International Journal of Science and Research (IJSR)
ISSN (Online): 2319-7064
Impact Factor (2012): 3.358

Dysencephalia Splanchnocystica

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Abstract: ***Dysencephalia Splanchnocystica also called as Meckel-Gruber syndrome is an autosomal recessive disorder[1]. It is characterized by a traid of occipital encephalocele, large polycystic kidneys, and postaxial polydactyly[2]. The major diagnostic criteria of MGS include at least 2 of these 3 classic manifestations, occipital encephalocele, cystic renal dysplasia, and polydactyly found in 90%, 100% and 83.3% respectively [3]. It is a rare syndrome with highest incidence in Gujarati Indians and Finnish population. We are reporting a case of Dysencephalia Splanchnocystica in a non Gujarati Indian from a municipal town, Sirkazhi in Nagapattinam district in Tamil Nadu, India.***

Keywords: Dysencephalia Splanchnocystica, Occipital encephalomeningocele, Postaxial polydactyly.

1. Clinical Report

A 23-year-old patient, with 32 weeks of gestation, with no antenatal check-ups and previous pregnancy loss at 20weeks of gestation delivered a female baby with multiple congenital malformations. There was second-degree consanguinity. She was not on any teratogenic drugs. The newborn weighed 1500g.

On physical examination the neonate showed postaxial polydactyly in both upper limbs and right lower limb **(fig.1a, f)**, microcephaly, micrognathia, low set ears, deformed pinna **(fig.1b)** microphthalmia, large nose, absent philtrum **(fig.1c)**, natal teeth, folded tongue, complete cleft lip and cleft palate **(fig.1d)** and occipital encephalomeningocele **(fig.1e)**. Ultrasonogram abdomen revealed absent corticomedullary differentiation with no cyst in both kidneys **(fig.2)**. 2 of the 3 classic manifestations of Meckel-Gruber syndrome present in our case. The neonate was delivered as still born. Parents were counselled regarding genetic evaluation for the future pregnancy.

2. Discussion

Dysencephalia Splanchnocystica is an autosomal recessive disorder[1]. It is characterized by a traid of occipital encephalocele, large polycystic kidneys, and postaxial polydactyly[2]. The major diagnostic criteria of MGS include at least 2 of these 3 classic manifestations, occipital encephalocele, cystic renal dysplasia, and polydactyly found in 90%, 100% and 83.3% respectively [3]. It is a rare syndrome with highest incidence in Gujarati Indians and Finnish population.

3. Future Scope of this Study

MKS identified on the basis of first trimester anomaly scan.On the basis of the sonographic findings, the patient should get elected termination of pregnancy, and post-termination following which gene sequencing should be done..mutation in CC2D2A is identified thus providing molecular confirmation of the clinical suspicion of MKS and opening the possibility for future prenatal diagnosis. This case highlights the ability of ultrasound to detect important anomalies in the first trimester, even in low risk situations. It also demonstrates the growing role of new sequencing technologies in fetal testing.

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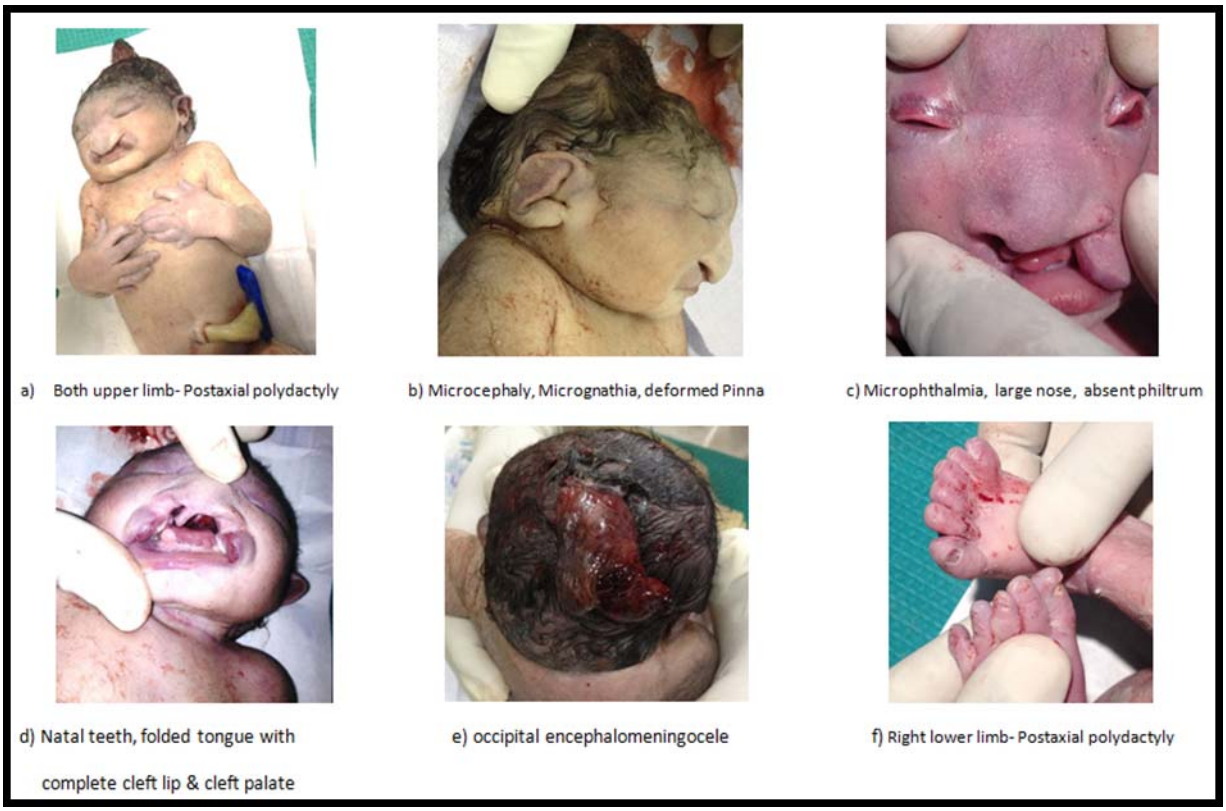

Author Profile

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