

Picture Story

A case of Meckel Gruber Syndrome

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Introduction

Meckel-Gruber syndrome (MGS), an autosomal recessive condition characterized by posterior encephalocele, cystic dysplasia of kidneys & postaxial polydactyly, was originally described by Meckel in 1822 & later by Gruber in 1934¹. We report an early neonatal death, a product of a non consanguineous marriage, with features of MGS.

Case report

A thirty year old lady was admitted with labour pains at 35 weeks period of amenorrhea. It was a planned pregnancy complicated by a threatened abortion during the first trimester and she was on periconceptional folic acid. Antenatal ultrasound scan was not done to assess fetal anomalies. A dysmorphic female baby weighing 2120g was delivered by an emergency caesarean section and died within an hour of delivery. Post mortem examination revealed posterior encephalocele, microcephaly, short neck, micrognathia, cleft palate, low set malformed ears and bilateral talipes equinovarus deformity (Figure 1). There were bilateral hypoplastic lungs, biliary atresia and bilateral large polycystic kidneys weighing 200g and 180g respectively (Figure 2). Sections of the kidney showed multiple cysts of varying sizes and sections from the liver showed bile ductular proliferation with focal fibrosis. The parents were referred for genetic counselling.

Discussion

MGS (Dysencephalia Splanchnocystica) is a rare autosomal recessive condition, where the locus has been mapped to 17q21-q24¹. It is characterized by prenatal growth retardation, occipital encephalocele, microcephaly with slopping forehead, cleft palate, ear anomaly, microphthalmia, micrognathia, short neck,

post axial polydactyly, dysplastic polycystic kidney, lung hypoplasia, liver fibrosis and biliary atresia¹. Infants with MGS die soon after surviving at the most for a few weeks. Death is usually due to renal defects or nervous system abnormalities¹.



Figure 1 - External appearance



Figure 2- Large polycystic kidneys

MGS is recognized as a cause of about 5% of neural tube defects with a 25% recurrence risk. It must be differentiated from other neural tube defects (NTD) which are usually polygenic and associated with a recurrence risk of about 5%. Periconceptional folate

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supplementation reduces the risk of non-syndromic NTD but this is not the case with MGS².

Prenatal diagnosis may be possible by an elevated alpha fetoprotein level when there is an encephalocele and ultrasonographic delineating of either the encephalocele or dysplastic enlarged kidney. Prenatal diagnosis causes parents to face a difficult choice between a technically illegal abortion and continuing a non-viable pregnancy³.

Patients seldom survive more than a few days to weeks. Death may be related to severe central nervous system defects and/or renal defects. Parents should be made aware of the 75% chance of a normal conception and 25% risk of MGS².

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