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Case Report

Meckel-Gruber syndrome: about a case identified during deliver

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ABSTRACT

Meckel-Gruber syndrome is an autosomal recessive disorder, usually lethal, most commonly characterised by the classic triad of polycystic kidneys, occipital encephalocele and polydactyly. Antenatal diagnosis can be made by ultrasound between 10 and 14 weeks of amenorrhoea. Recognition of this syndrome is important in order to establish the diagnosis and provide genetic counselling. Finally, well supervised termination of pregnancy should be the rule for lethal fetal malformations in order to relieve the psychological suffering of patients. However, in certain situations or working conditions, the diagnosis can be made late or even discovered during childbirth. We report a case of Meckel Gruber syndrome discovered at birth.

Keywords: Meckel-Gruber, Fetal malformations, Antenatal diagnosis, Genetic counselling

INTRODUCTION

Meckel-Gruber syndrome is an autosomal recessive disorder usually lethal characterized most commonly by polycystic kidneys, occipital encephalocele and polydactyly.[1] These findings can be associated with other cardiac, hepatic, splenic and facial abnormalities leading to a polymorphic clinic situation.[2] The prevalence of this disorder varies between regions. It is estimated at 1/140,000 live births in Great Britain and 1/3500 live births in North Africa.[3] Recognizing this syndrome is important not only for the management of the current pregnancy but also for the counseling of future pregnancies with an estimated recurrence rate of 25%. Antenatal screening by ultrasound is particularly important in couples with consanguinity, especially in African countries where genetic screening is not accessible to all.

We reported a case of Meckel Gruber syndrome which was diagnosed at birth and confirmed by pathological examination.

CASE REPORT

This was a 23-year-old patient, 4th gesture 3rd pare, with a background of a late abortion at 20 weeks of amenorrhea of a polymalformed. There was also a 1st degree of consanguinity with the partner. The patient was followed up with 4 antenatal consultations. She underwent an ultrasound scan at 28 weeks of amenorrhoea which concluded to a polymalformation syndrome without any further details. She was received in our department in the active phase of labour in pregnancy of 42 weeks of amenorrhoea. She gave birth vaginally to a female infant weighing 3500 grams, who was alive, non-viable and polymalformed with the following malformations: omphalocele, occipital encephalocele, polydactyly and cleft lip as shown in Figure 1.

In front of these manifestations, a fetopathological examination was required (with the parents' authorization) which found typical hepatorenal anomalies (Figure 2).

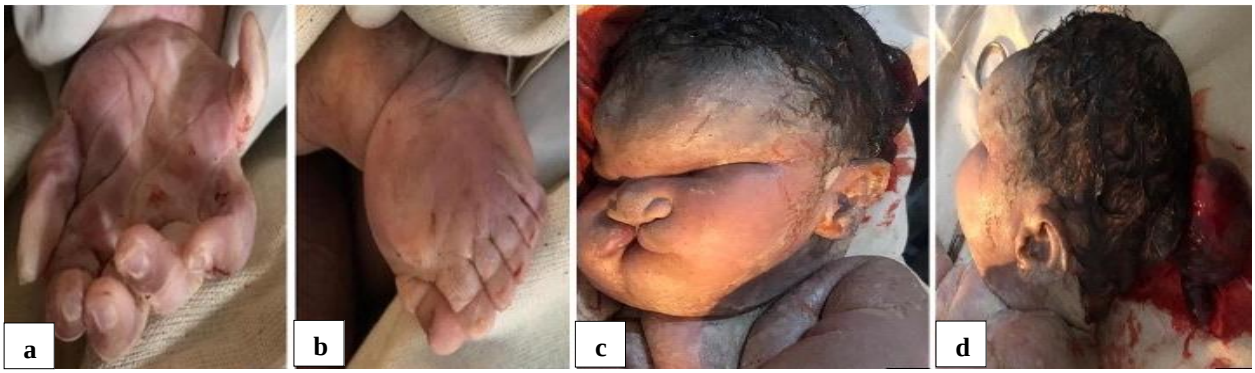

Figure 1: Fetal malformation (a) polydactyly of upper limb l, (b) polydactyly of lower limb, (c) cleft lip, and (d) occipital encephalocele.

