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CASE REPORT

Meckel-Gruber Syndrome (Dysencephalia Splanchnocystica)

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ABSTRACT

Meckel-Gruber syndrome is a rare lethal autosomal recessive condition which was first described by Johann Friedrich Meckel in 1822[1] and GB Gruber in 1934.[2] More than 200 cases have been reported worldwide with an incidence ranging from 1:13,250 to 1:140,000 live births.[3]

A 21-year-old female with G3 A2 L0, presented with twin pregnancy with history of previous two anencephalic pregnancies. The present pregnancy was a preterm vaginal delivery of female twins by face presentation at 35 weeks of gestation (diamniotic dichorionic twin gestation).

Neonatal autopsy revealed classical triad of occipital encephalocele, polycystic kidneys and lungs with postaxial polydactyly.

This case is presented for its rarity and its documented occurrence in Gujarati Indians.

Keywords: Meckel-Gruber, Dysencephalia splanchnocystica, Encephalocele, Polycystic kidney, Polydactyly.

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BACKGROUND

Meckel-Gruber syndrome is a rare, specific inherited, lethal, autosomal recessive disorder also known as dysencephalia spanchnocystica. which was first described by Johann Friedrich Meckel in 1822 and GB Gruber in 1934.[1-3] More recently the disorder was brought to recognition by Opitz and Howe.[4] More than 200 cases have been reported worldwide with an incidence ranging from 1:13,250 to 1:140,000 live births.[3]

CASE REPORT

A 21-year-old female with G3 A2 L0 presented with 9 months amenorrhea, pain abdomen and watery discharge per vagina with twin pregnancy. Patient had a past history of previous two anencephalic pregnancies. Marital history revealed nonconsanguineous marriage. No history of any drug intake during the pregnancy.

Obstetric history revealed that first pregnancy was after 1 year of married life. It was diagnosed as anencephalic fetus at 4th month by ultrasonography and was terminated.

Second pregnancy was 1 year after the first pregnancy. It was again diagnosed as anencephalic fetus at 4th month by ultrasonography and was terminated.

The present pregnancy was 1½ years after the second pregnancy. It was a preterm vaginal delivery of still born fetuses by face presentation at 35 weeks of gestation (Diamniotic dichorionic twin gestation).

Investigations revealed, TORCH panel—normal. Karyotype showed mother-46XX and father- 46XY. Alfa-fetoprotein was increased. Ultrasound examination showed anencephalic twin pregnancy.

Twin female fetuses received with placenta for complete autopsy revealed following features:

Common features were as follows:

1. Anencephaly (absence of cranium with replacement by cystic growth) (Figs 1 and 2).
2. Low set ears (see Figs 1 and 2).
3. Webbed neck (Fig. 1).
4. Polydactyly, talipes equinovarus (Figs 1 and 3).
5. Cyst in kidneys, lungs and liver (Figs 4 and 5).
6. Hypoplastic adrenal glands.

Differences

1st twin	2nd twin
Cleft lip and palate Fissured tongue with nodular outgrowths	Absence of eye balls

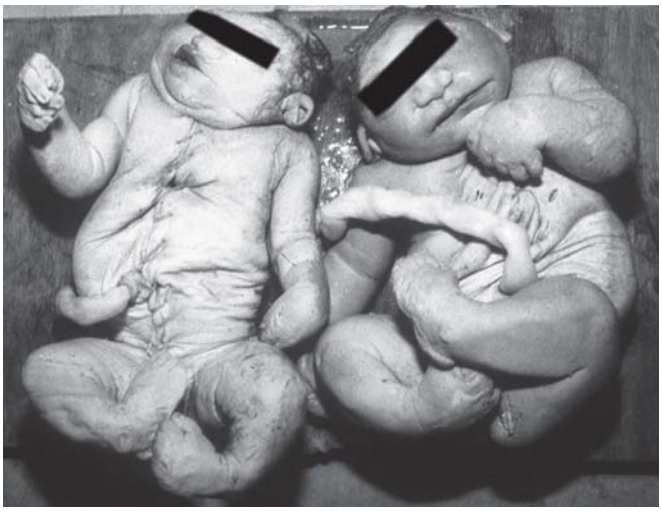

Fig. 1: Twin fetuses revealing anencephaly, low set ears, webbed neck, polydactyly and talipes equinovarus

