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Original Research Paper

Radio-Diagnosis

MECKEL GRUBER SYNDROME – A CASE REPORT

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ABSTRACT Meckel Gruber Syndrome (MKS) is a rare autosomal recessive malformation syndrome characterized by multiple congenital anomalies ultimately leading to the death of fetus in utero or shortly after birth. It is characterized by classical triad of occipital encephalocele, infantile polycystic kidneys and postaxial polydactyly. Diagnosis of MKS is made on the basis of ultrasonography, gross morphology & histopathological findings. Here, we describe a case of MKS presenting with the classical triad. (5)

KEYWORDS : Meckel Gruber, autosomal recessive, occipital encephalocele, polycystic kidneys, postaxial polydactyly

INTRODUCTION

Meckel-Gruber syndrome (MGS), also known as dysencephalia splanchnocystica (3) is classically characterized by the triad of:

1. Renal cystic dysplasia: multiple renal cysts (present in most cases)
2 Occipital encephalocele / holoprosencephaly (~70%)
3. Postaxial polydactyly: usually hexadactyly (~65%)

Epidemiology

The incidence is estimated to be 1:30,000. A disproportionately higher prevalence may be present in Finland, Belgium and in some parts of India in Gujarati population. (3)

Risk Factors

Recent studies suggested that pregnancies obtained following ART may have an increased rate of major malformations due to parental factors causing the infertility.

Furthermore, it was recently reported that IVF and intracytoplasmic sperm injection (ICSI) are associated with imprinting disorders in the offspring. (4)

Associations

Central Nervous System / Craniofacial

- Microcephaly
- Foetal ventriculomegaly
- agenesis of corpus callosum
- Dandy-Walker malformation
- micrognathia
- cleft lip +/- palate

Cardio Vascular Abnormalities

- Ventriculo- septal defect (VSD)
- aortic hypoplasia
- aortic coarctation
- aortic valve stenosis

Musculoskeletal Abnormalities

- club foot
- syndactyly
- clinodactyly

Genetics

Meckel-Gruber syndrome shares some features with trisomy 13 and is therefore also termed pseudo trisomy 13 [1]. Karyotyping is recommended if the above triad is seen on antenatal scanning. (1)

Markers

Maternal alpha-fetoprotein (MSAFP)

Case Study

Clinical History

A 26-year-old third gravida female having history of previous two 2nd trimester spontaneous abortions presented for her routine 2nd trimester antenatal scan for the first time without any active complaints/ history of any drug intake, DM, HTN, TB OR COVID

Imaging Findings

On antenatal ultrasound, there is occipital meningocele, multi cystic dysplastic kidneys, polydactyly, cerebellar and lung hypoplasia, right club foot and anhydramnios.

-Suggestive of Meckel Gruber syndrome.
With this provisional diagnosis, further foetal MRI was advised and ultrasound findings were confirmed.

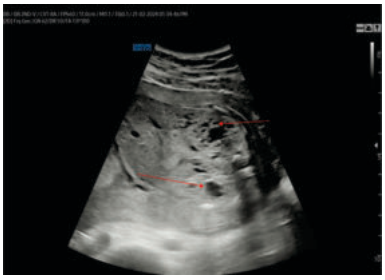

Figure 1a. Axial Image Of Antenatal USG Shows Multi Cystic Dysplastic Kidneys.

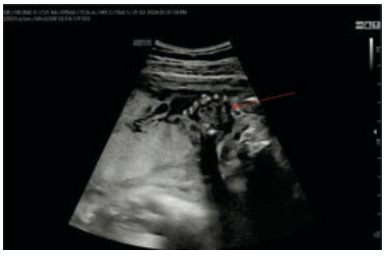

Figure 1b. Axial Image Of Antenatal USG Shows Polydactyly

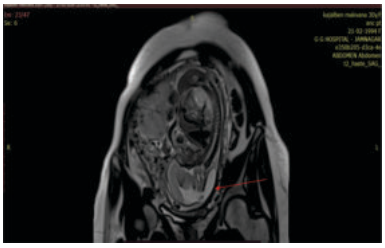

Figure 2a. MRI T2W Sagittal Image Shows Occipital Encephalocele.

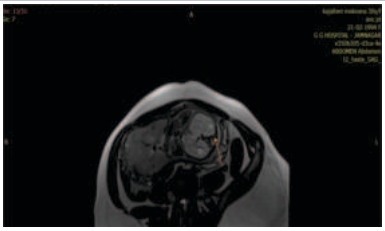

Figure 2b. MRI T2W Axial Image Shows Multi Cystic Dysplastic Kidneys.

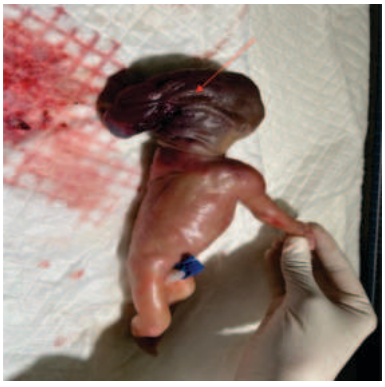

Figure 3a. Gross Image Of Baby Showing Occipital Encephalocele.

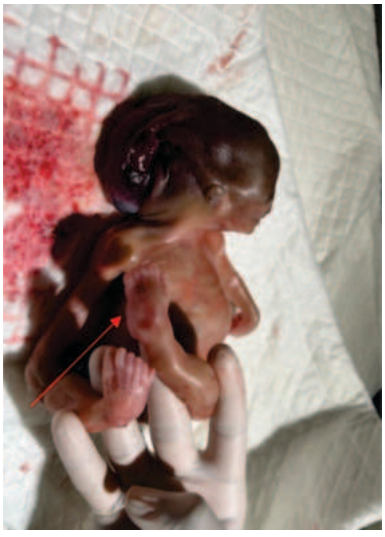

Figure 3b. Gross Image Of Baby Showing Polydactyly And Club Foot.

CONCLUSIONS

MGS is a lethal disorder. One cannot speak about survival of the fetus because of the pulmonary hypoplasia. The parents should be counseled about prognosis of the fetus and the outcome. Counselors should strictly give information about the recurrence risk for the next pregnancies. (6)

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