IJAM

RESEARCH PAPER ISSN (O) : 2663 - 046X | ISSN (P) : 2663 - 0451

MECKEL-GRUBER SYNDROME

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ABSTRACT Meckel Gruber is an autosomal recessive condition, usually characterised by a triad of Renal Cysytic Dysplasia, Encephalocoele, Post Axial Polydactyly. Hereby enlightening a case of a 22 year old Primi Gravida visiting for regular antenatal check up underwent a USG, which was suggestive of features of Meckel Gruber Syndrome (also known as Dysencephalia Splanchnocystica).

KEY WORDS : Meckel-Gruber syndrome, Renal Cysytic Dysplasia, Encephalocoele, Post Axial Polydactyly, Dysencephalia Splanchnocystica

INTRODUCTION:

Meckel Gruber syndrome is characterised by an autosomal recessive pattern of inheritance, the worldwide incidence varying from 1 in 13,250 to 1,40,000 live births. It can be caused by mutations in one of at least eight genes. The proteins produced from these genes are known or suspected to play a role in development of cilia. Mutations in the eight genes known to be associated with Meckel syndrome account for about 75 percent of all cases of the condition. In the remaining cases, the genetic cause is unknown.

CLINICAL FEATURES:

22 year old Primi Gravida, with history of non-consanguineous marriage, visiting for regular antenatal check up underwent a USG, gestational age being 13 weeks, 4 days. The USG illustrated Occipital Encephalocoele, Acromelic Shortening of Limbs, B/L Polycystic Kidneys, Hypoplastic Vermis, Agenesis of Corpus Callosum. The above mentioned features were suggestive of Meckel-Gruber Syndrome.

Management: Meckel-Gruber Syndrome is a lethal condition with a dreadful outcome. Thus, with the consent of parents, termination of the pregnancy was undertaken.

CONCLUSION:

Early identification followed by early diagnosis is essential. Genetic counselling plays a major role in management. It is imperative to inform the parents that the condition may reoccur in consequent pregnancies, and that no preconceptional diagnosis is available.

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Article History	Received	Accepted	Published
	25/11/2018	13/01/2019	20/03/2019

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