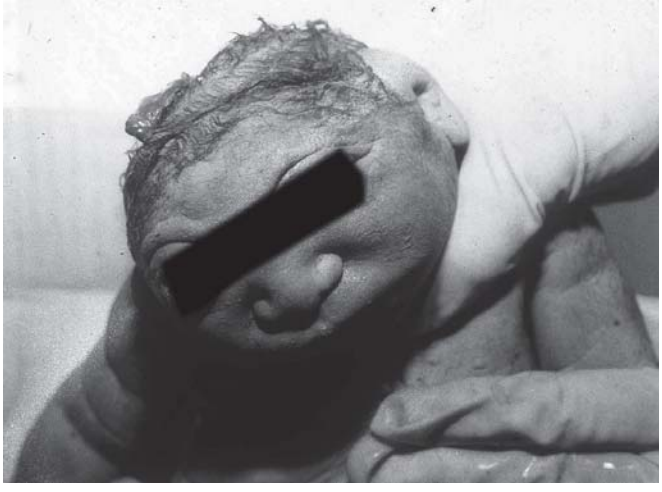

Fig. 2: One of the twin fetuses showing anencephaly and low set ear

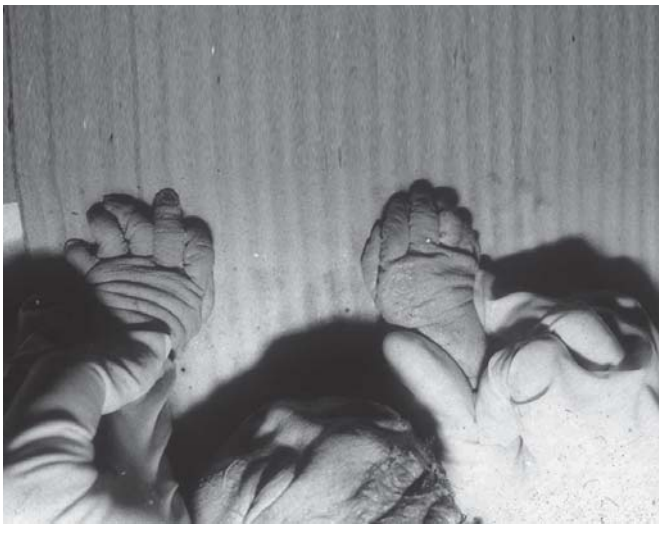

Fig. 3: Clinical picture revealing polydactyly

Microscopy

Brain: Area cerebrovasculosa
Kidneys, lungs and liver showed cystic lesions.

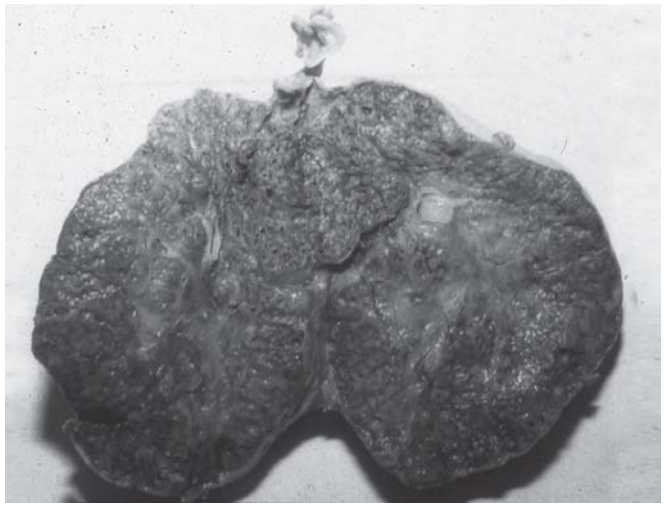

Fig. 4: Gross picture of kidney showing cystic changes

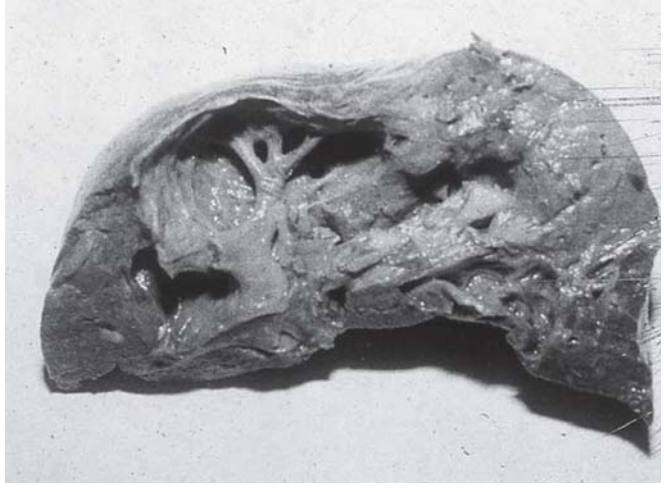

Fig. 5: Gross picture of liver showing cystic changes

DISCUSSION

Meckel-Gruber syndrome is a rare, lethal and autosomal recessive disorder which was first described by Meckel in 1822 and Gruber in 1934[1-3] and hence the name Meckel-Gruber syndrome.

More than 200 cases have been reported worldwide with an incidence ranging from 1:13,250 to 1:140,000 live births.[3] Frequency of incidence worldwide varies from 1: 3,000 (Belgium) to 1: 140,000 (Great Britain). There is high incidence of occurrence in Gujarati Indians (1: 1,300 – live births).[3]

Approximately one-third are still born and the remaining rarely lives more than 4 days. Death is due to lung and kidney failure. A genetic link was established only in 1969, showing an autosomal recessive inheritance mapped to six different loci in chromosomes suggesting genetic heterogeneity.[3,13]

Reliable diagnosis can be made by ultrasonography after the 11th week of gestation.[5,8] Magnetic resonance imaging,[9]

raised alpha-fetoprotein levels and chromosomal analysis also help in the prenatal diagnosis.[6]

The syndrome is characterized by classical triad of occipital encephalocele, polycystic kidneys and lungs with postaxial polydactyly. It is also associated with cleft lip, cleft palate, micrognathia, talipes equinovarus, microphthalmia, anophthalmia, hypoplastic phallus and cryptorchidism in males.

Meckel-Gruber syndrome requires to be differentiated mainly from trisomy 13 (Patau syndrome)[12] usually does not show encephalocele, polycystic lesions of kidneys/liver and polydactyly.[7,10,11]

CONCLUSION

Meckel-Gruber syndrome is a rare, lethal and autosomal recessive disorder which should be diagnosed prenatally and the parents should be counseled.

CLINICAL SIGNIFICANCE

Meckel-Gruber syndrome is an absolutely devastating congenital inherited disorder, when diagnosed prenatally, the mother has two choices: Either terminate the pregnancy or carry on to term. Both choices result in the death of the child.

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