Case Report

iMedPub Journals
http://www.imedpub.com

Medical Case Reports
ISSN 2471-8041

2021
Vol. 7 No. 11: 211

The Exceptional Case of Healthy Precious Infant Surviving with Meckel-Gruber Syndrome Variant

Rabiya Siraj* Rana Rehan, Sadaf Nasir, Bisma Rizwan, Mahum zaidi and Kanchan shadani

Department of Radiology, Liaquat National Hospital, Karachi, Pakistan

Corresponding author:

Rabiya Siraj, Department of Radiology, Liaquat National Hospital, Karachi, Pakistan

rabiya.siraj12@gmail.com

Citation: Siraj R, Rehan R, Nasir S, Rizwan B, zaidi M, et al. (2021) The Exceptional Case of Healthy Precious Infant Surviving with Meckel-Gruber Syndrome Variant. Med Case Rep Vol.7 No.11.211

Abstract

Meckel-Gruber Syndrome (MGS), also known as Dysencephalia Splanchnocystica is a fatal autosomal recessive congenital disorder designated as a triad of Renal Cystic Dysplasia, Occipital Encephalocele and Postaxial Polydactyly. It is correlated with broad spectrum of systemic malformations. Death typically occurs in utero or shortly after birth. The Worldwide incidence fluctuates to occur in 1 in 13,250 to 1, 40,000 live births. Phenotypic Variations are reported as well, in which one could present with any two of the cardinal features of MGS. Here we are presenting an exceptional and rare case of healthy baby who presented to us with occipital encephalocele and unilateral multicystic dysplastic kidney considered to have a variant of Meckel-Gruber syndrome.

Keywords: Meckel-Gruber Syndrome; Congenital disorder; Phenotypic variations

Received: November 15, 2021; **Accepted:** December 06, 2021; **Published:** December 15, 2021

Introduction

Meckel-Gruber Syndrome (MGS) is a congenital autosomal recessive disease which proved to be fatal due to its severe multisystem manifestations [1]. Its major characteristics triad consists of Hepatorenal Fibrocystic changes, Central Nervous System malformations (usually encephalocele) and Polydactyly [1,2]. Prenatal diagnosis is frequently approachable by detecting two striking features of MGS i.e. polycystic dysplastic enlarged kidneys and encephalocele [2]. But prenatal or perinatal demise is common. The worldwide incidence fluctuates from 1: 1300 to 140,000 births; however, higher prevalence may be present in Finland, Belgium and in some parts of India [3,4]. Clinical and genetic heterogeneity was previously reported in MGS, but still considered to be very rare as far as its prevalence is concerned. However the survival of variants is possible on contrary with lethal primary disease.

Case Report

A one day old infant presented to us in radiology department through NICU on 29th September 2021. He was born after gestation of 36 weeks with birth weight of 2.6 kgs and good APGAR score, as the child of multiparous mother. He was vitally stable and labs investigations are unremarkable for any abnormality. His ultrasound kidneys were done which shows enlargement of left kidney with loss of corticomedullary parenchyma. It measures about 7.9 cm (LS). Multiple cysts of varying sizes which are not communicating with each other are seen invading and involving whole of cortex and medullary parenchyma. Right kidney appears unremarkable for any pathology. Findings are most likely due to Unilateral Polycystic/Multicystic Dysplastic Kidney (**Figures 1 and 2**).

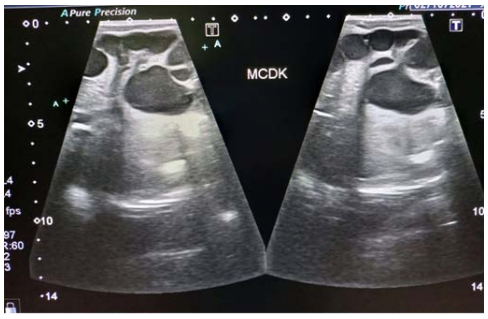

Figure 1 Ultrasound image of an enlarged left kidney showing multiple cysts of varying sizes with dysplastic cortex and medulla representing Multicystic Dysplastic Kidney.

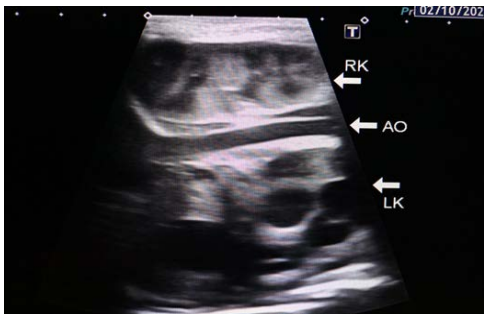

Figure 2 Ultrasound image showing midline aorta with right sided normal kidney and left sided multicystic kidney.

