

A case of Meckel-Gruber syndrome

Karoui A, Charfeddine M, Abid C, Abdelmoula B, Alioua I, Abouda H, Channoufi B
Maternity and Neonatology Center of Tunis, Department C, Tunis, Tunisia

Objective

Meckel-Gruber syndrome is a rare autosomal recessive lethal malformation characterized by typical manifestations of occipital encephalocele, bilateral polycystic kidneys and post axial polydactyly. The worldwide incidence varies from 1 in 13,250 to 1 in 140,000 live births. Highest incidence was reported in Gujarati Indians. We report a rare case of Meckel-Gruber syndrome from Tunisia and review of literature. Keywords: Encephalocele, Meckel-Gruber syndrome, polycystic kidney, polydactyly.

Methods

We report a rare case of lethal Meckel-Gruber syndrome in a male fetus with literature review.

Results

Patient B. A, 40 years old, hypertensive, G5P5, non-consanguineous marriage, carrier of a uterus, scar tissue, pregnant 23 SA admitted to our service for medical interruption of the pregnancy. The morphologic ultrasound made at 22 SA discovers a poly malformative syndrome associating a dolichocephaly with agenesis of the cerebellum, a bilateral nephromegaly with distended abdomen, anamnios with pulmonary hypoplasia. Caesarean section delivery: male fetus 1150 g Macroscopy: encephalocele, very distended abdomen, polydactyly compatible with Meckel syndrome. Evolution: death of the baby at H1 of life.

Conclusion

Meckel-Gruber syndrome is a rare and lethal poly malformative syndrome characterized by clinical polymorphism. The classic triad is not always constant. There is a risk of recurrence of 25%. Nowadays prenatal ultrasonographic signs still play a key role in the clinical diagnosis of MGS Genetic counseling and accurate isolation of the gene responsible for Meckel syndrome is a necessary step in antenatal diagnosis.