

Meckel-Gruber Syndrome : Sonographic Detection

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Abstract

Meckel–Gruber syndrome is a rare congenital foetal anomaly. We present a report of two cases where the diagnosis was made on antenatal ultrasound examination and confirmed later on when the pregnancies were terminated.

Key Words

Encephalocele, Polydactyly, Renal cysts

Introduction

Meckel–Gruber Syndrome is an autosomal recessive disease characterized by Occipital Encephalocele, Cystic Kidneys and Polydactyly. Other frequently associated anomalies include congenital hepatic fibrosis and hepatic cysts, micro–ophtalmia and abnormal genitalia (cryptorchidism). The disease is invariably fatal at birth due to pulmonary hypoplasia and renal failure.

CASE No. 1

A 29 years old woman in her third trimester of pregnancy and with previous bad obstetrical history was referred to the department of radio diagnosis for routine ultrasound examination. Sonographic examination was not performed earlier in this pregnancy.

Case No. 2

A 24 years old primigravida underwent routine sonographic examination in third trimester of pregnancy for the first time. She also had pregnancy induced hypertension.

On Ultrasound examination, following findings were observed in these patients :

1. Foetal skull vault showed deficient occipital bone with outpouching of meninges and brain tissue forming well defined encephaloceles in both the patients (Fig. 1).

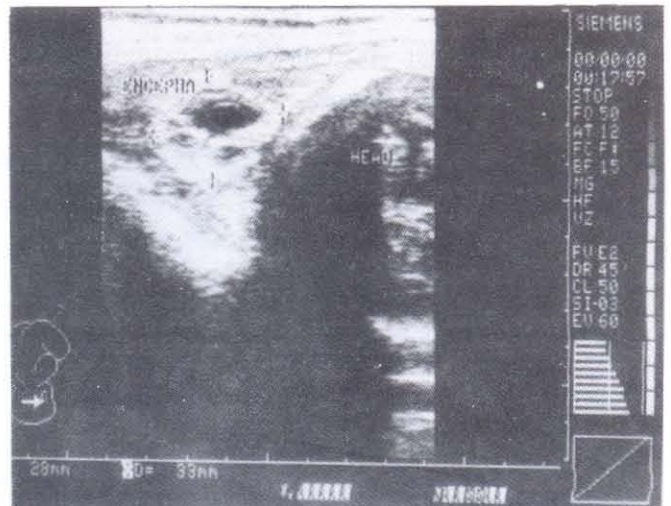


Figure 1. Foetal skull sonogram showing small occipital encephalocele.

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2. All the ventricles of foetal brain were dilated in both the patients (Fig. 2).

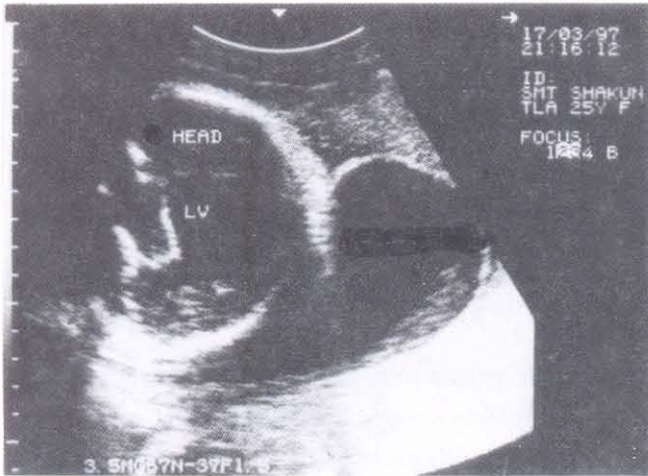


Figure 2. Foetal skull sonogram showing a large occipital encephalomeningocele and dilated lateral ventricles (LV)

3. Foetal abdomen revealed bilateral large hyperechoic kidneys with multiple small renal cysts in one foetus and multiple large renal cysts in other foetus (Fig. 3).

