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First trimester diagnosis of Meckel–Gruber syndrome

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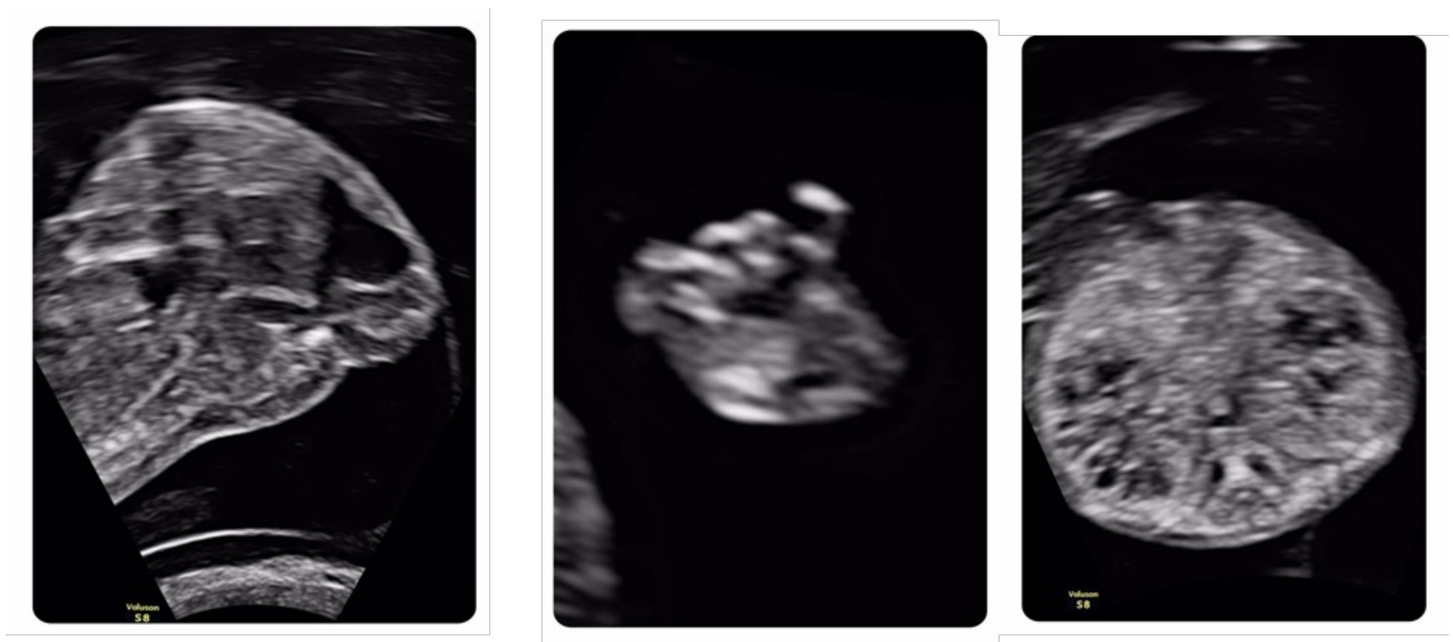
Introduction

Meckel–Gruber syndrome is a lethal autosomal recessive congenital anomaly syndrome caused by mutations in genes encoding proteins that are structural or functional components of the primary cilium. The typical structural anomalies are: occipital meningoencephalocele, bilateral polycystic kidney and postaxial polydactyly.

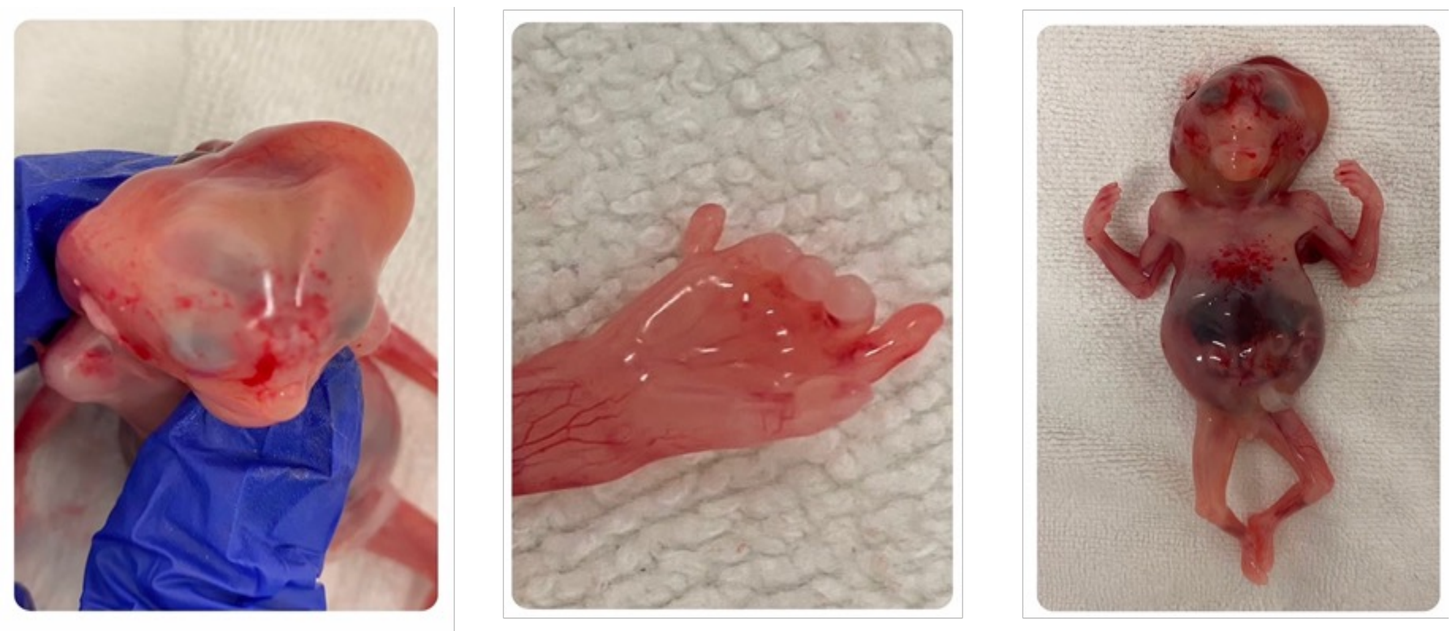
Case Report

A 29–year–old primigravida without risk factors and non-consanguineous marriage whose first routine ultrasonography scan at 13 weeks showed: occipital encephalocele, cystic dysplasia of both kidneys and bilateral postaxial polidactyly.



After discussing the prognosis, the patient opted for termination of pregnancy. An invasive test was offered, an informed consent was obtained for the procedure and a chorionic villus sampling was performed. The Quantitative fluorescence polymerase chain reaction (QF-PCR) and comparative genome hybridization (CGH) array were normal.

A clinical exome sequencing was performed and a variant of uncertain significance was found in the gen SHH c.1021G>A p.Ala341Thr. This is a missense variant that has been reported to be associated with cilium pathologies in an autosomal dominant inheritance.



A parental segregation study is being done to find out if it is a de novo mutation and the cause of the clinical features in the fetus.

The patient delivered at 14 weeks and a postmortem examination confirmed hernitation of the brain and postaxial polidactyly parental segregation study is being done to find out if it is a de novo mutation and the cause of the clinical features in the fetus.