Then on examination, there was also swelling noticed around occipital region, for which MRI Brain and Spine was done. MRI revealed a subcutaneous oval shaped cystic area measuring about 1.5 × 3.2 × 3.4 cms showing CSF signals is seen protruding through occipital region in midline with its neck at the level of external occipital protuberance. There is a small defect in posterior fossa in midline with no convincing communication with the cyst. The track is interrupted by a streak of low signal likely representing fibrous band. So we conclude it as an atretic Occipital encephalocele with possible in utero fibrosis of the calvarial defect. No evidence of hydrocephalus or other brain abnormality noted. The whole spine is normally aligned. No evidence of spina bifida or tethered cord at any level. On further examination, no polydactyly is noted in both hand and foot. On ultrasound examination of liver, no evident hepatic fibrosis is noted (**Figures 3 and 4**).

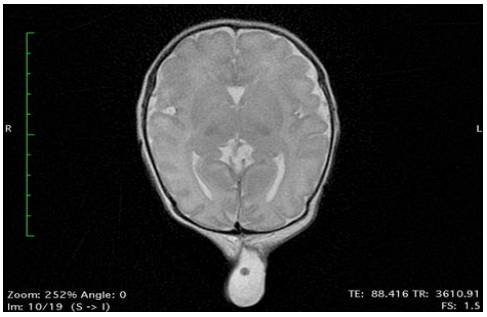

Figure 3 Axial Image of MRI Brain showing posterior defect in occipital region associated with atretic encephalocele.

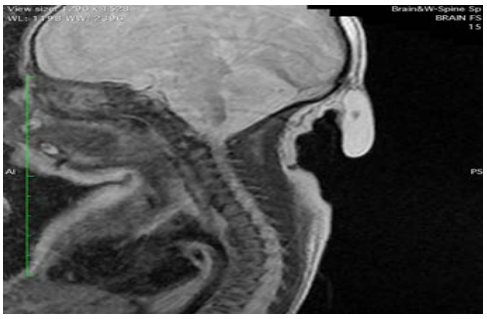

Figure 4 Sagittal Image of MRI Brain and spine showing posterior fibrotic occipital encephalocele with the rest of normal spine.

Per operatively 4*5 cm, Encephalocele is visualized with clear fluid. Small bony defect is identified through with cyst was communicating with it. Small rounded nodular node identified within the cavity which further came out to be angiomatous lesion on histopathology. Well repair done successfully.

Baby is now doing well and discharged home after repairing of occipital encephalocele and reminded for further follow up.

Discussion

Meckel-Gruber Syndrome (MGS), also known as Dysencephalia Splanchnocystica is a lethal autosomal recessive disease which was first narrated by Johann Friedrich Meckel in 1822 and later by Gruber [2,5]. It is the one of the rarest and severe ciliopathy which carries a worldwide incidence of 1: 1300 in Gujarati Indians to 1: 140,000 in England. Prenatal diagnosis is possible in second and third trimester by identifying large polycystic kidneys and occipital encephalocele in concomitant prenatal ultrasound scans [2-4]. But prenatal demise is common.

MGS is actually caused by several mutations in gene encoding proteins that are basic and functional constituent of the primary cilium [6]. It is labelled as the most severe ciliopathy of whole group of disorders and considered as heterogeneous group of disease with severe multisystem manifestations and contained twelve different mutations (designated MKS1 through MKS12) [3,7].

MGS is characterized by the debilitating triad of occipital encephalocele, postaxial polydactyly and polycystic kidneys [2,7,8]. An individual basically born with normal karyotype who at least presents with any two of the three characteristics feature. Most of them reported with sudden demise in utero or mother herself decide to end her pregnancy due to its 100 percent postnatal mortality [3]. The major causes of death are found to be either pulmonary hypoplasia or liver disease.

M Kaplan presented his case report in 1993, in which he highlighted the case of variant of MGS who survived postnatally even after presenting with two characteristics feature that are occipital encephalocele and unilateral multicystic dysplastic kidney. The infant showed an excellent prognosis after repairing of encephalocele and normal kidney function of unaffected kidney (Just as similar to our case) [8].

