Prenatal Diagnosis of Meckel-Gruber Syndrome with Dandy Walker Malformation

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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 polydactyly 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 occipital encephalocele, bilateral polycystic dysplastic kidneys and post-axial polydactyly (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 symphys 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 A B+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 polydactyly 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.

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Discussion

Meckel-Gruber Syndrome is characterized by cystic kidneys, occipital encephalocele and post axial polydactyly. 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 polydactyly. The remainder exhibit other variations. Farag et al (10) reported 5 Bedouin siblings with MGS lacking polydactyly; 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


