

## Meckel-Gruber Syndrome- A Rare Case Report

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### Abstract

**Introduction:** Meckel-Gruber Syndrome was first described by J R Meckelin the year 1822. It is an autosomal recessive disorder, and is caused by the failure of mesodermal induction. The typical triad of Meckel-Gruber Syndrome (MGS) involves meningo-encephalocele, polycystic kidneys and postaxial polydactyly. Any two of these features with a normal karyotype are diagnostic of this syndrome. The worldwide incidence varies from 1 in 1.300 to 1 in 140.000 live births.

**Case:** In this report, we present a case of MGS in which the diagnosis was made at around 15 weeks of gestation based on ultrasonographic findings (encephalocele, polycystic kidneys) These features were suggestive of the diagnosis of Meckel Gruber Syndrome (MGS). The patient was counseled regarding the lethal outcome of MGS. Fortunately, the family agreed for the termination of pregnancy..

**Conclusion:** 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.

**Keywords:** Encephalocele, Meckel Gruber, polycystic kidneys, postaxial polydactyl

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### I. Introduction

Meckel-Gruber Syndrome was first described by J R Meckel in 1822<sup>[1]</sup> It is an autosomal recessive disorder, and is caused by the failure of mesodermal induction. Bardet–Biedl Syndrome, Alstrom Syndrome and Joubert Syndrome belong to the same group of disease<sup>[2]</sup>The typical triad of Meckel-Gruber Syndrome (MGS) involves meningo-encephalocele, polycystic kidneys and postaxial polydactyly. Other abnormalities associated with MGS include cleft palate, cardiovascular diseases, hepatic ductal dysplasia, oligohydramnios, genital deformations, bowed legs, microcephaly and hydrocephalus. The worldwide incidence varies from 1 in 1.300 to 1 in 140.000 live births. The highest incidence was seen in the Gujarati Indians (1:1.300), and then in Finlands (1:9.000).<sup>[3]</sup>To date, above 200 cases of MGS have been reported in literature. In this report, we present a case of MGS in which the diagnosis was made at 15 weeks of gestation based on ultrasonographic findings of encephalocele and polycystic kidneys (**Figs. 1 and 2**).

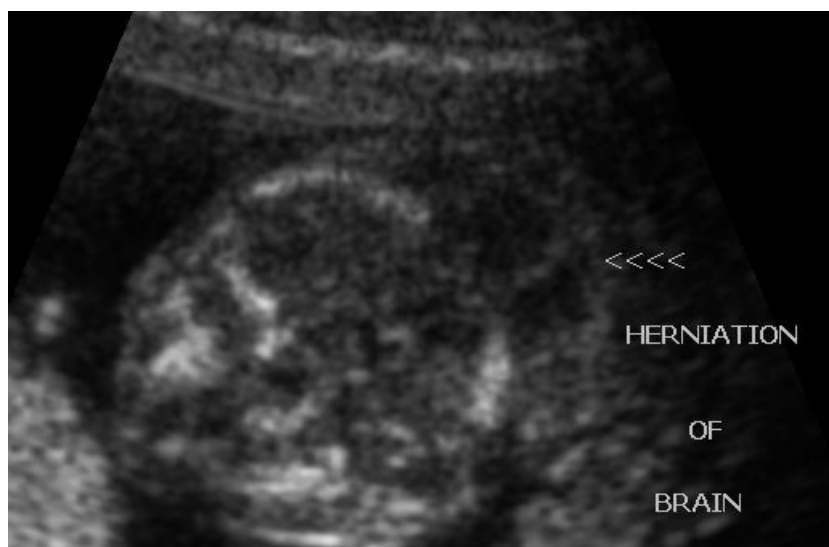


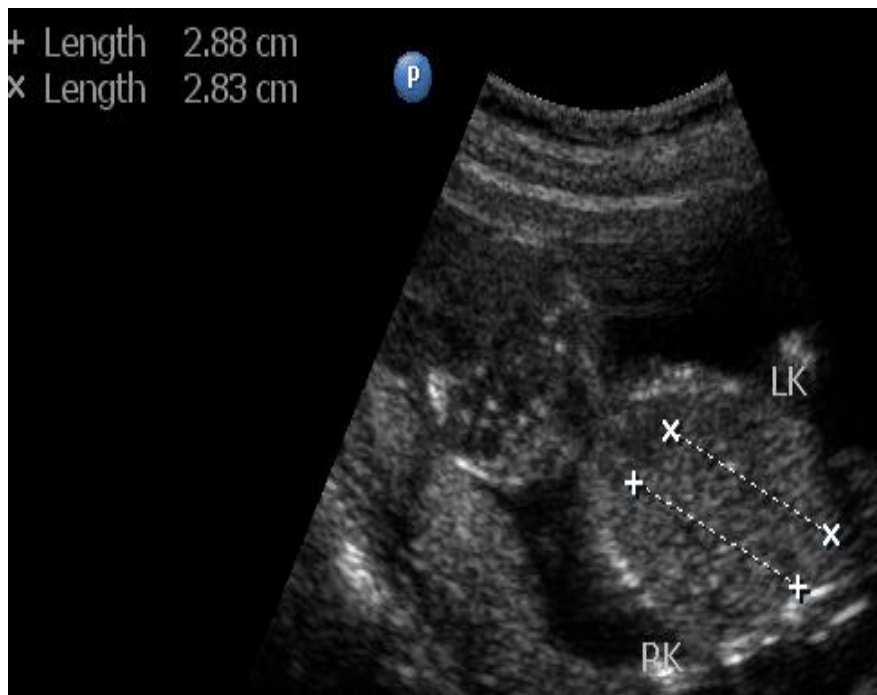
Fig 1 : Ultrasound image of Encephalocele



