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A Rare Case of Meckel–Gruber Syndrome

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Abstract: ***Meckel syndrome is a lethal, ciliopathic, genetic disorder, characterized by triad of renal cystic dysplasia, central nervous system malformations (occipital encephalocele) and polydactyly (post axial). Pulmonary hypoplasia due to oligohydramnios is also seen. It is rare disorder prevalence is <1/million and incidence is 1 in 13,250-1,40,000 live births. Recently we saw a case of Meckels syndrome (MS) baby delivered in our Unit. In this case, antenatal ultrasonagraphy revealed abnormalities indicative of minimal hydrocephalous, occipital encephalocele, hypoplastic limbs and ascites. Face was not clearly visualized. Upon delivery, baby showed hydrocephalous, shortened limbs with polydactyly, facial deformities supporting MS.***

Keywords: Occipital encephalocele, polydactyly, renal cystic dysplasia

1. Introduction

Meckel syndrome (MS, also known as Meckel–Gruber Syndrome, Gruber Syndrome, Dysencephalia Splanchnocystica) is a rare, lethal, genetic disorder. The first report on MS was published by Johann Friedrich Meckel in 1822. In 1934, G.B. Gruber published reports on individuals with MS and named the disorder dysencephalia splanchnocystica. This is characterized by the triad of brain malformation (mainly occipital encephalocele), large polycystic kidneys and polydactyly [1]. In addition other multiple congenital abnormalities such as cleft lip/palate, cardiac, genital anomalies, central nervous system malformations, liver fibrosis, and bone dysplasia.

Prevalence of this disease is <1/million with worldwide incidence is reported to be 1/13,250 to 1/140,000 live births [2]. The incidence is significantly higher in the Finnish population (1/9,000), Belgian and Kuwaiti Bedouin populations (1/3,500), and Gujarati Indians (1/1,300) [2]. No gender predilection is reported.

2. Case

A 27 year old muslim female G2P1L1 came to Maharani Laxmibai Medical College (Jhansi, India) OPD with a history of amenorrhea of 7 months and complaints of pain in abdomen and leaking per vaginum since 1 day. Per vaginal examination revealed that the patient was in labour with 3 cm dilatation of cervical os. She was admitted for delivery of the baby. Her ultrasonography (USG) revealed a live fetus with minimal hydrocephalous, occipital encephalocele, limb hypoplasia, severe oligohydramnios, pulmonary hypoplasia and ascites. Face was not clearly visualized in USG and there was no visualization of stomach bubble. Disparity was seen in average gestational age estimation on the basis of abdominal circumference and femur length. Risk regarding incompatibility of life of the fetus was explained to the parents.



Figure 1: Baby showing features of ascites (distended abdomen), short limbs, hydrocephalous, cleft lip with cleft palate with maxillofacial defects

A gasping male baby was delivered vaginally with no post partum obstetric complications. First and fifth minute Apgar scores were 1 and 0, respectively. Consequently, the baby died after 45 minutes despite neonatal resuscitation. Apart from hydrocephalous, there was cleft lip with cleft palate with maxillofacial defect (Fig 1-2). All the four limbs showed shortening with bowing of lower limbs and presence of post-axial polydactyly (Fig 3-4). Ascites was clinically evident. As consent for autopsy and genetic analysis was not given by parents further investigations could not be carried out. Based on the presence of classical features, a diagnosis of Meckel syndrome was made. Patient was screened for diabetes, TORCH, thyroid and VDRL to rule out any other etiology. Patient was found to be hyperthyroid.

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Figure 2: Lateral view showing distended abdomen, short curved limbs, hydrocephalous, and maxillofacial defects.

3. Discussion

The specific symptoms associated with Meckel syndrome vary greatly from one individual to another. In our case prenatal USG revealed minimal hydrocephalous with occipital encephalocele, limb hypoplasia and severe oligohydramnios with pulmonary hypoplasia. On delivery of the baby, USG findings were



Figure 3: Dosral view showing ascites and absence of neck.

confirmed by the presence of hydrocephalous, limb hypoplasia with further findings of polydactyly, ascites and maxillofacial defect in the form of cleft lip and cleft palate. Since autopsy was not performed, kidney abnormalities cannot be ascertained. However as per diagnostic criteria, two of the three characteristics of the triad viz., hydrocephalus-occipital encephalocele and polydactyly, were present to confirm the diagnosis of Meckels syndrome. Central nervous system, pulmonary or kidney abnormalities always result in perinatal death.



Figure 4: Features showing polydatyly and syndactyly.

Multiple cysts on the kidneys (polycystic kidneys) are the most common symptom associated with MS. Findings associated with polycystic kidneys includes loss of kidney function, leading to end-stage renal failure. Improper kidney function may also result in a reduction in the amount of amniotic fluid surrounding the developing fetus (oligohydramnios). In this case we observed oligohydromnios suggesting the kidney defects.

The most common central nervous system abnormality associated with MS is occipital encephalocele which was observed prenatally in our case through ultrasound presence of cleft lip and cleft palate and maxillofacial defects was detcted. In this case further support the craniopharyngeal growth defects (Fig 1-2). It is reported that affected infants may have distinctive facial features including an abnormally small jaw (micrognathia); enlarged, low-set and malformed ears; cleft palate; cleft lip; sloping forehead; and short neck. Affected children may have eye (ocular) abnormalities including abnormally small eyes (microphthalmia), and underdevelopment of the nerves of the eyes (optic nerve hypoplasia or coloboma) [3].

In our case, all the four limbs of the baby were shortened with bowing of lower limbs and presence of post-axial polydactyly which further confirms our diagnosis (Fig 3-4). Additional skeletal malformations include bowing of the long bones of the arms and legs, curvature of the fifth fingers (clinodactyly), webbing of the fingers and toes (syndactyly), and club foot where the foot are rotated internally (talipes equinovarus).

External genitalia seemed normal in our case. Further comment on the rest of the genitourinary system could not be made as autopsy was not done. It has been observed that in some cases defects in genitourinary tract may be present including failure of the one or both testes to descend into the scrotum (cryptorchidism), underdeveloped (hypoplastic) bladder, and incomplete development of genitalia.

Miscellaneous findings associated with MS include hepatomegaly, pulmonary hypoplasia, defects of biliary ducts, splenomegaly or asplenia, cardiac abnormalities. In our case pulmonary hypoplasia was evident in prenatal USG. In the absence of autopsy report, the clinically evident ascites in the baby after delivery is consistent with the presence hepatomegaly, splenomegaly or renal cyst.

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MS is an autosomal inherited recessive disorder and has a recurrence risk of 25% [2]. Parents who are close relatives (consanguineous) have a higher chance than unrelated parents. Though the parents in our case are not closely related, but consanguineous marriages are common practice in local muslim population which increases the risk to have children with a recessive genetic disorder. Since genetic study could not be undertaken in our case, we are unable to comment on the gene involvement.

4. Conclusion

MS is a rare neural tube defect associated with wide variety of malformations. No treatment is currently available for Meckel syndrome which has a fatal outcome. Prenatal diagnosis by ultrasonography is possible as early as 12 weeks of pregnancy. Encephalocele, cystic kidneys and polydactyly may be detected and alfa fetoprotein level may be suggestive. Since MS can be detected in early gestational period (12-14 weeks), genetic testing can confirm the diagnosis. Accordingly counseling and management should be provided to the affected families.

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