



## Case Report

# Meckel-Gruber syndrome: A case report

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## ABSTRACT

Meckel–Gruber syndrome (MGS) is a rare, lethal, autosomal recessive ciliopathy characterized classically by a triad of central nervous system malformations, postaxial polydactyly, and renal cystic dysplasia. The condition is associated with a poor prognosis, and early antenatal diagnosis plays a crucial role in parental counseling and pregnancy management. However, considerable phenotypic variability exists, and incomplete expression of the classical triad has been reported in a minority of cases. We report a case of MGS with an atypical presentation. A term male neonate was born to a 21-year-old mother from a non-consanguineous marriage. First- and early second-trimester antenatal ultrasonography scans were reported as normal. A third-trimester ultrasound revealed abnormalities suggestive of a congenital malformation. At birth, the neonate was noted to have an occipital encephalocele and postaxial polydactyly. Notably, renal involvement was absent, which is an uncommon finding in MGS. The diagnosis was established based on characteristic clinical features and imaging findings. This case highlights the phenotypic variability of MGS and emphasizes that absence of renal cystic dysplasia does not exclude the diagnosis. It also underlines the importance of detailed and serial antenatal imaging, particularly in pregnancies without early ultrasound abnormalities. Given the autosomal recessive inheritance pattern of MGS and a recurrence risk of 25% in subsequent pregnancies, early diagnosis is essential for appropriate genetic counseling and informed decision-making. Reporting such atypical presentations expands the existing knowledge of the clinical spectrum of MGS and reinforces the need for heightened clinical suspicion even when all components of the classical triad are not present.

**Keywords:** Congenital anomalies, Meckel–Gruber syndrome, Occipital encephalocele, Postaxial polydactyly, Renal cystic dysplasia,

## INTRODUCTION

Meckel-Gruber syndrome (MGS) is a ciliopathic disorder with a worldwide incidence ranging from 1 in 13,250 to 1 in 140,000 live births. It follows an autosomal recessive inheritance pattern, with a recurrence risk of 25% in subsequent pregnancies. The classical triad includes occipital encephalocele, postaxial polydactyly, and renal cystic dysplasia, though renal involvement may be absent in approximately 10-20% of cases.<sup>1-4</sup>

## CASE REPORT

A male neonate was born at term to a 21-year-old mother from a non-consanguineous marriage. First- and early second-trimester ultrasounds were normal. A third-trimester ultrasound at 7 months revealed hydrocephalus and occipital encephalocele. At birth, the neonate had an occipital encephalocele with cerebrospinal fluid (CSF) leak and postaxial polydactyly of both hands and feet [Figure 1].



**Figure 1:** Clinical photograph showing occipital encephalocele.

## INVESTIGATIONS

MRI brain revealed an occipital encephalocele with dysplastic brain parenchyma and an associated arachnoid cyst. Abdominal ultrasonography showed normal kidneys and liver.

## DISCUSSION

Although MGS can be diagnosed as early as 11-14 weeks of gestation, early diagnosis was missed in this case due to normal early ultrasounds and a lack of targeted anomaly scanning. The diagnosis was made based on two major criteria, which are sufficient according to established diagnostic standards.<sup>3-6</sup>

## CONCLUSION

This case highlights a rare presentation of MGS without renal cystic dysplasia. Early antenatal screening and genetic counseling are essential to reduce recurrence risk in future

pregnancies.

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