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"Meckel Gruber Syndrome with Anencephaly—A Rare Case Report"

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Abstract:
Meckel–Gruber syndrome (MGS) is a serious disorder of ciliopathic origin and is lethal in nature with guarded prognosis. As per available literature its incidence is quite rare. It is a multisystem developmental disorder associated with various malformations like occipital encephalocele, post-axial polydactyly and renal cystic dysplasia. Other nervous system abnormalities are less frequently reported which include hydrocephaly, anencephaly or malformation of cerebellum. We are presenting this case of MGS associated with anencephaly. Main goal of this case report is to highlight the importance of ante natal check-up along with ultrasound that precisely diagnose the malformations of Central Nervous System (CNS) and alerts a physician to prescribe medication e.g. folic acid as a preventive measure against major defects and also aids in active management of other neural tube defects

Key Words: *MGS (Meckel Gruber Syndrome), CNS (Central Nervous System),*

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I. Introduction

Meckel–Gruber syndrome (MGS) presents a rare and lethal ciliopathy, with an incidence of approximately 1:30,000 live births with a wide range varying between1:13000 to 400000. This geographical variation is seen particularly in India, Belgium and Finland where incidence is more. It follows an autosomal recessive approach with 25% risk of recurrence in each pregnancy [1, 2, 3]

MGS is a developmental disorder characterized by malformations involving multiple systems. Classically, the features apparent are renal cystic dysplasia, occipitalencephaloceleandpost-axialpolydactyly. Presence of at least two of the threeclassicfeatures makes diagnosis of MGS possible. According to the frequency dysplasticcystickidney is found in 100% followed by occipital encephalocele (90%) and polydactyly (83%) respectively.[4,5] Other significant CNS abnormalities associated with MGS are not frequently reported in literature e.g. hydrocephalus or anencephaly.

II. Case Report

A 28-year-old woman from higher mountainous reaches of far flung area of district Doda, J&K, having married consanguineously with gravida 2 para 1 presented to our hospital at 36 weeks of gestation with the complaints of abdominal pain (Labor pain). Her previous obstetric history revealed that she had given birth previously to a healthy boy who was doing well at 5 years of age. As a routine procedure, patient was sent for ultrasonographic scan for fetal well-being and bio physical profile. USG report revealed live fetus with multiple anomalies including dysplastic kidneys, marked oligohydramnios, anencephaly (floating brain matter) and prominent orbits. (Fig. III) The mother was counselled regarding the poor fetal prognosis.In next few hours,sheexperiencedlabourpains and gave

birth to a male child weighing 2250grams vaginallywith an Apgar score of 5 in 1 min and 7 in 5 min. On physical examination, new-born revealed anencephaly(Fig.II)withan occipito frontalcircumferenceof 24cm,occipital encephalocele (Fig. II), abnormal ears (Fig. II),edematous lids of both eyes(Fig. I),distendedabdomen, club feet andpost-axialhexadactylyonboth sides of hands and feet.New-born was kept under warm incubator care with wet dressing over exposed occipital