4. Polydactyly in upper and lower extremities was observed in both the foetuses.

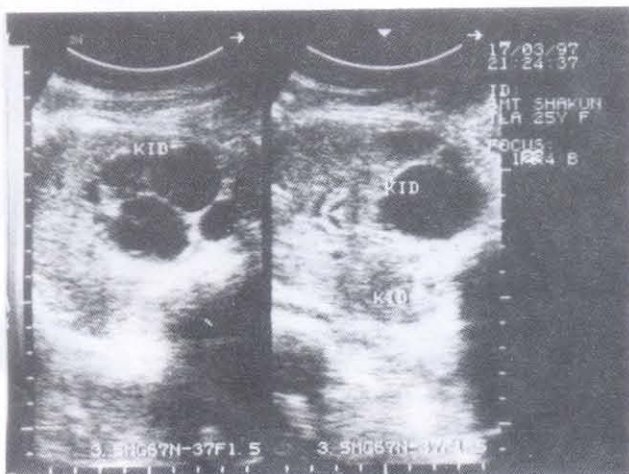


Figure 3. Bilateral large renal cysts in foetal kidneys (KID) on antenatal foetal abdominal ultrasonography.

On the basis of above mentioned findings, involving multiple organs of the foetuses, the diagnosis of Meckel-Gruber syndrome was made.

The patients were kept on follow up. One patient delivered full term foetus which survived for 12 hours after birth. In other case, the pregnancy was terminated as desired by the parents after the prognosis was explained. All the sonographic findings were confirmed on the delivery.

Discussion

Incidence of Meckel-Gruber syndrome is 1 : 140,000 to 1 : 13,250 (i), more common among Yemenite Jews. Risk of recurrence is 25% and carrier frequency is 1 : 56. History of affected siblings may be there. Parental consanguinity has been reported which supports evidence for autosomal recessive inheritance of this disorder. Most consistent abnormality in Meckel's syndrome is renal dysplasia. Renal cysts are frequently large and they probably originate from the collecting ducts. The affected kidneys vary in size from marked hypoplasia to large cystic masses, and usually are not functioning. The clinical and pathological characteristics of Meckel's syndrome were reviewed by Opitz and Howe (2) who suggested the eponym. The condition is recessively inherited and leads to death perinatally or in infancy. Most infants die at or shortly after birth. However, prolonged survival upto 28 months has been reported (3). In Finland, where there is a very high frequency of recessive diseases, the probable incidence of Meckel's syndrome was found to be 1 : 9000 births with equal sex ratio (4). The classical triad is polydactyly, occipital encephalocele, and cystic kidneys. It is associated with other CNS malformations, cranial rachischisis, eye anomalies, cleft palate, congenital heart disease, hypoplasia of adrenal gland, pseudohermaphroditism in males and other malformations. There are sometimes cysts in liver and pancreas. In a study of 38 siblings with



Meckel's syndrome by Fraser and Lytwyn (5), all had cystic dysplasia of kidneys, 63% had occipital meningocele, 55% had polydactyly, and 18 % had no reported brain malformation. Diagnosis of Meckel's syndrome is not considered valid in the absence of cystic renal dysplasia. All the 5 cases reported by Rohder and Labbe (6) had bilateral diffuse renal disease with changes resembling adult type polycystic kidneys leading to extensive renal enlargement and distension of abdomen.

Four cases were described by Jammal *et al* (7), all had polydactyly. Hydrocephalus and micropolygyria was also seen in these patients. In a review of 40 cases in Finland by Salonen (8), cystic dysplasia of kidneys and portal fibrosis was observed in all. Hepatic cysts were also present. Salonen reiterated that minimal diagnostic criteria for Meckel's syndrome include cystic dysplasia of kidneys with hepatic portal fibrosis and occipital encephalocele or some other CNS malformation. Because of posterior encephalocele, maternal and foetal alpha fetoprotein levels may be elevated. Differential

diagnosis include Smith-Lemili-Optiz syndrome, Hydrolathus syndrome, Trisomy 13, and Ivemark syndrome.

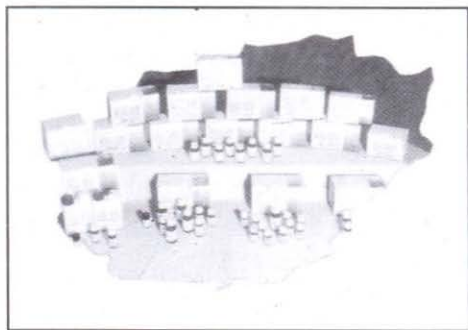
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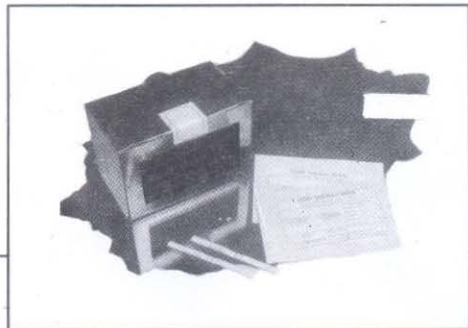
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