CASE REPORT

Prenatal Diagnosis of Meckel-Gruber Syndrome with Dandy Walker Malformation

Madhu Gupta*, Anil Mehta, Renu Gupta, Rajni Gupta, Sangeeta Singh**

Abstract

We report a 38 week old female fetus affected by Meckel-Gruber syndrome. Polycystic dysplastic kidneys, oligohydroamnios, symmetrical IUGR, hydrocephalus with Dandy-Walker cyst was observed on ultrasonographic (USG) examination. Small occipital encephalocele with polydactly was noted after delivery of child.

Key Words

Meckel-Gruber syndrome, Dandy-Walker malformation

Introduction

Meckel Gruber Syndrome (MGS) is a rare and lethal autosomal recessive disorder characterized by occipltal encephalocele, bilateral polycystic dysplastic kidneys and post-axial polydactly (1,2).

Although numerous abnormalities associated with MGS were previously reported in the literature, Dandy-Walker malformation (3-5), microcephaly, intrauterine growth retardation (IUGR), single umbilical artery, cardiovascular defects, cleft palate (6,7) several genital abnormalities (4,6,7), oligohydramnios (3,7) are the most well known. Hepatic periportal fibrosis (5) and hydrocephalus (8) were also noted in some cases. The incidence of this rare syndrome has been estimated as 1 in 50,000 (9). We report a prenatally diagnosed case of Meckel-Gruber syndrome with Dandy-Walker malformation.

Case Report

A 24 year old full term pregnant woman was admitted in the Department of Obstetrics & Gynaecology at Govt. Hospital, Gandhinagar, Jammu with a bad obstetrical history. The first female child died 4 days after birth. She had an abortion in the first trimester which was followed by another first trimester abortion. She reported to the hospital for antenatal checkup for the first time at 38 weeks. Clinical examination of patient revealed breech presentation with disparity between the gestational age and the symphysis fundal height. The fundal height was 4cms less than the expected. There was no clinical history of consanguineous marriage, exposure to x-ray or medication. Patient's blood group was AB+ve with haemoglobin 9gm%. Ultrasonography of the patient revealed bilateral enlarged polycystic kidneys and dilated lateral and third ventricles. Posterior fossa cyst with gross aplasia of cerebellum suggestive of Dandy-Walker cyst was also noted. No definite liquor pocket was seen which suggested severe oligohydramnios. Features of symmetrical IUGR were also observed. Patient was induced and delivered vaginally without any intranatal or postnatal complications. On examination, the fetus revealed a moderate hydrocephaly with a small occipital encephalocele and post-axial polydactly of the left hand. On the abdominal examination of the fetus large masses were palpable bimanually in both lumbar regions. Genital examination revealed here female sex. Left foot showed congenital talipes equinovarus.

From the Departments of *Radiology, Gynaecology and **Anesthesia, Government Hospital, Gandhi Nagar, Jammu (J&K).
Correspondence to: Dr. Anil Mehta, Consultant Gynae & Obstetrics, 681, B/A Gandhi Nagar, Jammu (J&K).

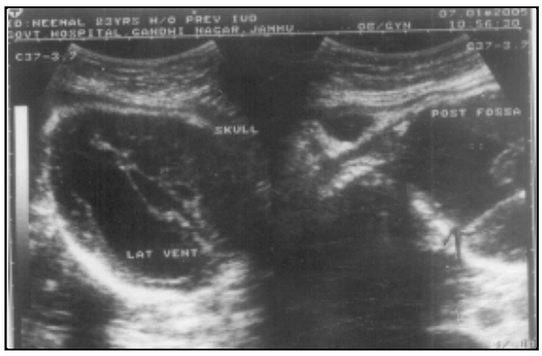

Fig. 1 USG revealing hydrocephalus with posterior fossa cyst & cerebellar aplasia

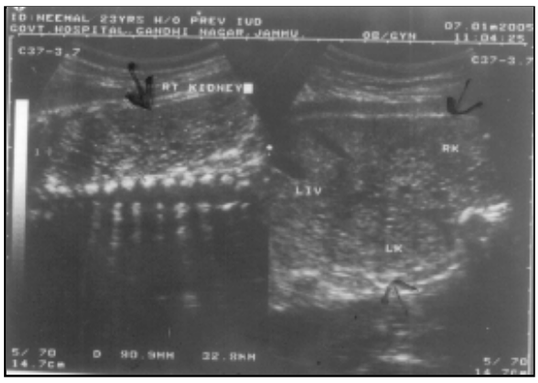

Fig. 2 Enlarged polycystic kidneys demonstrated by fetal sonography

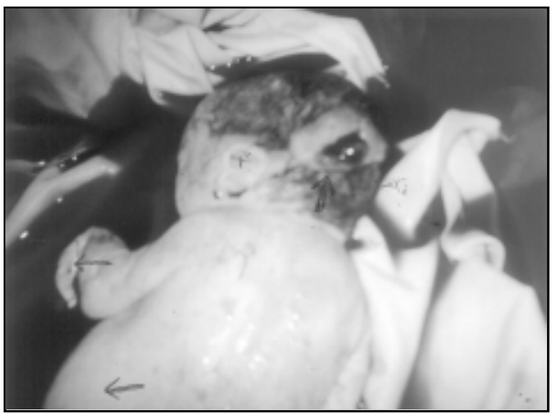

Fig. 3 External appearance of fetus with occipital encephalocele, post-axial polydactly and fullness of flanks