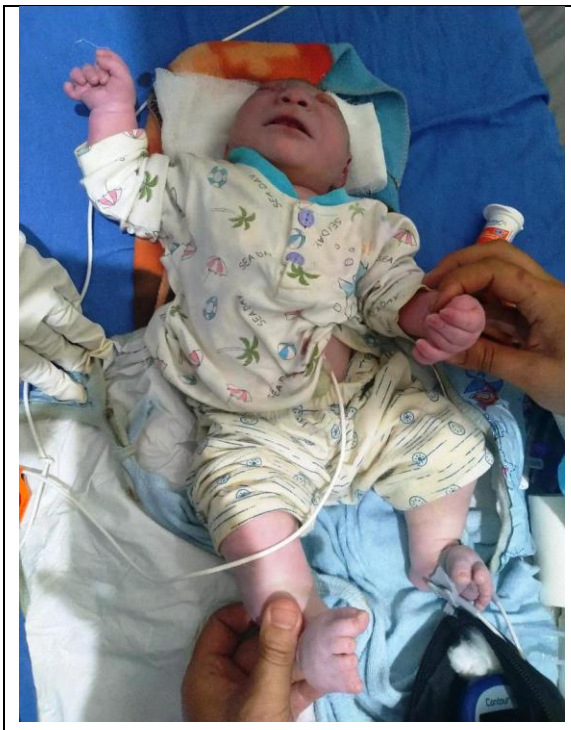
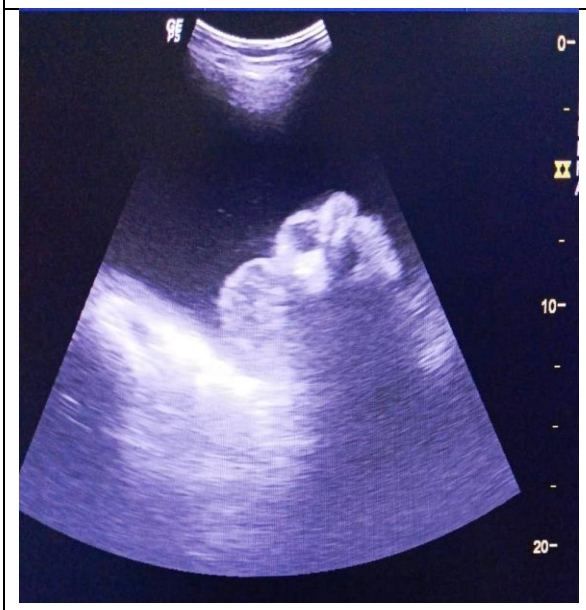


Fig I: MGS with anencephaly and hexadactyly of hands and feet.

Fig II: MGS with occipital encephalocele and abnormal ears.



FigIII: Showing floating brain matterin anencephaly with prominent orbits.

Encephalocele. Upon insistence of parents baby was transferred to a tertiary care hospital for further management. However, afterfew hours baby expired.Based on classical clinical features we diagnosed the case as MGS. Parents didn't give consent for autopsy and genetic analysis further on.

III. Discussion:

It was Opitz and Howe who coined the termMeckel Gruber Syndrome, however actually, the syndrome is named after famous German anatomistJohann Friedrich Meckel (also popular for Meckel diverticulum) and famous physician Georg Gruber who had termed it asdysencephalia splanchnocystica. [6] MGS is lethal in nature causing serious anomalies of central nervous system (CNS), cystic dysplasia of the kidneys and polydactylyof the extremities. Two of the above 3 cardinal features need to be present for calling it MGS. Various other anomalies have been observed to be associated withMGS includingintrauterinegrowthretardation(IUGR), facial abnormalities (cleft lip/palate), cardiovasculardefects,genitalabnormalities andoligohydramnios. [7,8]With advancement of ultrasonography where 2D/3D/4D scans are available, gives a clear picture of fetus and its body parts. Anomaly scans are now part and parcel of ante natal checkups. It can precisely diagnose such CNS abnormalities. Neural tubes start closing after 11 weeks of gestation and upto 14 weeks, diagnosis can be made and active management can be taken depending upon current medical/surgical options available. CNS abnormalities associated with MGS have been previously reported also in the literature including occipital encephalocele, hydrocephaly, microcephaly, Dandy–Walker malformation and even holoprosencephaly.[7, 9] Anencephaly and hydrocephaly are seen less frequently as per literature available. Data provided by European Surveillance of Congenital Anomalies (EUROCAT) network show a recent population-based study of 173 cases of MGS where only 3.5% cases were associated with anencephaly. Another study done in Finland, where 67 cases of MGS showed only onecasewithanencephaly[3,6] As far as our case is concerned , we found all the cardinal features of MGS along with anencephaly which is quite rare. Differentially we need to keep certain chromosomal anomalies in mind, like trisomy 13 where midline CNS abnormalities are more frequently found [5, 9]

IV. Conclusion:

As specific biochemical investigations and chromosomal studies are not routinely indicated. Therefore,prenatal ultrasonography is important for the diagnosis, which can be confirmed later by genetic analysis and careful post-mortem findings. The importance of early diagnosis is effective timely counseling of affected mothers regarding the medical/surgical options of termination and prenatal genetic testing in future pregnancies.

CONFLICT OF INTEREST:
None
CONSENT:
Informed written consent was taken from the guardians of the patient.

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