International Journal of Contemporary Pediatrics
Verma M et al. Int J Contemp Pediatr. 2018 Jan;5(1):262-264
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20175598

Case Report

Meckel Gruber Syndrome: a rare case report

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Received: 29 October 2017
Accepted: 25 November 2017

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ABSTRACT

Meckel gruber syndrome or dysencephalia splanchnocystica, is a rare autosomal recessive disorder caused by failure of mesodermal induction. Worldwide incidence of MGS is 1 per 13,500-140,000 live births. It is characterized by triad of occipital Meningoencephalocele, polycystic kidneys and post-axial polydactyly. Most fetuses affected with this syndrome die before birth due to oligohydramnios, renal failure or pulmonary hypoplasia. We report a rare case of MGS who delivered live at birth with classical features.

Keywords: Occipital encephalocele, Polysystic kidney, Polydactyly, Pulmonary hypoplasia

INTRODUCTION

The first reports of MGS were published in 1822 by Johann Friedrich Meckel, later Gruber GB also reported similar patients in 1934 and gave the term dysencephalia splanchnocystica.[1,2] Meckel gruber syndrome is a pleiotropic autosomal recessive disorder caused by dysfunction of primary cilia during early embryogenesis. Classical triad consist of cystic renal disease, central nervous malformation most commonly occipital encephalocele and polydactyly mostly postaxial.[3-5] Additional hepatic development defect or hepatic fibrosis may occur. Affected children or foetuses may also have abnormalities affecting the craniofacial area, lungs, heart and genitourinary tract. 12 different loci responsible for MGS have been mapped on chromosomes showing genetic heterogenicity.[6]

CASE REPORT

26 years old female, second gravida, non-consanguineous marriage came for routine antenatal check-up around 28th week of gestation. Abdomen ultrasonography showed breech presentation and moderate oligohydramnios.

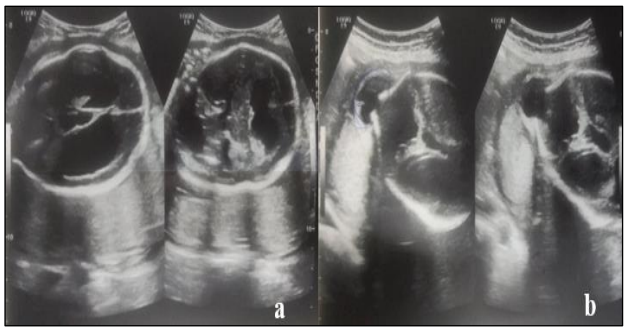

Figure 1: (a) Dilatation of fetal ventricular system; (b) cystic swelling projecting from occipital region.

Fetal head showed severe dilatation of ventricular system (V/H ratio >85%). Both fetal kidneys were enlarged in size with presence of multiple small cystic areas, probably multicystic renal dysplasia. Cystic area is seen projecting out from occipital region, suggestive of occipital

encephalocele. Fetal hepatomegaly and smaller thorax were also seen. Later mother delivered at 36th week by normal breech delivery. Apgar score at birth was 1 and baby survived for 45 minutes.

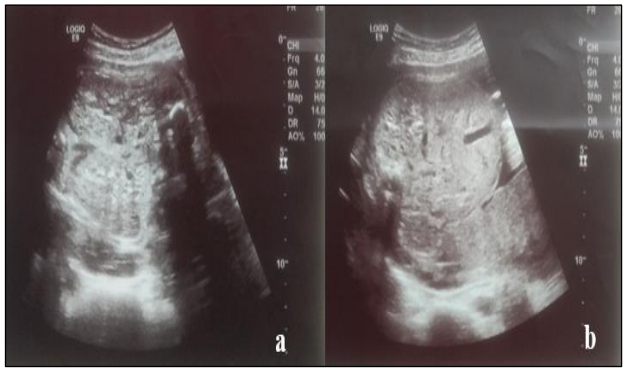

Figure 2: Both kidneys enlarged and showing multiple cystic area.

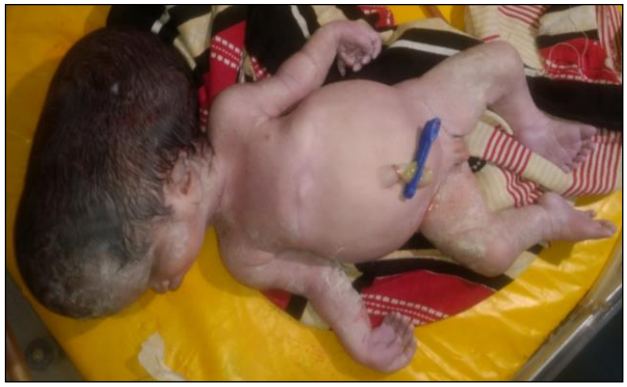

Figure 3: baby with occipital encephalocele, low set ears, retrognasthia, short neck, small chest with widely placed nipples, flanks full in abdomen, postaxial polydactyly, b/l CTEV.

