Case Report

Meckel Gruber Syndrome — A Case Report

Kalyani. R., *Professor, Department of Pathology,*

Mandeep Singh Bindra, *Resident, Department of Pathology,*

Subhashish Das, *Assistant Professor, Department of Pathology,*

Hemalatha Mahansetty, *Professor & HOD, Department of OBG*

— Sri Devaraj Urs Medical College, University, Tamaka, Kolar. Karnataka.

Abstract

Meckel Gruber Syndrome is a rare and interesting congenital anomaly characterised by triad of occipital encephalocele, bilateral cystic renal disease and postaxial polydactyly. We report this syndrome in a male fetus of 31 weeks' gestational age and review the literature.

Keywords

Fetus, Meckel Gruber Syndrome, Dysencephalia splanchnocystica

Introduction

Meckel Gruber Syndrome (MGS) dysencephalia splanchnocystica is an autosomal recessive inherited disorder characterised by triad of occipital encephalocele, bilateral cystic renal disease and postaxial polydactyly[1,2]. Other associated abnormalities include heart, liver, genital, facial and skeletal defects[3,4].

Worldwide the incidence varies from 1 in 13,250 to 1 in 1,40,000 live births affecting male and female babies of all ethnic background[3,5]. It accounts for 5% of all neural tube defects.1 The defective genes are identified in chromosome 13, 17 and 11.5 Recurrence in subsequent pregnancy is 25% (1 in 4)[1,3]. Diagnosis can be confidently done at 11-14 weeks of gestation[6].

We present a case of MGS and review the literature regarding the various congential abnormalities and discuss the differential diagnosis.

Case Report

A 22-year female, gravida 2, living 1, attended antenatal clinic regularly. At 30 weeks of gestation, an antenatal ultrasound scan revealed single live fetus with severe oligohydramnios, occipital encephalocele, colpocephaly and bilateral cystic renal disease. Hence patient underwent medical termination of pregnancy. A clinical autopsy was performed on the fetus.

Autopsy Findings

The fetus was 38 cm in length and weighted 2200 gm. External examination revealed skin covered cystic swelling in occipital region measuring 11x8x2 cm, depressed nasal bridge, low set ears, cleft palate, ugnathia and short neck (**Fig. 1**). Both upper limb palms showed polydactyly (6 fingers) and lower limb feet showed syndactyly. Scrotum was normal but penis was not formed. Anal opening was normal with meconium staining.

Umblical cord was 6 cm in length, cut section showed normal three blood vessels. X-ray of the fetus was normal except for polydactyly of upper limbs. On systemic examination brain showed features of occipital encephalocele with colpocephaly on cut section (**Fig. 2A**). Right kidney measured 5x3x2 cm and and left kidney 4.5

Address for correspondence: Dr Kalyani. R., Professor of pathology, H.No: 127/13, "Sri Ganesh", 4th main, 4th cross, P. C. Extension, Kolar – 563101, Karnataka. E-mail: drkalyanir@rediffmail.com

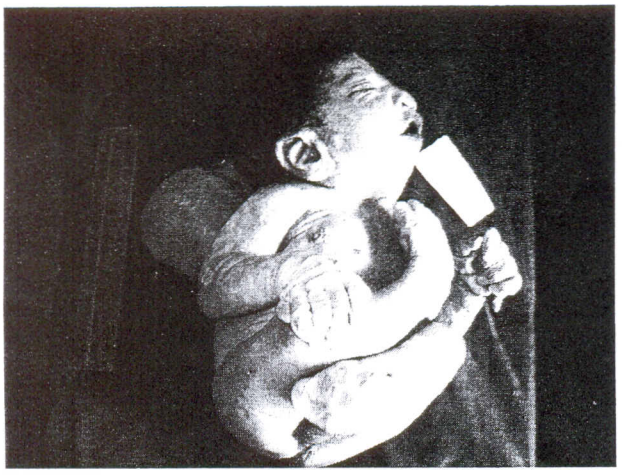
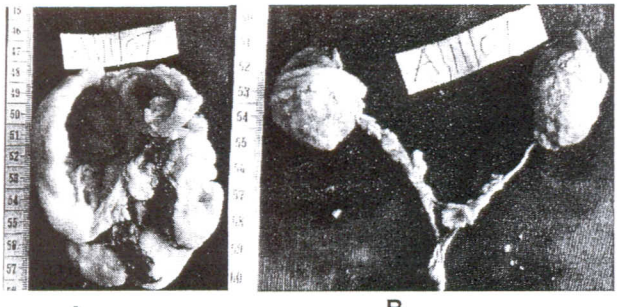


Fig 1
Gross photograph of fetus showing occipital encephalocele, depressed nasal bridge, postaxial polydactyly of hands and syndactyly of feet.



