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MECKEL GRUBER SYNDROME: CLASSICAL CLINICAL DIAGNOSIS OF A CASE IN A NON-CONSANGUINEOUS MARRIAGE

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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.

Key Words: *Non consanguinity, Occipital encephalocele, Polysystic kidney,post axial Polydactyly.*

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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. 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.(1,2) 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.

CASE REPORT:

1 hr old male child presented in pediatric emergency Mayo hospital lahore with complaints of respiratory distress since birth.Baby was born via SVD at 37th weeks of gestation by gravida two para 2 mother at home.Mother's history revealed that her last pregnancy had history of early neonatal death due to suspected Syndrome. Because of her cultural and religious constraints,mother didn't had her antenatal follow up visits regularly. She has history of nonconsanguineous marriage. Antenatal ultrasonography was suggestive of occipital encephalocele &,microcephlic. No anomaly scan was done. Baby born with poor APGAR 2/10 with in first minute & 3/10 up to 5mins after delivery and a weight of 2.4 kg. The new-born was examined in ER and the external examination revealed Occipital encephalocele (Fig. 1)with head circumference 25.5 cm, upper slanting of eyes,overriding of sutures,(Fig 2) widely spaced nipple with small chest and distended abdomen due to bilateral renal mass (Fig. 4), flexion of both wrist joints, knee joint and ankle joints bowed legs with clubbed feet, shown in infantogram (Fig,3 & 4) post-axial hexadactyly on both hands & feet(Fig 5,) We admit the newborn in NICU with suspected MGS, based on the presence of the classical features. The parents were counselled regarding the syndrome & prognosis of neonate. They denied consent for genetic analysis.The baby expired at 9th day of life due to pulmonary hypoplasia and renal failure.