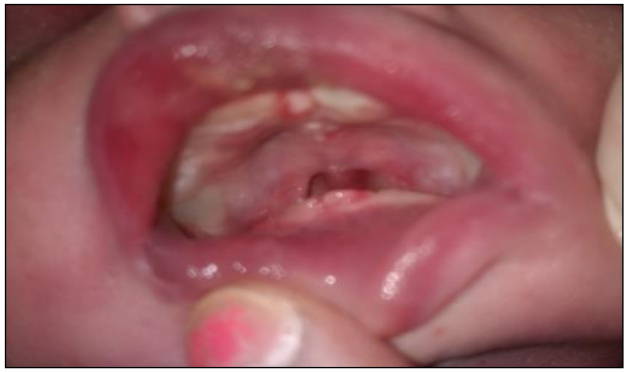

Figure 4: Cleft palate in posterior part with tongue fallen back.

Baby had large head with cystic swelling both in frontal and occipital region suggestive of encephalocele. Baby also had retrognathia, low set ears, smaller mouth opening, cleft palate, glossoptosis, short neck, small chest with widely spaced nipples, flanks full in abdomen (due to renal enlargement), polydactyly in all limbs and bilateral CTEV. During neonatal resuscitation it was almost impossible to intubate the baby due to smaller mouth opening and glossoptosis, which were not allowing blade of laryngoscope to be introduced inside. Patient had no spontaneous breathing at birth and expired within one hour of birth, probably due to pulmonary hypoplasia and associated co-morbidities (Figure 1, 2, 3, 4 and 5).

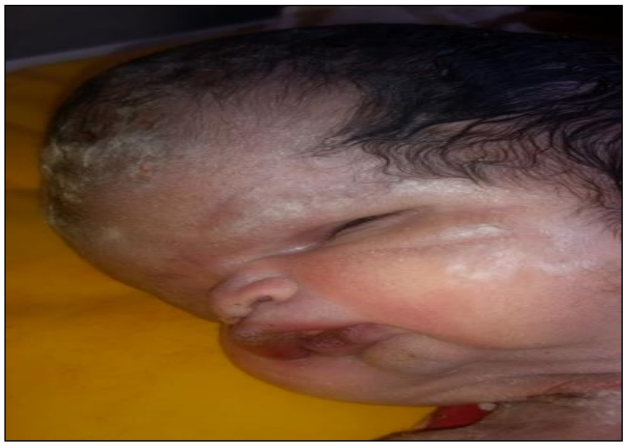

Figure 5: Bulging forehead, flat nasal bridge and retrognathia.

DISCUSSION

MGS was first described by Johann Friedrich Meckel in 1822 in two siblings with identical malformations of occipital encephalocele, polycystic kidneys and polydactyly. Gruber GB in 1934 reported many familial cases with similar features and coined the term "dysencephalia splanchnocystica". In 1969, Opitz and Howe proposed the name Meckel's syndrome (MKS).[4,7]

MGS is characterized by the triad of:

- Renal cystic dysplasia- multiple renal cysts (present mostly),
- Occipital encephalocele or holoprosencephaly (~70%),
- Postaxial polydactyly- usually hexadactyly (~65%).

Additional variety of malformation may be associated with this syndrome but most agree that cystic renal dysplasia must be present with at least one of the other two anomalies in the classic "triad". Craniofacial abnormalities include microcephaly, ventriculomegaly, corpus callosal agenesis, dandy walker malformation, micrognathia and cleft lip/palate. Cardiovascular abnormalities include cardiac rotation defects, VSD, aortic hypoplasia, aortic coarctation and aortic valvular stenosis. Musculoskeletal abnormalities include webbed neck, club foot, syndactyly and clinodactyly. Abnormalities like lung hypoplasia and club foot are secondary to oligohydramnios.[8,9] This condition is usually diagnosed by antenatal USG and

elevated maternal serum alpha fetoprotein. The mean gestational age at diagnosis is 19 weeks.[10] Its almost always fatal, baby die in utero or shortly after birth due to pulmonary hypoplasia and neonatal renal failure.[11,12] OMIM online article number 24900019 has given list of 12 types of MKS with mutation on gene locus of that particular chromosome.[13]

CONCLUSION

MGS is rare syndrome and can be diagnosed during antenatal USG. Our case diagnosed relatively late but alive with classical features involving renal, craniofacial, musculoskeletal and lung malformations. Parents should be counselled about 25% risk of subsequent sibling being affected.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Verma M, Sharma S, Suthar K, Thada B. Meckel Gruber Syndrome: a rare case report. Int J Contemp Pediatr 2018;5:262-4.