Discussion

Meckel-Gruber Syndrome is characterized by cystic kidneys, occiptal encephalocele and post axial polydactly. 2 of the 3 major anomalies are sufficient for the definitive diagnosis. It was reported that 57% of the cases had 3 cardinal findings, but 16% had only polycystic kidney and polydactly. The remainder exhibit other variations. Farag et al (10) reported 5 Bedouin siblings with MGS lacking polydactly; this could be explained by the phenotypic variability of MGS pleiotropic gene (10).

The spectrum of the phenotype in the MGS is very wide, encompassing various combinations of some quite common anomalies. Some cases may even be evaluated as a different syndrome (11). In the present case, the fourth pregnancy, from a non consanguineous family the prenatal diagnosis was made sonographically by the detection of polycystic kidneys and CNS malformation at 38 weeks' gestation. In view of these findings, the fetus was accepted as MGS. In association with the above findings, the fetus also showed cystic dilation of fourth ventricle occupying the posterior fossa with gross aplasia of cerebellum suggestive of Dandy-Walker malformation. CNS malformations of MGS are variable and show a broad spectrum ranging from occipital encephalocele, Chiari malformation, hydrocephalus, polymicrogyria, arhiencephaly, holoprosencephaly, agenesis of corpus callosum and anencephaly (8).

MGS is a lethal syndrome, generally results in utero or neonatal death within a few hours of life; thus, prenatal diagnosis is very important. The condition can be diagnosed sonographically in the first and second trimester (12,13). In our case the patient reported late in the third trimester and sonography was performed at 38 weeks.

Conclusion

A good family history, accurate prenatal diagnosis with USG and measurement of maternal or aminotic fluid alpha-fetoprotein levels in the second trimester are essential in the pregnancies complicated by MGS, for pregnancy management and counseling for abortion and for evaluation of recurrence risk in the future.

References

1. Schmidt W,von Host T, Shroeder T, Kubli F. Prenatal diagnosis of Meckel-Gruber syndrome by ultrasound. Z Geburtshilfe Perinatal 1981; 185: 67-71.
2. Vernekar JA, Mishra GK, Pinto RG, Bhandari M, Mishra M. Antenatal ultrasonic diagnosis of Meckel-Gruber Syndrome (a case report with review of literature). Australas Radiol 1991; 35: 186-88.
3. Neyberg DA, Hallesy D, Mahony BS, Hirsch JH, Luthy DA, Hickok D. Meckel-Gruber Syndrome, Importance of prenatal diagnosis. J Ultrasound Med 1990; 9: 691-96.
4. Balcy S, Onal B, Ercal MD, Beksac S, Erzen C, Akban O. Meckel-Gruber Syndrome: A case diagnosed in utero. Turk J Pediatr 1992; 34: 179-85.
5. Cinncinati P, Neri ME, Valentini A. Dandy-Walker anomaly in Meckel-Gruber Syndrome. Clin Dysmorphol 2000; 9: 35-38.
6. Jones KL. Smith's Recognizable Patterns of Human Malformation (5th ed). In: Jones KL (ed). Philadelphia: WB Saunders. 1997; pp. 184-85.
7. Su SL, Liu CM, Lee JN. Prenatal diagnosis of Meckel-Gruber Syndrome case reports. Gao Xiong Yi Xue Ke Xue Za Zhi 1995; 11: 127-32.
8. Ahdab-Barmada M, Classen D. A distinctive triad of malformations of the central nervous system in the Meckel-Gruber Syndrome. J Neuropathol Exp Neurol 1990; 49: 610-21.
9. Schmidt W, Kubli F. Early diagnosis of severe cogenital malformations by ultrasonography. J Perinat Med 1982; 10: 233-41.
10. Farag TI, Usha R, Uma R, Mady SA, al-Nagdy K, el-Badramany. Phenotypic variabiality in Meckel-Gruber Syndrome. Clin Genet 1990; 38: 176-79.
11. Yapar EG, Ekici E, Dooan M, Gokmen O. Meckel-Gruber syndrome concomitant with Dandy-Walker malformation: prenatal sonographic diagnosis in two cases. Clin Dysmorphol 1996; 5: 357-62.
12. Van Zalen-Sprock RM, van Vugt JM, van Geijn HP. First trimester sonographic detection of neurodevelopmental abnormalities in some single gene disorder. Prenat Diagn 1996; 16: 199-202.
13. Flessa A, Rempen A, Schumausser B, Marx A. Meckel-Gruber Syndrome. Z Geburtshilfe Neonatol 1996; 2000: 66-68.