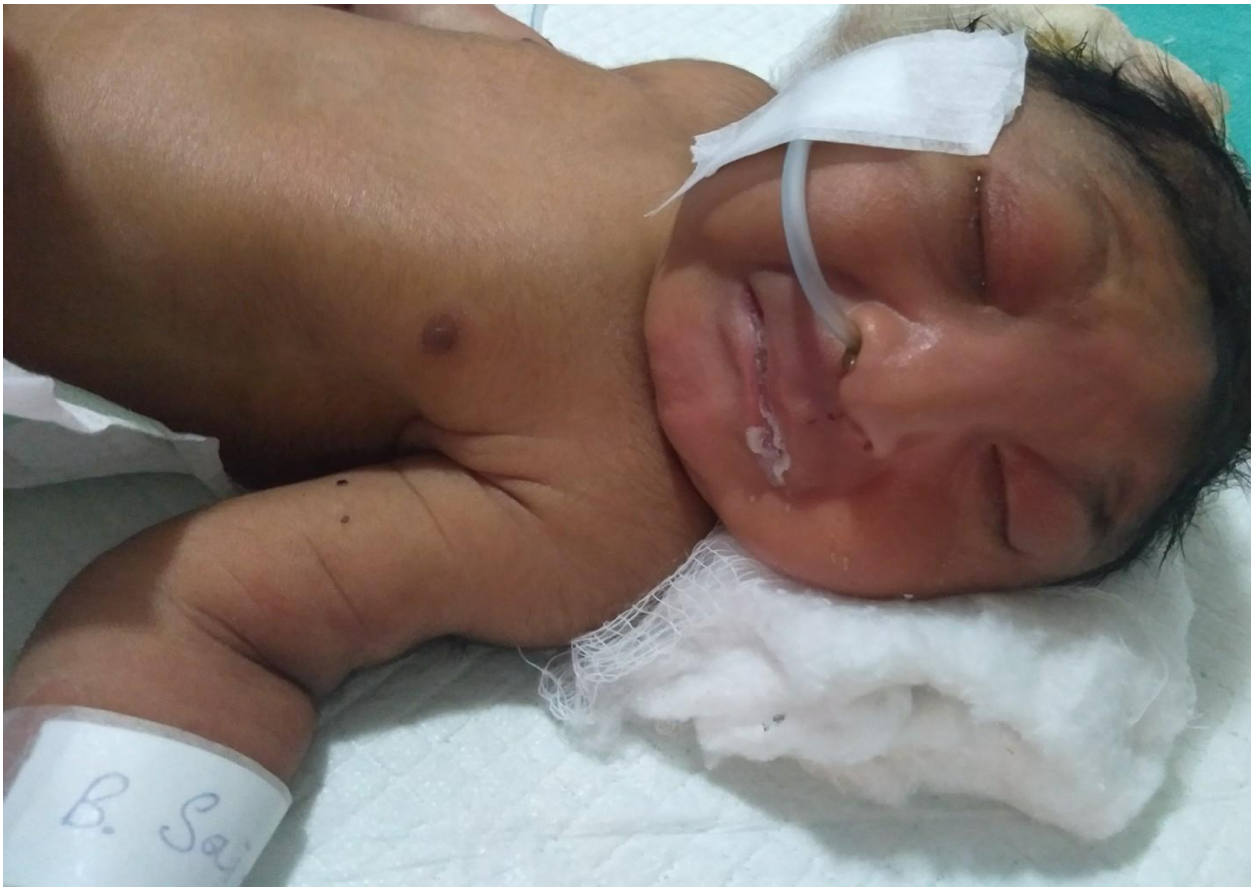
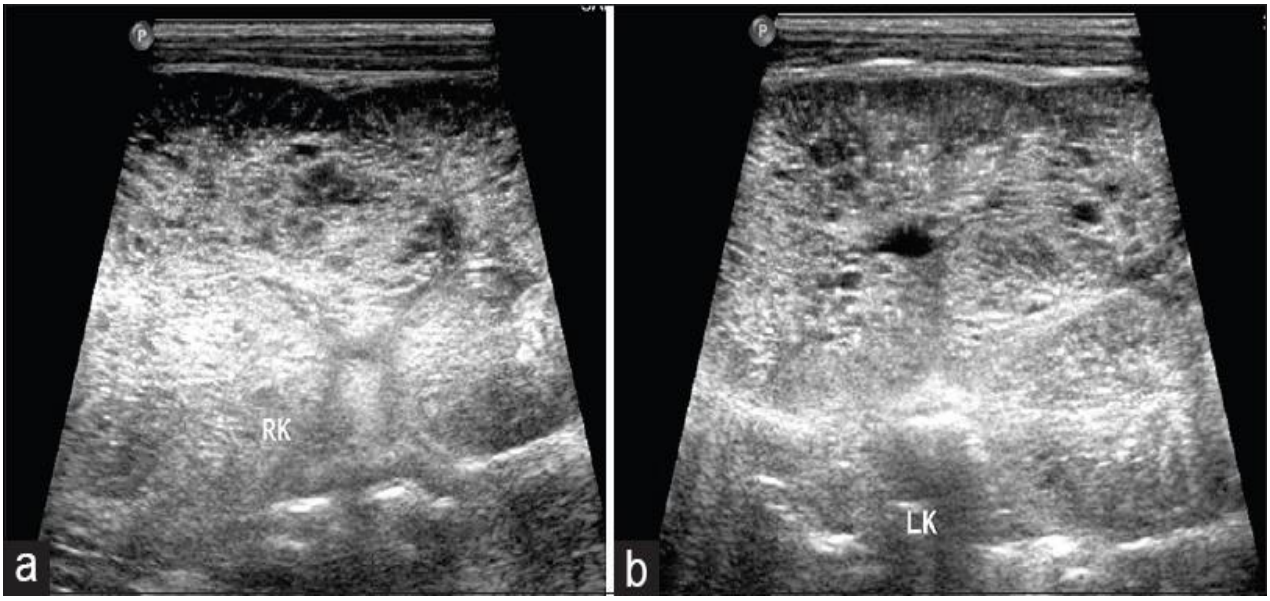


Fig 2 shows microcephalic,coarse facies, upward slanting eyes,



Ultrasound KUB shows Both large polycystic dysplastic kidneys.

