Goldston Syndrome: A Rare Case Report with a New Variant?

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Abstract: Goldston Syndrome is a rare entity describing the association of polycystic kidneys and Dandy-Walker malformation with autosomal recessive inheritance. Here, we are presenting a very rare case report of a 3 day old male living neonate born to consanguineously married couple as a 2nd baby with a large head, delayed cry & decreased activity, but clinically stable baby. On evaluation it is found to have Dandy-Walker malformation, bilaterally enlarged kidneys with small cortices and antenatal scan showed polyhydramnios as against oligohydramnios which is usually seen in Goldston syndrome. Such living neonates need to undergo further genetic studies to confirm whether it is a part of Goldston syndrome or any other new variant?

Keywords: Goldston Syndrome, Dandy-walker malformation, Polycystic kidneys, genetic studies, new variant.

1. Introduction

Goldston syndrome, also known as cerebro-renal syndrome which was first described by Goldston in the year 1963, is a rare disorder with few case reports further seen in the literature. It is characterized by renal dysplasia and Dandy-Walker malformation with oligohydramnios. The aim of our rare case report is to present a 3 day old living neonate with Dandy-Walker malformation, polycystic kidneys along with polyhydramnios in the mother as against oligohydramnios seen in previous case reports.

2. Case Report

A 3 day old male baby, 2nd in birth order, born by normal vaginal delivery with good Apgar scores to a 3rd degree consanguineous parentage was referred from a private hospital to our pediatric department, Guntur medical college, Guntur with the clinical presentation to us as delayed cry, decreased activity & abnormal head size for which the neonate was admitted to NICU and evaluated further. Parents informed us that the baby was taking feeds, passed meconium & urine. Mother had an uneventful antenatal hospital to our pediatric department, Guntur medical college, Guntur with the clinical presentation to us as delayed cry, decreased activity, but clinically stable baby. On evaluation it is found to have Dandy-Walker malformation, bilaterally enlarged kidneys with small cortices and antenatal scan showed polyhydramnios as against oligohydramnios which is usually seen in Goldston syndrome. Such living neonates need to undergo further genetic studies to confirm whether it is a part of Goldston syndrome or any other new variant?

3. Discussion

The association of renal syndromes with central nervous system abnormalities is reported as a rare entity. The first syndrome that had been described of this combination was Meckel-Gruber syndrome and also has been described as the prototype syndrome and comprised of several serious anomalies such as occipital meningo-encephalocele, ocular anomalies, cleft palate, polydactyly, cystic kidneys, pseudohemaphroditism and other malformations which are incompatible with life. Approximately 1/5th of patients with Meckel’s syndrome have hepatic anomalies as hepatic fibrosis. Another syndrome associated with Dandy-Walker malformation, cystic dysplastic renal lesions along with congenital hepatic fibrosis is Miranda syndrome, which is a rare familial disorder. Goldston syndrome is a distinct entity with a combination of cystic dysplastic kidneys and Dandy-Walker malformation.

There have been few reported cases of Goldston syndrome in the available literature and all of them were diagnosed antenatally with all the reported cases had evidence of oligohydramnios and to our knowledge, probably none of them survived. Our living neonate is probably the first possible surviving baby of Goldston syndrome diagnosed postnatally. Another distinguishing feature is that our neonate had sonographic evidence of polyhydramnios as against oligohydramnios which is usually seen in the Goldston syndrome. The cause for polyhydramnios in our case report could be due to fetal swallowing difficulty, secondary to mechanical compression of vagus nerve either by posterior
fossa cyst or enlarged 4th ventricle.12 There are no case reports of Goldston syndrome associated with polyhydramnios as per the available literature.

Furthermore, genetic studies couldn’t be done in our case and they should have been done to further enlighten us whether it is a Goldston syndrome or a new variant of it?

4. Conclusion

The objective of this rare case report is to identify such cases antenatally and to evaluate further for any associated anomalies in Goldston syndrome and also genetic studies need to be carried out postnatally in such living neonates so as to detect any new variants and to manage & prognosticate them accordingly.

References


Clinical Photograph of Our Case Report With Goldston Syndrome

AT BIRTH NOW AT 8 MONTHS OF AGE

MRI BRAIN AXIAL T1W&T2W IMAGES OF POSTERIOR FOSSA SHOWING DANDY-WALKER MALFORMATION