A case of Meckel-Gruber Syndrome

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Objective
Firstly to report a rare case of Meckel syndrome and the contribution of early ultrasound in prenatal diagnosis, secondly to insist on genetic heterogeneity and polymorphism of this syndrome and thirdly to evoke the place of the genetic study and its contribution to genetic counseling of couples at risk.

Methods
Meckel syndrome is a lethal syndrome associated with a rare autosomal recessive genetic disorder which was originally defined with occipital meningo-encephalocele, multicystic kidneys and polydactyly. However, the extreme variability of the clinical presentations found in the literature shows that the polymorphism of this syndrome is an essential feature. We report a rare case of Meckel Syndrome that was diagnosed prenatally by morphological ultrasound examination.

Results
22 year old woman, G3P0, with a history of a termination at 20 weeks because of prenatal diagnosis of meningo-encephalocele and a miscarriage in the first trimester was referred to our clinic at 17 weeks of gestation. Our findings were occipital myelomeningocele, multicystic kidneys bilateral and short and curved lower limbs and oligohydramnios. The mother opted for termination of pregnancy. The macroscopic examination revealed occipital meningocele, hypertelorism, microphthalmia, low-set ears and hurt lobulated, sexual ambiguity, polydactyly and a very distended abdomen. The dissection of the fetus conclude to occipital defect of 1x1. 5 cm with occipital meningocele without nerve tissue, large multicystic kidneys bilateral, ureters threadlike bilateral and tubular bladder. The section of the kidney showed no corticomedullary differentiation. The fetal karyotype was 46 XY, PS21. The histological examination showed the presence of spherical cysts sparing corticis cortex in the kidneys and whose size increases towards the medulla, which is deserted. The liver had a fibroadenomatosis with corbelled bile duct. The diagnosis of Meckel Gruber Syndrome was then retained.

Conclusion
Meckel syndrome is a rare syndrome caused by a recessive genetic disorder characterized by genetic heterogeneity. Early detection through ultrasound is critical. The requirement is to promote genetic research to locate almost all gene loci responsible, and have access to genetic counselling for all couples at risk.