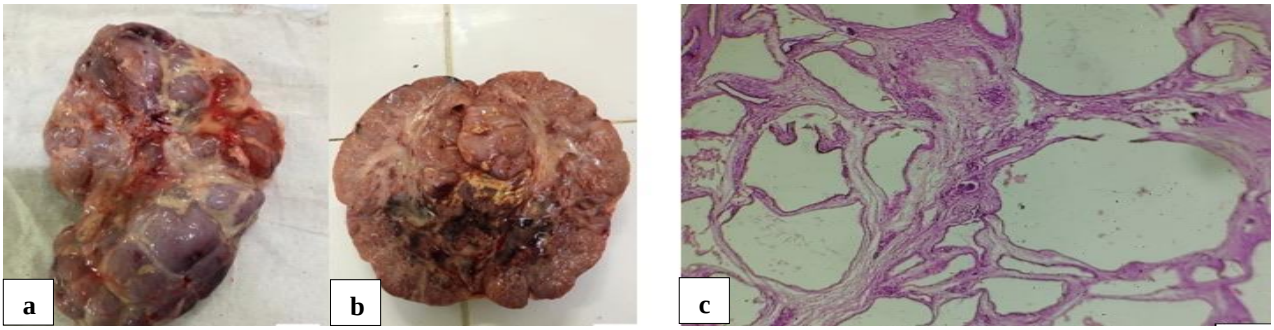

Figure 2: Histo-foetopathological examination, macroscopic aspect (a) surgical specimen with a bumped kidney surface, (b) cross-sectional appearance with several cystic cavities with myxoid content, and (c) histological aspect showing cystic cavities with serous content, lined with a simple cylindrical epithelium. Between these cystic formations, an inflammatory residual renal tissue is observed, with fibrosis.

DISCUSSION

First described in 1822 by the German anatomist Johann Friedrich Meckel, Meckel syndrome includes occipital cephalocele, microcephaly, cleft lip and palate, polycystic kidneys and polydactyly.[3] In 1934, George B. Gruber reported several familial cases suggesting a genetic origin of the disease.[4] Only in 1969, Opitz and Howe reviewed the clinical features of this disorder and suggested its autosomal recessive mode of transmission.[5] Worldwide, the prevalence is between 1 and 10.7 cases per 142,860 births.[3]

Early diagnosis can be made antenatally by ultrasound from 14 weeks of amenorrhea.[5,6] Ultrasound is the examination of preference for diagnosis, thus the interest of promoting screening ultrasound to identify pregnancies "at high risk" of fetal malformations and to guide them for diagnostic ultrasound.

Nevertheless, screening and diagnosis require different levels of expertise in different situations. This suggests that more attention should be given to antenatal diagnosis of malformations through the training of qualified medical personnel. In some situations or labor conditions, the diagnosis can be made late or even discovered during delivery, as it was in our case.

The differential diagnosis, once the diagnosis is suspected, will be made by the fetal karyotype mainly with trisomy 13.[7]

Meckel-Gruber syndrome is secondary to mutations in genes encoding proteins that are structural or functional components of the primary cilium. The disorders caused by mutations in ciliary genes are collectively called ciliopathies, and Meckel syndrome represents the most severe affection of this group of disorders. It is a complex syndrome with extreme genetic heterogeneity. To this day, mutations in 14 genes have been identified as causal for Meckel syndrome.[8]

Major congenital malformations cause psychic trauma that is lived painfully by the mothers.[9] This case report highlights the importance of early antenatal diagnosis of congenital diseases such as Meckel syndrome and the need to make genetic investigations accessible. This will facilitate doctors and parents in planning the subsequent management of the pregnancy. As Meckel syndrome is not compatible with life, medical termination should be offered to parents. But medical termination raises complex ethical, legal, social, and religious considerations. Indeed, Senegal's legislation on abortion is among the most restrictive in the world. According to the law on reproductive health, voluntary termination of pregnancy is

banned.[10] The medical code of ethics, in its article 35, provides for a single exception: therapeutic abortion can only be performed if this intervention is the only way to save the life of the mother. Therapeutic abortion is thus prohibited in cases of fetal malformation, or when the pregnancy threatens the mental or physical health of the mother, as long as her life is not seriously at risk. Further research on the psychological impact on mothers with life-incompatible polymalformed fetuses and the consequences of these pregnancies should be conducted in order to extend the law to these mothers.

CONCLUSION

Meckel-Gruber syndrome is a lethal autosomal recessive congenital anomaly. Antenatal diagnosis can be made in the first trimester by ultrasound. Diagnosis and genetic counseling are important for subsequent pregnancies. At last, carefully supervised termination of pregnancy should be the rule for lethal fetal malformations in order to relieve the psychological suffering of patients.

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