

Meckel Gruber Syndrome (About a Case and Reviews of the Literature)

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Abstract:- Meckel-Gruber syndrome, also known as Meckel syndrome or Gruber syndrome is an infrequent but lethal congenital malformation that combines encephalocele, cystic kidney dysplasia, and polydactyly. The variability clinical pictures found in the literature show that the polymorphism of this syndrome is an essential characteristic.

The current ultrasound examination, at the present time, is the best means of antenatal screening for this lethal poly malformation.

We report a case of Meckel syndrome discovered on ultrasound during pregnancy.

Keywords:- prenatal diagnosis; MeckelGruber syndrome; Meckel syndrome ; Gruber syndrome.

I. INTRODUCTION

Meckel-Gruber syndrome, also known as Meckel syndrome or Gruber syndrome is a severe ciliopathy.

It is a developmental disorder first described by Meckel in 1822, and later by Gruber in 1934 [1]. It is a combination of encephalocele, cystic kidney dysplasia, and polydactyly.

The variability clinical pictures found in the literature show that the polymorphism of this syndrome is an essential characteristic. [3-4].

The current ultrasound examination, at the present time, is the best means of antenatal screening for this lethal poly malformation [2]. The mortality rate of this syndrome is 100%, the main cause of death is pulmonary hypoplasia due to oligoamnios and liver disease.

We report a case of Meckel syndrome discovered on ultrasound during pregnancy, which is said to be at term.

II. CASE REPORT

Patient aged 24 years, primiparous primigravida, having as an antecedent a first degree consanguinity. Admitted for the first time in our training for CEP of abdominal-pelvic pain of the uterine contraction type on a pregnancy that is said to be at term in a patient in active labour.

The fetal ultrasound was performed to determine an evolving mono-fetal pregnancy with fetal malformations including: major hydrocephalus (Figure 1) + bilateral renal dysplasia (Figure 2) and posterior encephalocele associated with amniosis.



Figure 1 :show aencephalocel in a full-term foetus



Figure 2::show a bilateral kidney dysplasiain a full-term foetus

A vaginal delivery was made giving birth to a male baby, weight 2400g. Macroscopic examination shows: at the cephalic pole: retrognathism (Figure 3) and posterior encephalocele (Figure 4); at the abdomen: hepatosplenomegaly with ascites; at the limbs: polydactyly (Figure 5) on the 4 distal segments, clubfoot (Figure 6) and a curved aspect of the humerus (Figure 7); examination of the external genital organs objectively shows the presence of a micropenis.

The diagnosis of Meckel Grubersyndrome was retained .



Figure 3 :fetal retrognathism



Figure 4 :occipital encephalocele



Figure 5 :polydactyly with a curved appearance of the long bones of the humerus



Figure 6 :micropenis.

III. DISCUSSION

Meckel syndrome is a rare congenital poly malformative syndrome of autosomal recessive transmission, first described in 1822 in the German literature, caractérisé by a group of congenital malformations affecting in particular the central nervous system and the kidneys. It is generally lethal shortly after birth [2].

It affects 1 in 13,250 to 1,140,000 people worldwide. It is most common in Finland, where its prevalence at birth is 1 in 9,000 and the mutation frequency is 1% [5]. This high prevalence has been described with a very high inbreeding rate, as in the case of our patient who presents with first degree inbreeding. Three genes have been described: MKS1 on chromosome 17, MKS2 on 11, and MKS3 on 8.

Meckel syndrome is a monogenic disease characterized by a combination of renal cysts and other manifestations [4]: abnormalities in the development of the central nervous system (occipital encephalocele); polydactyly; dysplasia of the bile ducts and liver cysts.

This syndrome is generally defined by the triad: Occipital encephalocele, cystic kidney dysplasia and polydactyly. Polydactyly is most often post-axial (6th finger), but can sometimes be pré-axial (duplication of the thumb). A curvature of the long bones of the limbs is present in 1 in 6 cases. D'autres abnormalities may be present: cleft lip and palate, anophthalmos or microphthalmos, atresia of the limbs urètre, heart and genital malformations.

In the literature many authors report diagnostic criteria that have divided it into major criteria: polycystic renal dysplasia is a mandatory criterion for establishing the diagnosis associated with anamnios [6-7]; minor criteria: hepatic fibrosis; occipital encephalocele; polydactyly; other central nervous system malformations: Dandy-Walker malformation and malformation of Arnold Chiari.

The antenatal diagnosis may be suspected in the association of these abnormalities, as in the case of our patient who presented a bilateral multi-cystic dysplasia associated with an occipital encephalocele and anamnios.

MRI has no interest in the diagnosis and CEP of these abnormalities. Amniocentesis may reveal an elevated level of alpha-fetoprotein amniotic fluid due to encephalocele [8]. Karyotype remains the best way to confirm the diagnosis. If the pregnancy is carried to term, the newborn dies in the perinatal period.

Diagnostic difficulties may arise in the face of other polymalformative syndromes associating encephalocele, renal cystic dysplasia, polydactyly such as Carpenter-Hunter syndrome, and others but also generalized bone lesions.

Polydactyly is also found in Ellis von Creveld syndromes, short rib polydactyly, Moon-Bardet-Biedl, holoprosencephaly-polydactyly (pseudo-trisomy 13). A great help in diagnosis will be provided by the isolation of the Meckel syndrome gene.

Meckel syndrome is lethal with an average survival of less than 24 hours. However, Genuardi describes a case of Meckel syndrome that initially included polycystic kidneys, a Dandy-Walker, and postaxial polydactyly and survived for 43 months before dying of renal failure [10]. Genetic counseling aims to inform parents of an individual achieves that the risk of recurrence is 25% for subsequent pregnancies.

Unfortunately for our case, the patient was not followed, and a vaginal delivery was performed giving birth to a male infant who died a few hours after birth in a context of respiratory distress.

Genetic counseling was offered to the patient.

IV. CONCLUSION

In any fetus with two of the following three abnormalities: occipital encephalocele; polycystic large kidney; and postaxial polydactyly. Additional abnormalities may be noted. Because of the strong similarity to trisomy 13, which is much more common, it is essential to obtain a karyotype. Chorionic villi sampling or amniocentesis may be performed during the prenatal period. Karyotyping is particularly important because the risk of recurrence of trisomy 13 is very low, whereas the risk of recurrence of Meckel-Gruber syndrome is 25%.

In families with a previous child with Meckel-Gruber syndrome, prenatal diagnosis by chorionic villi sampling or amniocentesis is possible if the genetic defect in the previously affected individual has been identified or if typical ultrasound findings are present.

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