A case of meckel gruber syndrome
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Objective
Mickel Gruber syndrome detected in early second trimester. Meckel-Gruber syndrome is a lethal, rare, autosomal recessive (genetic heterogeneity), developmental disorder caused by dysfunction of primary cilia during early embryogenesis.

Methods
A 24 year old, G3 P2 was referred to FMU with a diagnosis of posterior fossa cyst at 18 weeks. She had no family history of any congenital anomalies, except her cousin. She was married to her first cousin and was known case of thalassemia (both partners carriers). She was taking multivitamins and thyroxine only as had history of Thyroid disorder. Ultrasound scan confirmed a large occipital encephalocele 2x2x3.5 cm allowing some of the brain tissue and spinal fluid to bulge out. Very large polycystic kidneys, occupying the whole abdomen and pushing towards the chest. There’s limb deformity (club foot). Placenta very bulky. Heart deformity. The chest is very narrow. Both limbs look deformed with talipes and very difficult to identify. Possible diagnosis of Mickel Gruber syndrome was confirmed. Bad prognosis was explained to the couple who opted for medical termination of pregnancy which was carried out without any problem soon after. She was referred to the geneticist for further counselling and PGD was suggested for the future pregnancy.

Results
Classically defined by the classic triad of large polycystic kidneys, occipital encephalocele, and postaxial polydactyly. Affected children may also have abnormalities affecting the head and face, liver, lungs, and genitourinary tract. The leading cause of death in Meckel-Gruber syndrome is pulmonary hypoplasia which results from oligohydramnios caused by kidneys which have failed to develop properly. Associated abnormalities include oral clefting; genital anomalies; CNS malformations, including Dandy-Walker and Arnold-Chiari malformation; and liver fibrosis. Improvements in ultrasonography have enabled prenatal diagnosis as early as 10 weeks’ gestation. The prognosis is grim, with death occurring in utero or shortly after birth; prenatal diagnosis has led to therapeutic abortion of many affected fetuses. The mortality rate is 100% with most fetuses surviving only a few days to weeks.

Conclusion
Because the phenotypic overlap with trisomy 13 is considerable, the gene for Meckel-Gruber syndrome was postulated to be on chromosome 13. It is essential to exclude trisomy 13, which Meckel-Gruber syndrome mimics as Trisomy 13 carries a 1% recurrence risk, as opposed to the 25% for Meckel-Gruber syndrome. Linkage or mutation analysis is not yet available however recent identification of a gene that causes Meckel-Gruber syndrome and Joubert syndrome, PGD remains a possibility in future As consanguinity is an association proper family history is essential.