

Meckel-Gruber Syndrome: A Case Report

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Citation

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Abstract

Meckel-Gruber syndrome is a rare and lethal autosomal recessive disorder characterized by occipital encephalocele, postaxial polydactyly and bilateral dysplastic cystic kidneys. It can be associated with many other conditions. Antenatal ultrasound examination establishes the diagnosis by identifying at least two of the major features described.

INTRODUCTION

Meckel-Gruber syndrome is a rare and lethal autosomal recessive disorder characterized by a multitude of congenital anomalies. Antenatal ultrasound examination aids to correct diagnosis by identifying various anomalies. We report a rare case of Meckel-Gruber Syndrome identified by antenatal ultrasound.

CASE REPORT

A 26 year old multigravida (G2P1L0) presented at 8 weeks gestation with previous obstetric history of a spontaneous fetal loss at twenty six weeks. As per her previous records pathological examination of the fetus revealed an encephalocele. Pedigree analysis revealed a history of first degree consanguinity between the parents. Family history was non-corroborative. There was no history of intake of teratogenic drugs and other relevant past illness. She was on regular folic acid supplementation since beginning of conception. An anomaly scan at 12 weeks gestation performed with level II high resolution ultrasound revealed an encephalocele, bilateral polycystic kidneys and syndactyly. (Fig.1).

Figure 1

Fig.1. Transverse section of the fetal abdomen – bilateral enlarged, hyperechoic cystic dysplastic kidneys



Considering the syndromic association between these features a possibility of Meckel Gruber's syndrome was hinted and the situation with its possible outcome explained to the parents. The couple decided to terminate the gestation and misoprostol was administered. The 12 week fetus was delivered intact. Gross examination revealed a small cranium, micrognathia, encephalocele, and syndactyly.(Fig.2).

Figure 2

Fig. 2. Fetus after delivery: small head with encephalocele, micrognathia, protuberant enlarged abdomen, bilateral clubfeet and polydactyly



Autopsy reported the presence of bilateral polycystic kidneys. The diagnosis of Meckel Gruber syndrome was confirmed and the parents were counseled about the possibility of recurrence in future gestations.

DISCUSSION

Meckel -Gruber syndrome (also called Dysencephalia splanchnocystica) is an extremely rare association with an incidence of 1 in 13,250 to 1 in 140,000 live births.¹ It is an autosomal recessive disorder with the locus at chromosome 17p region2, bands1-4.² No particular gender predilection has been noted. Renal disorders are noted in 95-100% cases and may range from bilateral cystic dysplasia with grossly enlarged kidneys, ureter, bladder and urethral anomalies. Occipital encephalocele is reported in 60% to 80%. Post-axial polydactyly is present in 55% to 75%. Other limb

anomalies such as bowing and shortening may also be present.³ Facial abnormalities, ambiguous genitalia, cardiac septal defects and gastrointestinal anomalies like omphalocele also occur in varying combinations.³ The diagnosis of Meckel Gruber Syndrome relies on the constellation of systemic abnormalities detected on anomaly scan. The combination of cystic dysplastic kidneys, oligohydramnios, cranial and limb defects is mandatory for diagnosis. Maternal and amniotic fluid alpha fetoprotein may be elevated in case of associated encephalocele or anencephaly.⁴ A karyotype examination is required to exclude chromosomal aberrations. Meckel syndrome is a lethal disorder. Most infants are stillborn or die hours or days after birth. Occasionally they survive a few months with poor quality of life.⁵ If the diagnosis is made before viability, termination can be offered. When the family decides to continue the pregnancy, or if the diagnosis is made after viability, the standard obstetrical management is not altered. Parents should be counseled of the likely recurrence of Meckel-Gruber syndrome in subsequent gestations.

References

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