**Fig 2 :** Ultrasound image of polycystic kidneys.

## II. Case Report

A 23 yr old primigravida with consanguineous marriage presented to our clinic for routine antenatal checkup at 15 weeks of gestation. She had previous 2 healthy full-term pregnancies. Triple test was performed, and neural tube defect risk was found to be elevated (108 IU/ml - 3.7 MoM).USG done on this patient revealed enlarged kidneys Fig 3 showing numerous cysts Fig 4& occipital encephalocele Fig 5 There was no polydactyly. The abdominal circumference was increased due to enlarged kidneys, corresponding to 16 weeks 2 days Fig 6. However, femur length corresponded to 14wk 5days fig 7. No other abnormalities were noted as per the gestational age of the fetus .Liquor was adequate for gestational age.The fetal karyotyping done did not reveal any chromosomal anomaly.Thus a diagnosis of Meckel Gruber Syndrome was made.Fortunately, the family agreed for termination of pregnancy.The fetus had an occipital encephalocele protuberant abdomen, and palpable kidneys. Genetic or pathologic postmortem investigation could not be performed because of the patient's reluctance. Diagnosis of MGS was ascertained based on prenatal and postnatal features.



**Fig 3** antenatal scan showing enlarged kidneys



Fig 4 antenatal usg showing bilateral polycystic kidneys.

