Meckel-Gruber Syndrome

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Abstract

Meckel-Gruber syndrome is a rare autosomal recessive disorder. It is associated with a spectrum of clinical manifestations like occipital encephalocele, microcephaly, polydactyly and congenital nephrosis. This condition maybe diagnosed in utero by determining L-fetoprotein levels and Ultrasound measurement of the biparietal diameter. Management requires multidisciplinary team approach. Genetic counselling is the mainstay of management. We are reporting this case for the first time.

Keywords: Autosomal recessive; encephalocele; L-fetoprotein.

Introduction

Meckel-Gruber syndrome is a rare autosomal recessive condition that is characterized by an occipital encephalocele, polydactyly, polycystic kidney, microcephaly and other congenital anomalies like cleft lip or palate and abnormal genitalia.1 As inheritance is autosomal recessive, it is very lethal. Clinically, this syndrome presents from birth and both sexes are affected equally. This condition maybe diagnosed during intrauterine life by determining L-fetoprotein levels and ultrasound measurement of biparietal diameter. In most of the mothers, there is an evidence of polyhydraminios. The first case of Meckel-Gruber syndrome was reported in 1822.2 Gruber reported another case in 1934.3 Since then a number of cases of this syndrome have been reported. This is the first case of Meckel-Gruber syndrome being reported in Nepal.

Case report

A 5-day old male neonate was brought by his mother with a history of poor sucking, jaundice, difficulty in breathing and cyanotic attacks off and on for 3 days.

The child is the 5th in the family, and has a history of full-term normal vaginal hospital delivery. His mother was on regular antenatal check-up. His birth history was uneventful. USG examination during the third trimester gives the evidence of polyhydraminios. The child borne from non-consanguinous parents. The first two children died. The first child was a girl having occipital encephalocele and died at the age of one year. The second child was a boy with occipital encephalocele and died two hours after birth. The third child, 6 years old girl, polydactyly in the left hand without encephalocele is crippled and surviving. The fourth child of 2 years, a girl, is perfectly all right.

Fig. 1: Showing occipital encephalocele.

Fig. 2: Showing polydactyly in all limbs.

Anthropometric measurement showed that his weight was 3 kg, length - 50 cm OFC 31.6 cm. Craniofacial examination showed that he was having ocipital encephalocele with diastasis of sagital suture and slightly bulged fontanelle. Examination of the chest showed that he was tachypnoeic with intercostal and subcostal recession. Cardiovascular examination showed that he was having systolic murmer. On abdominal examination bilateral palpable kidneys were found.

External genitalia was normal. Examination of the limbs revealed that he was having polydactyly in all the four limbs. No abnormalities were detected on dermatographic examination.

Investigation of the chest X-ray showed enlarged cardiac silhouette skull. Ultrasonogram revealed mild ventricular dilatation.

Discussion

Though no evidence of consanguinity of the marriage was found in the patient's parents, the history of other siblings of both sexes suffering from similar types of manifestations suggests possible diagnosis of autosomal recessive pattern of inheritance.

The pedigree chart showed that four out of five siblings were affected.

To add to this, the occipital encephalocele, microcephaly, polydactyly and renal anomaly present in this patient was a favourable diagnostic clue in coming to a diagnosis of Meckel-Gruber syndrome. But there were no defects in mouth (like cleft lip or palate) and also in genitalia. In our patient, besides the typical presentation there were atypical findings like systolic murmur and mild cranial ventricular dilatation on USG examination. So, it showed that Meckel-Gruber syndrome is also associated with rare presentation of hydrocephalus and congenital heart diseases4; more than 50 cases of this severe disorder were reported by Meckel & Hsia.5,6

Together with history and clinical presentation with subsequent chest X-ray and Ultrasonogram (USG) investigation, a diagnosis of Meckel-Gruber syndrome is a possibility.

Multidisciplinary team intervention is required to overcome the different manifestations. In a specialized centre, treatment for encephalecele and renal anomaly can be anticipated. Genetic counselling is an important aspect for the management. In this case, the parents were given genetic counselling. The patient was treated with antibiotics, oxygen, phototherapy, parentral fluid infusion and nasogastric feeding. Parents left the hospital with baby against medical advise.

Conclusion

Though Meckel-Gruber syndrome is a rare condition and carries a bad prognosis, still it has a great academic importance. Early diagnosis and proper management can prevent death at an early age. Parents should be given proper advise and genetic counselling for future management of the infants.

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