

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

Dr. A Thirupathi Reddy<sup>1</sup>, Dr. K. Vani Bai<sup>2</sup>, Dr. Jhansi Rani<sup>3</sup>

<sup>1, 2, 3</sup>Associate Professor of Pediatrics, Guntur Medical College, Guntur, India

**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 2<sup>nd</sup> 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<sup>1</sup> in the year 1963, is a rare disorder with few case reports further seen in the literature<sup>2</sup>. It is characterized by renal dysplasia and Dandy-Walker malformation with oligohydramnios.<sup>3,4,5</sup> 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 course except for polyhydriomnios detected by ultrasound done in 3<sup>rd</sup> trimester of pregnancy. No history of previous abortions or any other congenital abnormalities that had been reported in the family. Elderly sibling is a 4 year old healthy living female child.

Examination of this 3<sup>rd</sup> day old neonate who was crying, irritable but relatively active with slightly diminished neonatal primitive reflexes showed wide fontanelle, sutural diastasis with low set ears and with no other major external congenital malformations. Head circumference was 38cm, length 52.5cm, weight 3 kg and chest circumference was 33cm. The baby had stable vital signs including normal blood pressure. On abdominal palpation, there were bilaterally symmetrical palpable renal masses with all other systems being normal.

On Investigation, the routine laboratory tests including liver & kidney function tests were all normal. Ultra sound abdomen showed bilateral enlarged echogenic kidneys with small cortices. MRI Brain imaging revealed a large midline

posterior cranial fossa cyst communicating with the 4<sup>th</sup> ventricle, which is prominent & also there is a partial agenesis of corpus callosum suggesting Dandy-Walker malformation.

The possibility of Goldston syndrome was considered and the hospital course of the neonate was uneventful and discharged on 16<sup>th</sup> day of life in stable condition. He is now 8 months old with no significant developmental delay and advised to be on close follow-up. We couldn't do genetic studies to further establish whether it is a classical 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.<sup>2,6</sup> 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, pseudohermaphroditism and other malformations which are incompatible with life. Approximately 1/5<sup>th</sup> of patients with Meckel's syndrome have hepatic anomalies as hepatic fibrosis.<sup>6</sup> 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.<sup>7</sup> Goldston syndrome is a distinct entity with a combination of cystic dysplastic kidneys and Dandy-Walker malformation.<sup>1,3,4</sup>

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<sup>2,8,9,10,11</sup> 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 antenatal scan done at 3<sup>rd</sup> trimester of pregnancy in 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 4<sup>th</sup> ventricle.<sup>12</sup> 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

- [1] Goldston AS, Burke EC, D'Agostino A, Maccaughey WT. Neonatal polycystic kidney with brain defect. *Am J Dis Child* 1963; 106: 484-8.
- [2] Hussain Z, Masroor I, Haider QU et al. Goldston Syndrome. *J Coll Physicians Surg Pak* 2011; 21(4): 242-4.
- [3] Glucan YH, Duman N, Kumral A, Sagol O, Lebe B, Kavukcu S et al. Goldston syndrome: report of a case. *Genet Couns* 2001; 12: 263-7.
- [4] Menon RK, Nadkarni TD, Desai KI et al. Dandy-Walker malformation associated with polycystic kidney: Goldston syndrome Revisited. *J Clin Neurosci* 2006; 13(8): 875-877
- [5] Mesfer S Al Shahrani. Antenatal diagnosis of a rare fetal syndrome. Is this a new syndrome or a variant of Goldston Syndrome? *Bahrain Medical Bulletin* 2012 jun; 34(2): 1-5
- [6] Bernstein J, Brough AJ, McAdams AJ. The renal lesion in syndromes of multiple congenital malformations. Cerebro-hepato-renal syndrome; Jeune asphyxiating
- [7] thoracic dystrophy; tuberous sclerosis; Meckel syndrome. *Birth Defects Orig Artic Ser* 1974; 10; 35-43
- [8] Kudo M, Tamura K, Fuse Y. Cystic dysplastic kidneys associated with Dandy Walker malformation and congenital hepatic fibrosis: Report of two cases. *Am J Clin Pathol* 1985; 4: 459-63.
- [9] Gloeb DJ, Valdes-Dapena M, Salman F et al. The Goldston syndrome: Report of a case. *Pediatr Pathol* 1989; 9(3): 337-43.
- [10] Avou S, Akdeniz H, Unal O, et al. Goldston Syndrome in a fetus: Case Report and Literature Review. *Fetal Pediatr Pathol* 2010; 29(5): 353-8.
- [11] Hunter, Jimnez, Tawagi. Familial renal-hepatic-pancreatic dysplasia and Dandy-Walker syndrome? *Am J Med Genet.* 1991; 41: 201-207
- [12] Walpole, Goldblatt, Hockey, Knowles S. Dandy-Walker malformation (variant), cystic dysplastic kidneys and hepatic fibrosis : a distinct entity or Meckle syndrome? *Am J Med. Genet* 1991; 39: 294-298
- [13] George C Newman, Anthony I, Buschi, Normank Sugg, Thaddeus E Kelly and James Q Mille. Dandy Walker Syndrome diagnosed in utero by ultrasonography. *Neurology (Ny)* 1982; 32: 180-4.

#### Clinical Photograph of Ours 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

