

## Meckel-Gruber syndrome

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### Abstract

Meckel-Gruber syndrome is a rare lethal, autosomal disorder. It has been linked to chromosome 17. It consists of a triad of occipital meningoencephalocele, large polycystic kidneys and post-axial polydactyly. Death is mainly due to pulmonary hypoplasia. We report this rare case which presented with many associated defects.

**Key words:** - MKS syndrome, meningoencephalocele

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### Case Report

A male baby was brought to this hospital at the age of 9 hours. He was born to a 3<sup>rd</sup> gravida mother. She had attended antenatal check-ups on one occasion at the local health. No USG was done. The baby cried weakly after birth

On examination at the hospital the following were noted: -

Anthropometric data Wt -- 3.1 kg, Length—45cm, Head circumference—31cm

Assessed gestational age –full term

Mild peripheral cyanosis

Respiratory rate—70/min, Heart rate-160/min, Spo2-88%

Congenital anomalies—Large occipital encephalomeningocele—15x10cm

Central cleft palate, micrognathia and retrognathia, sloping forehead, microcephaly, partial absence of nasal septum, short neck, cardiomegaly with pansystolic murmur and small poorly developed scrotum with undescended testes.

### Investigations done

USG –grossly enlarged kidneys though cystic changes were not obvious, liver showed prominent biliary system but no cystic changes. X-Ray chest showed cardiomegaly, neurosonogram showed an encephalomeningocele but no other abnormality of the brain. CT scan was not agreed to by the parents.



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## Discussion

MKS is also called Dysencephalia Splanchnicocystica. It was originally described by Meckel in 1822 and later by Gruber<sup>1</sup>. More recently it was described by Howe and Optiz in 1969. More than 200 cases have been reported to date. It occurs in all races and ethnic communities though the highest incidence has been reported in the Finnish population with an incidence of 1 in 9000<sup>2</sup>. The incidence worldwide has been reported as 1 in 13,250 to 1 in 140,000 live births. Male to female ratio is more or less equal which is consistent with an autosomal inheritance pattern.

Many congenital anomalies are present of which the most striking is the occipital encephalocele<sup>3</sup>. This consists of extrusion of parts of the brain parenchyma, which is fully covered by a dural sac. Growth is variable and intra-uterine growth delay may be present.

Head and neck: - occipital encephalocele, microcephaly, anencephaly, cerebral or cerebellar hypoplasia, hydrocephalus with or without Arnold - Chiari malformation, absence of olfactory tract or lobe, absence of Corpus Callosum and septum pellucidum are all seen in this condition. Dandy-Walker malformation is seen in some patients. Microphthalmia, cleft palate, sloping forehead, micrognathia, ear abnormalities and short neck may also be seen.

Though postaxial polydactyly is a feature of MKS in some cases it is not exhibited<sup>4</sup>. Although all four limbs may be affected this is the most variable feature of the classic triad of major abnormalities.

Kidney: -Cystic dysplasia is the most constant characteristic feature of MKS. Due to this there is oligo-hydramnios and pulmonary hypoplasia which is the main cause of death. Kidneys may be enlarged 10-20 times the normal size.

Hepatic lesions: - these are consistent features but can be considered hidden abnormalities as they are often seen only on post mortem examination. There is arrested development of the intrahepatic biliary system with varying degrees of reactive bile duct proliferation, bile duct dilatation, portal fibrosis and portal fibrous obliteration.

Genital: - cryptorchidism with incompletely developed external or internal genitalia may be seen. Genital ambiguity may lead to difficulty in sex assignment to the baby.

Occasional abnormalities: - many other defects are seen including Craniosynostosis, coloboma of the iris, Hypoplastic optic nerve and hypoplastic or absent nasal septum. Cleft lip, lobulated tongue, cleft epiglottis, prenatal teeth, short webbed neck and short bowed limbs are also seen. Cardiac defects include VSD and ASD as well as PDA and pulmonary stenosis. Abdominal examination may reveal Single umbilical artery, patent urachus, omphalocele, malrotation of the gut, accessory spleen, adrenal hypoplasia, imperforate anus and hypoplasia of the urinary bladder.

Natural history and management: - this condition has 100% mortality with death in utero or soon after birth. They seldom survive more than a few days. The longest known survivor was 4 months. Since there is no treatment it is preferable to diagnose the condition prenatally and abort the affected foetus. Foetal USG can detect many defects. Chorionic villi sampling can be done at 14 weeks of gestation. MRI also can be done prenatally to see foetal defects.

MKS can mimic Trisomy 13 which has a 1% chance of recurrence while MKS has a 25% chance hence chromosomal studies should be done. Because of this resemblance to Trisomy 13 the defective gene for MKS was postulated to be on Chromosome 13. However analysis of polymorphic DNA markers from 5 Finnish families revealed the MKS locus to be Chromosome bands 17q21-q24. A subset in the Middle East and Northern African families did not show this linkage to chromosome arm 17q. A second locus (MKS2) has been mapped to band 11q13.(5) Recently MKS 3 has been localized to chromosome 8q24 and has less frequency of polydactyly as compared to the other two types.

In conclusion we present this rare and interesting case of MKS. In our case there was no consanguinity. However the parents have been counselled to come for a check up if the mother again becomes pregnant.

TM and KSJ were involved in documenting all the findings and helped in collection of data. UR was responsible for the preparation of the paper.

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