Abstract

Meckel-Gruber Syndrome (MGS) is a lethal autosomal recessive disorder characterized by anomalies of the central nervous system resulting in mental retardation, cystic dysplasia of the kidneys, and malformations of the hands and feet. In this report, we present a case of MGS, whose ultrasonographic findings containing the typical triad of the disease (encephalocele, polycystic kidneys, and polydactyly). 33 year old, Gravida 2 Parite 1, patient admitted to our clinic first time. Typical ultrasonographic findings including occipital encephalocele, bilateral cystic kidneys, oligohydramnios and polydactyly were observed. The family did not approve the genetic or pathologic investigation after the termination of the pregnancy. Therefore the diagnosis of MGS was based on prenatal and postnatal features. Most infants with MGS are stillborn or die within few hours or days after birth. It is important to remember that MGS is an autosomal recessive disorder that can repeat itself 25% between siblings, it would be prudent to give this information to families aswell.
Introduction

Meckel-Gruber syndrome (MGS) is a lethal autosomal recessive disorder characterized by anomalies of the central nervous system resulting in mental retardation, cystic dysplasia of the kidneys, and malformations of the hands and feet [1,2]. Also posterior encephalocele, hepatic ductal dysplasia, cleft palate, and postaxial polydactyly observed [3]. The worldwide incidence varies from 1 in 1.300 to 1 in 140,000 live births. The highest incidence was seen in the Gujarati Indians (1:1,300), and then in Finlands (1:9,000) [4]. In this report, we present a case of MGS, whose ultrasonographic findings containing the typical triad of the disease (encephalocele, polycystic kidneys, and polydactyly).

Case-report

33 year old, Gravida 2 Parite 1, patient admitted to our clinic first time. Typical ultrasonographic findings including occipital encephalocele, bilateral cystic kidneys, oligohydramnios and polydactyly were observed. These features were suggestive of the diagnosis of MGS. The family approved the termination of the pregnancy. The fetus had an occipital encephalocele (Fig. 1), postaxial polydactyly (Fig. 2), protuberant abdomen and palpable kidneys (Fig. 3). The family did not approve the genetic or pathologic investigation. Therefore the diagnosis of MGS was based on prenatal and postnatal features.

Figure 1: Occipital encephalocele

Figure 2: postaxial polydactyly

Figure 3: Protuberant abdomen and palpable kidneys

Discussion

The diagnosis of MGS can be made prenatally during routine ultrasonographic screening for fetal chromosomal abnormalities at 11 to 14 weeks of gestation [5,6]. MGS is characterized by typical sonographic findings, including occipital encephalocele, postaxial polydactyly, and cystic kidneys [3]. To ascertain the diagnosis, two of the three major abnormalities should be observed. The most specific abnormality is encephalocele, whereas the most seen abnormality is the polycystic kidneys [7].

MGS is associated with defects in the MKS1 gene (located at 17q23) and MKS2 gene (located at 11q13) [8,9].
MGS is a lethal disorder. Most infants are stillborn or die within few hours or days after birth [10]. Therefore prenatal diagnosis is crucial. Thorough ultrasonographic examination is vital especially between 11-14 weeks of pregnancy. Also it is important to remember that MGS is an autosomal recessive disorder that can repeat itself 25% between siblings, it would be prudent to emphasize this information to families as well.

As a conclusion, MGS is a fatal disorder. The parents should be informed about prognosis of the fetus and the outcome. Information about the 25% recurrence risk for the next pregnancies should be given.

References