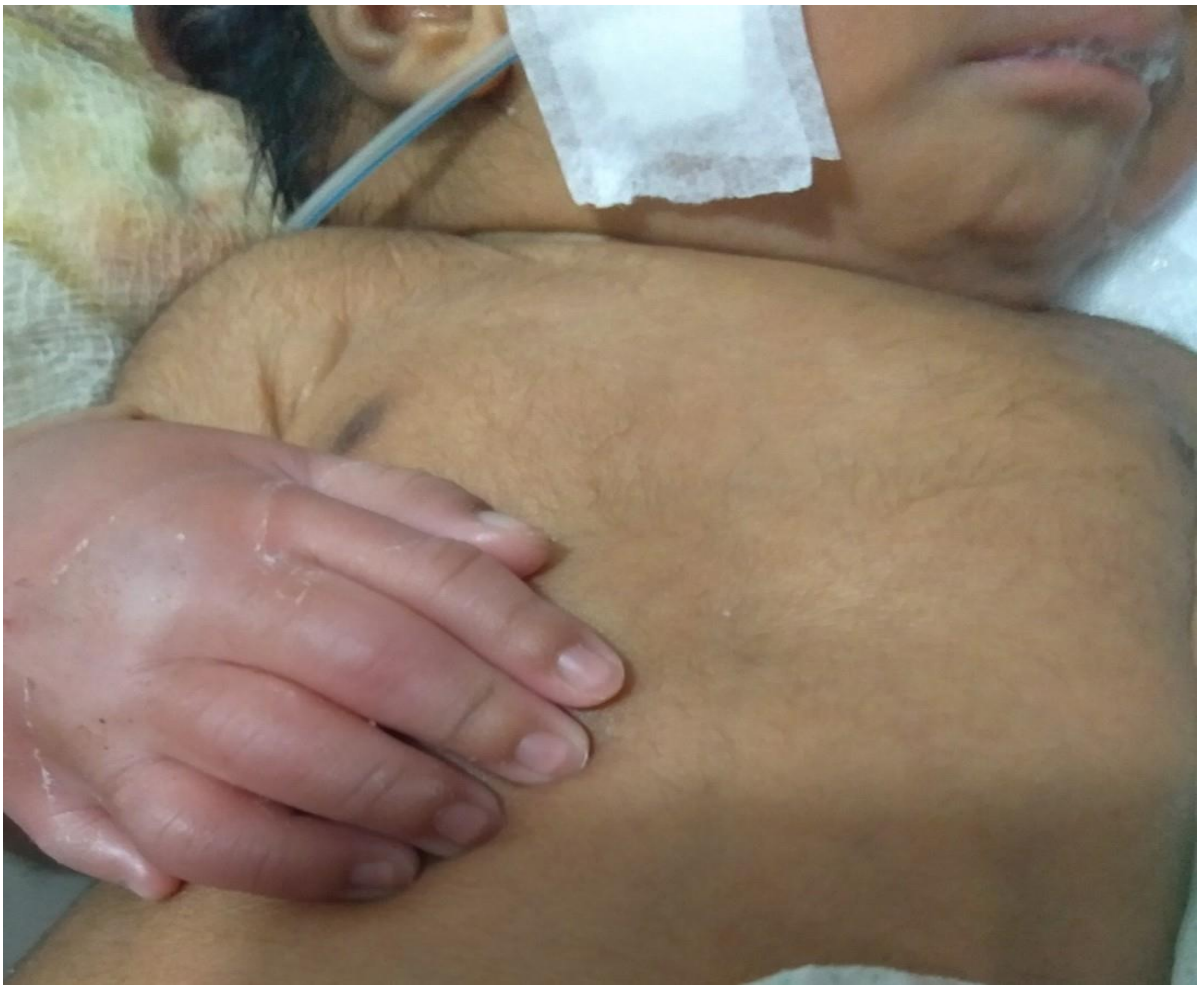


Fig 5: shows post Axial polydactyly in both hands and feet

Fig 1 shows occipital encephalocele.

