Parents left with the baby against medical advice at 27 hours of life and the baby expired at 48 hours of life.

DISCUSSION

Meckel Gruber Syndrome is a lethal syndrome causing anomalies of CNS, cystic dysplasia of kidneys, malformation of extremities. Other anomalies associated with MGS are IUGR, single umbilical artery, CVS defects, cleft lip and palate, several genital abnormalities, oligohydramnios and hepatic fibrosis[5].

In accordance with the disease, our patient had occipital encephalocele, polydactyly, syndactyly, cleft lip, cleft palate,

bilateral CTEV. Ultrasound whole abdomen findings showed increased echogenicity with prominent renal pyramids but no prominent cystic lesions were visible in both the kidneys.

MGS is usually diagnosed prenatally during second trimester by ultrasonography with classical triad most commonly seen before 14th week of gestation. If diagnosis is done by prenatal USG, termination of pregnancy can be done. When available, autopsy and genetic analysis are the gold standard for diagnosis[8].

Our patient was diagnosed live and diagnosis was considered on the basis of clinical and radiological findings. The family did not accept autopsy after death.

There is no effective treatment for MGS and mortality is 100%. Most infants are stillborn or die early. Gazioglu et al reported an unusual case of MGS dying at 7 months of age[10].

Our case expired at 48 hours of life after LAMA and cause of death is not known.

CONCLUSION

Meckel Gruber Syndrome is a lethal syndrome but it can be diagnosed during antenatal USG. As the recurrence rate is 25%, the parents were counselled accordingly.

Our aim is to spread awareness regarding the need for earlier diagnosis of rare anomalies and the importance of antenatal visits in a developing country, like, India.

REFERENCES:

1. Yuksel MA, Mamaelov 2, Sofiyara Avici, Davutoglu E, Temel Yuksel J, Madazli R. An unusual case of Meckel-Gruber syndrome associated with visceroatrial and facial anomalies. J. Obstet Gynaecol 2016, 36:524-4
2. Jha T, Bardhan J, Das B, Patra KK, Dhali B, Seth S. Meckel-Gruber syndrome. A rare entity. J Indian Med. Assoc. 2010; 108:611-2
3. Basisic I, Boban L, Loane M, Galve E, Wellesley D, Calzolari E, et al. Meckel Gruber syndrome-a population based study on prevalence, prenatal diagnosis, clinical features and survival in Europe En J Hum Genet 2015;23:746-52
4. Jeevika M, Reddy TA, Kumar K A, Lona Reddy R. Meckel Gruber syndrome- a case report. IOSR J Dent Med Sci Ver III 2016;15:2279-861
5. Nargiz S, Sezer SD, Altinkaga SO, Yuksel H. Meckel Gruber syndrome- a case report and review of literature. Gynecol Obstet Reprod Med. 2016; 20
6. Gupta M. Mehta A, Gupta R, Singh S. Prenatal diagnosis of MGS with Dandy walker malformation. J K SG J Med Educ Res 2005;7:164-6
7. Szymanska K, Berry I, Logan CU, et al. Founder mutations and genotype phenotype correlations in Meckel Gruber syndrome and associated ciliopathies. Cilia 2012; 1:18-26
8. Myageri A, Grampurohit V, Rao R. Meckel Gruber Syndrome: Report of two cases and review of literature. J Family Med Prim Care 2013;2:106-18
9. Panduranga C, Kangle R, Badami R, Patil PV. Meckel Gruber syndrome: report of two cases. J Neurosci Rural Pract. 2012;3:56-9
10. Gaziglu N, Vural M, Seckin MS, et al. Meckel Gruber syndrome. Childs Nerv system 1998;14:142-5