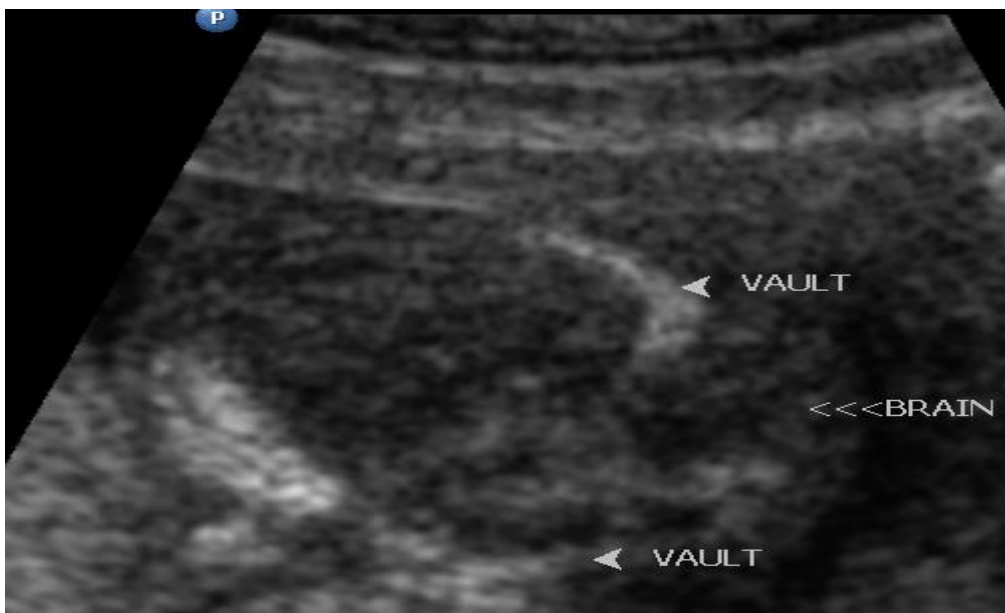


Fig 5 Antenatal USG showing occipital encephalocele

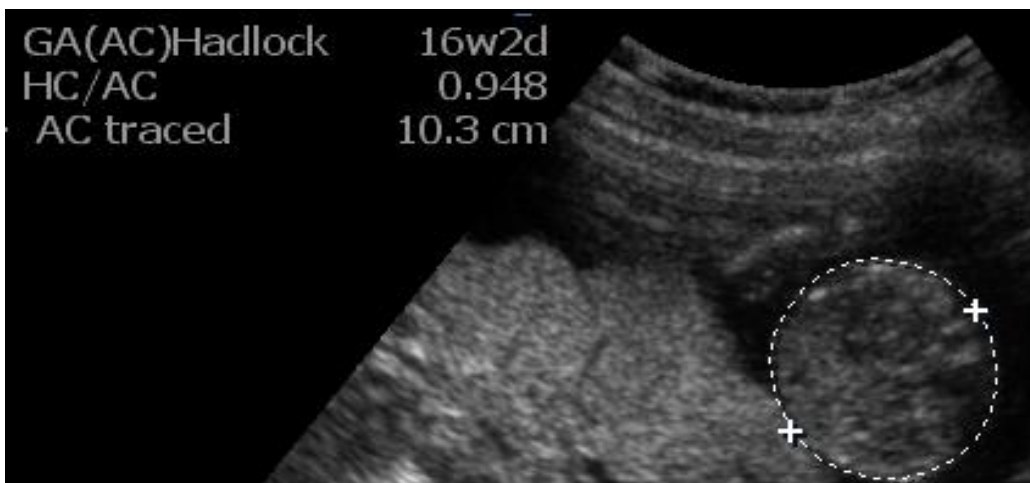


Fig 6 Antenatal scan showing increased AC

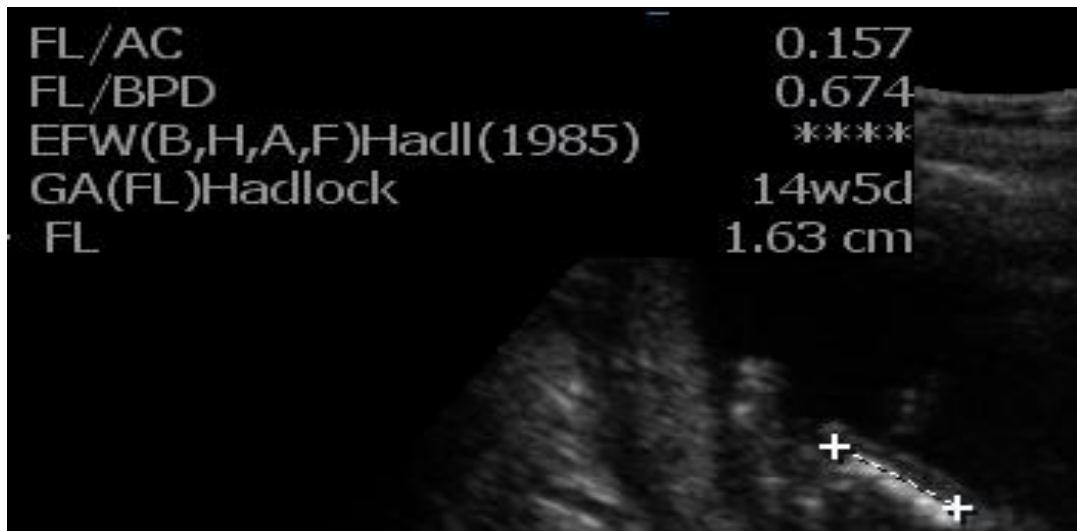


Fig 7 FL corresponding to 14wk 5days.

### III. Discussion

MGS was first described by J R Meckel in 1822 in 2 neonatal babies who died because of multiple organ abnormalities (encephalocele, polycystic kidneys, polydactyly).<sup>[1]</sup> In 1934, George B Gruber reported familial cases with similar features.<sup>[4]</sup> MGS affects both genders equally, and consanguinity has been reported to be an important factor in the genetic basis of the disease. Six different loci in different chromosomes have been shown in MGS. These loci are 17q21–24 [transmembrane protein meckelin (MKS) 1], 11q13 (MKS2), 8q21.3–q22.1 (MKS3), 12q21.31–q21.33 (MKS4), 16q12.2 (MKS5), and 4p15.32 (MKS6).<sup>[5]</sup> This variability in gene loci suggests genetic heterogeneity in MGS. The typical presentation of MGS involves the clinical triad of meningo-encephalocele, enlarged polycystic kidneys and post axial polydactyly. To ascertain the diagnosis, 2 of the 3 major abnormalities should be present. The most specific abnormality is encephalocele, whereas the most seen abnormality is the polycystic kidneys.<sup>[6]</sup> Oligohydramnios, and therefore, pulmonary hypoplasia occur because of nonfunctional dysplastic kidneys, which enlarge even up to 15–20 times. MGS is usually diagnosed on ultrasonography in the second trimester. The mean gestational age at diagnosis is 19 weeks.<sup>[7]</sup> In such cases,  $\alpha$ -fetoprotein (AFP) levels rise in maternal serum between 11 and 16 weeks of gestation.<sup>[3]</sup> Chromosome analysis by amniocentesis or chorionic villous sampling is also an important diagnostic method.<sup>[6]</sup> Review of the literature reveals that mortality due to MGS is 100%. Neonates die just after the delivery. Just only 2 cases of MGS could survive beyond infancy.<sup>[2],[3],[4]</sup> These cases survived at most 14<sup>th</sup> and 28<sup>th</sup> months.

### IV. Conclusion

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.

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