A B

Fig 2
A. Gross photograph of cut section of brain showing colpocephaly. B. Gross photograph showing bilateral cystic kidneys.

x 3 x 2 cm (**Fig. 2B**). External surface of both kidneys showed multiple cystic swellings with multiple cysts in cortex and medulla on cut section. Histopathologically both kidneys showed features of cystic renal dysplasia. Both lungs grossly and on histology showed features of non-areated lungs. Organs of other system were normal. A final diagnosis of MGS was made.

Discussion

Meckel Gruber Syndrome was the first described by German Anatomist Johann Friedrich Meckel in 1822 in a pair of siblings later German Pathologist George Benno Otto Gruber in 1934, coined the term dyseticephalia splanchnocystica, and more recently by Optiz and Howe in 1969[2,3,4]. The syndrome has prediliction for Belgian (1/3000) and Finish (1/9000) population with M:F:1:13. Analysis of polymorphic DNA markers from finish families revealed MGS locus on 17q21 - q24, second locus on 11q13 and third locus on 8q24[3]. Both parents of the fetus carry copies of the defective gene who may have more than one infant with this disorder[5]. Recurrence of a neural tube defect is 1.3% in a given family[3].

The classic triad includes neural tube defects particularly occipital encephalocele in 80-90% cases, postaxial polydactyly in 75-83% cases and bilateral cystic renal disease in 95-100% cases. Defect in occiptial bone posteriorly with brain matter covered by meninges herniating through it associated with severe dilatation of venticular system is the usual presentation[4]. Other rare neurological defects are celebellar and cerebral hypoplasia and hydrocephalous[4]. Apart from postaxial polydactyly other skeletal abnormalities reported are syndactyly, clindodactyly, clubbed foot, short webbed neck, cleft lip/palate, micrognathia and symmetrical short limbed dwarfism[3,4,5]. Infantile polycystic kidney is also reported[4]. Genital anomalies like hypoplastic penis, cryptorchism, mullerian duct remnants, epididymal cysts, septate vagina and hypoplastic/bicornuate uterus are also noted4. Other anomalies includes heptaic fibrosis, accessory spleen, hypopitutarism, congenital heart disease and pulmonary hypoplasia[3,4]. A single case of MGS with Rokintansky-Kuster-Hauser syndrome has been reported4.

Diagnosis of MGS was done by ultrasound scan between 11 - 14 weeks of gestation. Diagnosis in late pregnancy becomes difficult due to severe oligohydramnios[3,6]. The differential diagnosis for MGS are in **Table 1**[1,3]. Prognosis is lethal as fetus die shortly after birth because of too small lungs and abnormal kidney which cannot support life. Hence termination of pregnancy becomes mandatory before the viability of fetus.

In our case the male fetus diagnosed at 30 weeks had classical triad and in addition showed other anomalies like depressed nasal bridge, low set ears, cleft palate, ugnathia, short neck, syndactyly and incomplete formation of penis. Fetus died immediately after birth. Genetic study on the fetus and parents would have helped for genetic counselling in the present case.

Table 1
Differential Diagnosis of Meckel Gruber Syndrome

Smith-Lemi-Optiz Syndrome • Autosomal recessive • Multiple malformation CNS / GU System • Postaxial polydactyly • Abnormal remodeling of liver ductal plate. • Defect of enzyme 7 - dehydrocholesterol 67-reductase in cholesterol biosynthesis • Broad and high forehead • Bilateral ptosis • Epicanthal folds • Transverse palmar crease	**Hydrolethalus Syndrome** • Autosomal recessive • Lethal malformation • Polydactyly (Duplicate big toe) • Micrognathia • Hydrocephaly • Absent midline structures of brain	**Trisomy 13** • CNS malformation • Cystic renal dysplasia • Postaxial polydactgly • No liver fibrosis • Pancreatic dysplasia • CVS malformation • Occular abnormality
Bardet - Biedl Syndrome • Multisystem disorder • Postaxial polydactyly • Progressive renal dystrophy • Obestiy • Hypogonadism • Liver anomalies • Encephalocele absent	**Joubert Syndrome** • Autosomal recessive • Meningoencephalocele • Microcephaly • Hypoplastic / aplastic cerebellar vermis • Polydactyly • Low set ears • Kidney & liver abnormality • Retinal dysplasia • Soft tissue tumour of the tongue	**Larsen Syndrome** • Polycystic kidney disease • Bilateral dysgenesis • Osteosclerosis • Multiple joint dislocation

Conclusion

Knowledge of this syndrome and appropriate care during antenatal scanning would enable the sonologist to make an early antenatal diagnosis. As the anomalies in MGS is incompatible with life, termination of pregnancy is warranted.

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