INTRODUCTION
Meckel-Gruber Syndrome (MKS) is an autosomal recessive disorder, characterized by a combination of central nervous system malformation (occipital encephalocele), post-axial polydactyly, and enlarged polycystic kidney dysplasia. With a recurrence risk of 25% this lethal syndrome can be detected in early screening by ultrasound. However, to the authors’ knowledge, association of MKS with unilateral renal agenesis was reported only once until now. Here, we describe a case of 26-year primigravida with 23 weeks pregnancy whose detailed sonographical examination of the fetus revealed large encephalocele through the posterior fontanelle microcephaly, anhydramnios, unilateral left enlarged polycystic kidney and right sided renal agenesis. The pregnancy was willfully terminated on medical grounds. Risk for subsequent pregnancies was explained to the parents.

CASE REPORT
A 26-year primigravid patient attended the clinic for the first time during her pregnancy for an obstetric scan due to the oligohydramnios. Her gestational age by last menstrual period was 23 weeks. There was history of first-degree consanguineous marriage. The father’s mother had a stillbirth with fetal anomalies, but the mother’s family history was non-contributory. She was not on any teratogenic drugs. The detailed sonographical examination of the fetus revealed large encephalocele through the posterior fontanelle (Figure 1A), microcephaly, anhydramnios, unilateral enlarged polycystic kidney (left side: 31 x 30 x 40 mm), but the other side had renal agenesis (Figure 1B). The fetal extremities were not appreciated due to the anhydramnios. Fetal biometric measurements except head circumference were consistent with 20 weeks of gestation.

After extensive clinical and genetic non-directive counselling, the parents gave informed approval or termination of pregnancy. A chromosome analysis was performed (fibroblasts skin culture) to rule out chromosome abnormalities such as trisomy 13, and showed normal fetal karyotype (46, XY). The parents of the fetus refused an autopsy. They did not want to undergo DNA analysis of fetus for high cost. On the external physical examination, all the four limbs showed the presence of post-axial polydactyly, the extra fingers and toes being on the same side of the extremities. Based on the above features, antenatal diagnosis of MKS associated with unilateral renal agenesis was made.

Information was given to family about this syndrome; the risk of relapse was communicated 25% and also early (between 11th and 14th gestational weeks) sonographical examination was advised for next pregnancies.

DISCUSSION
Meckel-Gruber Syndrome (MKS) is a lethal, rare and an autosomal recessive disorder. The worldwide incidence
Meckel-Gruber syndrome and renal agenesis

of MKS varies from 1 in 13,250 to 1 in 140,000 live births. It is characterized by the triad of occipital encephalocele, large polycystic kidneys, and postaxial polydactyly. Associated abnormalities include oral clefting; genital anomalies; central nervous system malformations, including Dandy-Walker and Arnold-Chiari malformation; and liver fibrosis. Pulmonary hypoplasia is the leading cause of death.1,6,7 Improvements in ultrasonography have enabled prenatal diagnosis as early as 10 weeks' gestation. The diagnosis of MKS is possible in most fetuses at early gestation, provided that a careful systemic survey is included routinely as part of first trimester scan. Diagnosis is really easier at this time, as in the presence of oligohydramnios in the second trimester could easily cause encephalocele and polydactyly to be missed, while in 11-14 weeks of pregnancy the diagnosis is much facilitated as actually the amount of amniotic fluid is not affected by the renal problem.7,8

Failure of mesodermal induction has been suggested to cause Meckel-Gruber syndrome. The induction cascades of early morphogenesis involve numerous growth factors, homeobox genes, and paired domain genes. The male-to-female ratio is almost equal, which is consistent with autosomal recessive inheritance.6

Oligohydramnios that results from dysplastic kidneys leads to fetal pulmonary hypoplasia. Because the prognosis is grim, with death occurring in utero or shortly after birth, prenatal diagnosis has led to therapeutic termination of many affected fetuses. The mortality rate is 100% with most die before or just after birth, but those who survive longer might have less severe abnormalities. Autopsy provides valuable information that aids in diagnosis and genetic counselling for future pregnancies.5,6

The diagnosis of MKS should be made prenatally during routine first trimester ultrasonographic screening because of the mortality rate is 100%. It should be noted that unilateral renal agenesis may be seen in MKS.

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REFERENCES