Another studies quoted in 1990, where five Bedouin sibs diagnosed with MGS with phenotypic variabilities and different abnormalities in which each of them presented with only two of the three manifestation of primary disease occipital encephalocele and Polycystic kidneys, lacking polydactyly (similar to our case) [9,10].

Talking about some cardinal features of MGS, first and foremost that always came across is Polycystic/multicystic dysplastic kidneys. It can be unilateral or bilateral. Kidney shows enlargement of size with multiple cysts of variable sizes that actually replaced off whole of the kidney parenchyma [11]. Mostly it shows association with hepatic duct dysplasia and fibrosis just as similar in cases of autosomal recessive polycystic kidney disease [12]. Second we should talk about CNS malformation, in which posterior encephalocele is very common which may be associated with other various CNS related abnormalities like aplasia of cerebellum, agenesis of corpus callosum, Dandy-walker cyst, holoprosencephaly, microcephaly, ventriculomegaly, Spinal dysraphism, Joubert syndrome and long list of other CNS related abnormalities. Preaxial or postaxial polydactyly is another cardinal feature of the syndrome, along with cleft lip palate, omphalocele,

cardiac abnormalities, hypertelorism and many more [2,13-16].

Conclusion

The primary purpose of presenting this rare case is to highlight the one of the variant of Meckel Gruber Syndrome who survived and kept healthy even after carrying the two main cardinal features of syndrome. As we already discussed that genetic heterogeneity and phenotypic variability can be present in this syndrome. So proper evaluation should be necessary prior to decide about the termination of pregnancy or counselling the mother about life sustaining medical treatment who already delivered.

Acknowledgement

None

Conflict of Interest

None

References

1. Paavola P, Salonen R, Baumer A, Schinzel A, Boyd PA, et al. (1997) Clinical and genetic heterogeneity in Meckel syndrome. Hum Genet 101: 88–92.
2. Velasco MA (2009) Meckel Gruber Syndrome. Ultra. Med and Bio 35:8.
3. Adetola F, Anthony LJ, Rachael OO, Bradshaw J (2018) Obstetric Imaging: Fetal Diagnosis and Care (Second Edition). 559-562.e1.
4. Alexiev BA, Lin X, Sun CC, Brenner DS (2006) Meckel-Gruber syndrome: pathologic manifestations, minimal diagnostic criteria and differential diagnosis. Arch Pathol Lab Med 130:1236–8.
5. Gruber G (1934) Contributions to the question of “coupled” malformations (acrocephalo-syndactyly and dysencephalia splanchnocystica). Beitr Pathol Anat 93:459–76.
6. Hartill V, Szymanska K, Sharif SM, Wheway G, Johnson CA (2017) Meckel–Gruber Syndrome: An Update on Diagnosis, Clinical Management and Research Advances. front. Pediatr 5:244.
7. Darouich SI, Bettaieb M, Khila T, Kitova D, Chelli E, et al. (2015) Phenotypic variability in Meckel-Gruber syndrome. Cilia 5 4(Suppl 1):P1.
8. Connor AKO, Woodford LMG (2016) Polycystic Kidney Diseases and Other Hepatorenal Fibrocystic Diseases, Kidney Development, Disease, Repair and Regeneration p241-264.
9. Richards NQ, Couser NL, Pande C (2019) Genetic Abnormalities with Anophthalmia, Microphthalmia, and Colobomas. Ophthalmic Genetic Diseases p41-52.
10. Farag TI, Usha R, Uma R, Mady SA, Al-Nagdy K, et al. (1990) Phenotypic variability in Meckel–Gruber syndrome. Clin Genet 38(3):176-9.
11. Kaplan M, Ben-Neriah Z, Achiron R (1993) Survival in an infant with a prenatally diagnosed Meckel syndrome variant. Am J Perinatol 10(2):172-4.
12. Puliyanda DP (2013) Cystic Diseases of the Kidney Angela Sun. Emery and Rimoin's Principles and Practice of Medical Genetics p1-33.
13. Kollias SS (2014) Dandy–Walker Syndrome. Encyclopedia of the Neurological Sciences (Second Edition) p935-941.
14. Blaas HGK (2018) Fetal Diagnosis and Care (Second Edition). Holoprosencephal p190-204.e1.
15. Jenkins D, Beales PL (2013) Genes and Mechanisms in Human Ciliopathies. Emery and Rimoin's Principles and Practice of Medical Genetics p1-36.
16. Yanoff M, Sassani JW (2015) Congenital Anomalies. Ocular Pathology (Seventh Edition) p29-52.e4.