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Meckel-Gruber Syndrome
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Introduction

Meckel-Gruber syndrome is an autosomal recessive disease caused by primary ciliary dysfunction during early embryogenesis. It is characterized by the following triad: bilateral renal cystic dysplasia (100%), occipital encephalocele (90%), and postaxial polydactyly (83.3%). The prevalence ranges from 1:/1,300 - 140,000 births
30-year-old female, 5th pregnancy, with one preterm birth, no postnatal studies, and untreated pre-gestational diabetes, was referred to a Level III Hospital

A 38-week fetus in breech position, microcephaly, occipital encephalocele, ventriculomegaly, multicystic renal dysplasia due to absent amniotic fluid.

Finger counting or fetal face view could not be confirmed. The gastric chamber and bladder were not observed

Conclusion

Meckel syndrome is a rare, lethal polymalformative syndrome with autosomal recessive inheritance. Prenatal diagnosis is possible through suspected ultrasound and, in many cases, through fetal DNA testing. Early prenatal diagnosis of the condition is important, as it will allow for appropriate subsequent genetic and reproductive management