Fig 3

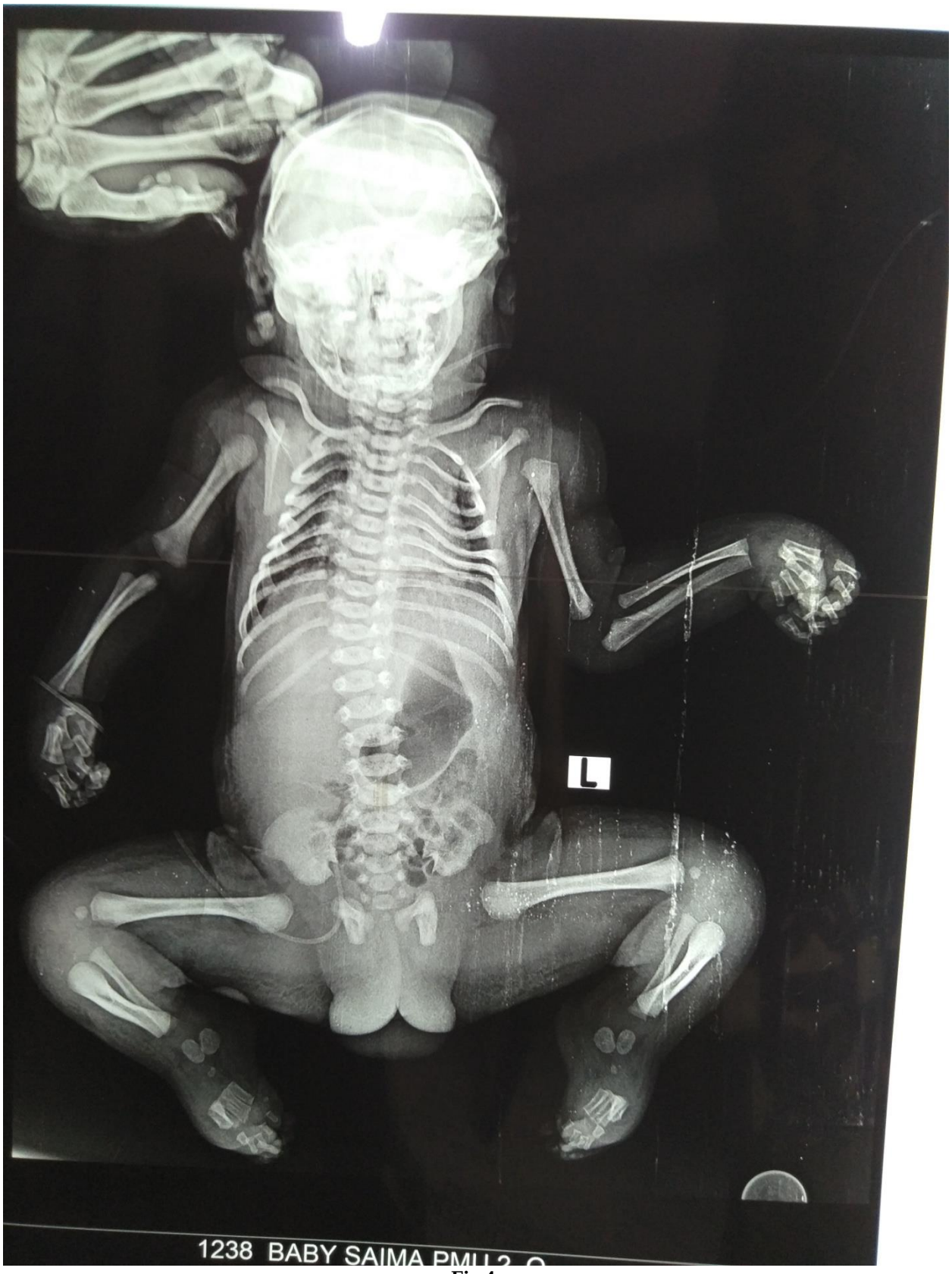


Fig 4

DISCUSSION:

MGS is a very rare syndrome, reported only 1-2 cases in last 3-4 decades in Pakistan (3). MGS including anomalies of the central nervous system (CNS), cystic dysplasia of the kidneys, and malformations of the extremities. Other anomalies associated with MGS are intrauterine growth retardation (IUGR), single umbilical artery, cardiovascular defects, cleft palate, several genital abnormalities, and oligohydramnios and hepatic periportal fibrosis (4). Abnormalities like lung hypoplasia and club foot are secondary to oligohydramnios. (5,6)

The differential diagnosis of MGS includes trisomy 13, trisomy 18, Joubert syndrome, Bardet–Biedl syndrome and Smith–Lemli– Opitz syndrome (7). Trisomy 13, Patau synrome is the most likely syndrome to be confused with MGS. Enlarged kidneys,oligohydramnios and the presence of an occipital encephalocele favours the diagnosis of MGS, whereas holoprosencephaly or other midline CNS anomalies favors trisomy 13 (8).

MGS is best diagnosed prenatally by ultrasonography early in the second trimester. No specific biochemical and chromosomal studies indicate the presence of the MGS. Therefore, the prenatal ultrasonography detection of MGS is important for the diagnosis, which can be confirmed later by genetic analysis. Clinical diagnosis is suggested on the basis of the presence of classical clinical features and when the syndrome recurs in subsequent pregnancies (9,10).

prenatal diagnosis is important for the counselling of the parents regarding the poor foetal prognosis and to explain the chances of recurrence in subsequent pregnancies (9).

MGS effects both genders equally, and consanguinity has been reported to be an important factor in the genetic basis of the disease (1). Although in our case,born to non-consanguineous parents. genetic analysis was not done.prenatal USG findings, the presence of cardinal features and history of recurrence of similar anomalies that were clinically felt to represent MGS were used to establish the diagnosis. MGS is inherited in an autosomal recessive manner so, the chance of giving birth to another child with MGS is 1 in 4 (25%) for each pregnancy (11,12)

CONCLUSION:

MGS is rare syndrome and can be diagnosed during antenatal USG. Our case diagnosed relatively late, born to a non-consanguineous marriage.but alive with classical features involving renal, craniofacial, musculoskeletal and lung malformations. Parents should be counselled about 25% risk of subsequent sibling being affected. One cannot speak about survival of the fetus because of the pulmonary hypoplasia and renal failure.

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