July 2004

First trimester diagnosis of meckel gruber syndrome in pregnancy

S Munim
Aga Khan University, shama.munim@aku.edu

S Nadeem
Aga Khan University

R Sikandar
Aga Khan University, raheel.sikandar@aku.edu

First Trimester Diagnosis of Meckel Gruber Syndrome in Pregnancy

S. Munim, S. Nadeem, R. Sikandar
Department of Obstetrics and Glynaecology, Aga Khan University Hospital, Karachi.

Meckel Gruber syndrome was originally described by Meckel in 1822, later by Gruber and more recently by Opitz and Howe.[1] It is a lethal autosomal recessive disorder characterized by the triad of encephalocele, polycystic kidneys and polydactyly. Prenatal ultrasonographic diagnosis of this condition has been reported extensively during the second and third trimeter. In the low risk population the estimated prevalence of the condition is about 1 in 20,000 pregnancies.[2] The disorder is more frequently encountered in the children of consanguineously married couples. The prevalence of this disorder in Pakistan is expected to be higher as the recent Pakistan Demographic and Health. Survey (DHS) shows that almost two-thirds of marriages in Pakistan are consanguineous.[3]

A case of Meckel Gruber Syndrome diagnosed by ultrasound examination is presented.

Case Report

A 35 years old gravida 5, para 4, was referred at 11 weeks gestation for ultrasound at our fetal medicine unit. The indication for the ultrasound was her previous history of a baby at 28 weeks with microcephaly, polydactyly and polycystic kidney.

The ultrasound examination performed at 11 weeks 6 days revealed a single fetus corresponding to dates. The Crown Rump Length (CRL) was 55.3 mm. In addition to the anomaly survey revealed encephalocoele and hyperechogenic kidneys. Bladder and stomach were not visible. Hands and feet were visible but polydactyly was not appreciated. The nuchal translucency measurement at this stage was found to be 5.2 mm. In view of above findings the provisional diagnosis of Meckel Gruber syndrome was made. She was counseled for an invasive test, which she declined and instead opted for a repeat ultrasound scan in two weeks.

A repeat scan was performed at 15 weeks, which showed fetus corresponding to 15 weeks of gestation. The amniotic fluid appeared normal at this stage and the placenta was posterior high. Encephalocele and enlarged echogenic kidneys were reconfirmed. The parents were counseled of the likely diagnosis of Meckel Gruber syndrome. Following counseling the parents opted for termination of pregnancy.

Extra-amniotic termination-of-pregnancy using Prostaglandin F_2 alpha was carried out. A fetus weighing 65 grams baby, with encephalocele, polydactyly in both hands and feet with short extremities was born (Figures 2 and 3). The diagnosis of Meckel Gruber syndrome was confirmed clinically. The couple refused for an autopsy.

Figure 1. Fetus with encephaloceole and increased nuchal translucency.

Figure 2. Fetus showing an encephaloceole.

Discussion

An association of Meckel Gruber syndrome with Rokitansky syndrome and short limb dwarfism has been reported in literature.[4] Other inconstant features include fascial cleft, microcephaly, cerebellar and cerebral hypoplasia, hydrocephalus, sloping forehead, congenital heart disease and pulmonary hypoplasia. Association genital anomalies in males are hypoplastic penis, cryptorchidism, Mullerian duct remnants and epididymal cysts, while septate vagina and hypoplastic or bicornuate uterus may be the associated anomalies in females.[5] However, Meckel Gruber syndrome may demonstrate variation in phenotypic expression when some malformations are different from those traditionally accepted and cases may be evaluated as a different syndrome.

Figure 3. Fetus showing polydactyly.

In this case the patient had a previous history of delivering a fetus with microcephaly, polydactyly and polycystic kidneys. Similar cases have been reported in the literature.[6] The earliest prenatal diagnosis of Meckel Gruber syndrome has been reported at 10 weeks by means of embryoscopy in a case at risk for this condition. The invasiveness of thisprocedure, however, precludes its use as a screening test or even in high risk pregnancies because a previously affected fetus.[2]

We were able to make the diagnosis of Meckel Gruber syndrome at 11 weeks and 6 days. As it is possible to diagnose most cases of Meckel Gruber syndrome at this early gestation, provided that a careful systemic survey is included routinely as part of 11 to 14 weeks scan. It is in fact easier at this stage, as in the presence of oligohydramnios in the second trimester could easily cause encephalocele and polydactyly to be missed. Whereas, early in pregnancy the diagnosis is greatly facilitated by the fact that the amount of amniotic fluid is not affected by the renal anomaly. Association of increased nuchal translucency has been reported with a number of genetic syndromes, as was in this case.[7]

In conclusion, nuchal translucency measurement and first trimester mini-anomaly scan can prove to be an effective screening tool for Meckel Gruber syndrome ensuring early diagnosis and timely termination of pregnancy.

References

1. Balci S, Onol B, Ercal MD, et al. Meckel Gruber syndrome: a case diagnosed in utero. Turk J Pediatr 1992;34:179-85.
2. Sepulveda W, Sebire NJ, Souka A, et al. Diagnosis of the Meckel-Gruber syndrome at eleven to fourteen weeks' gestation. Am J Obstet Gynecol 1997;176:316-19.

3. Hussain R. Community perceptions of reasons for preference for consanguineous marriages in Pakistan. J Biosoc Sci 1999;31:449-61.
4. Agapitos E, Christodoulou C. Meckel-Gruber syndrome associated with Rokitansky-Kuster-Hauser syndrome. J Clin Ultrasound 1995;23:452-55.
5. Malguria N, Merchant SA, Kiran KV, et al. Meckel-Gruber syndrome associated with short limbed dwarfism. J Postgrad Med 1996;42:55-6.
6. Mazneukova V, Kamenov E, Dimitrova L. Ultrasound diagnosis of Meckel-Gruber syndrome at 13 weeks of gestation in families at risk - a case report and literature review. Akush Ginekol (Sofiia) 2002;41(5):42-5.
7. Souka AP, Krampl E, Bakalis S, et al. Outcome of pregnancy in chromosomally normal fetuses with increased nuchal translucency in the first trimester. Ultrasound Obstet Gynecol 2001;18:9-17.