International Journal of Medical and Health Sciences

Journal Home Page: http://www.ijmhs.net ISSN:2277-4505

Case Report

Fused multicystic kidneys with Persistent Cloaca – Case report

A.K.Manicka vasuki[1*], T.K.Aleyamma fenn[2], M.Nirmaladevi[3], A.Pavai[4], Deborah joy hepzibah[5]

[1&5]Assistant professors, [2]Former Professor & HOD, [3]Associate professor Department of Anatomy, PSG Institute of Medical Sciences and Research, Coimbatore,

[4]Professor & HOD, Department of Pediatric surgery, PSG Institute of Medical Sciences and Research, Coimbatore.

ABSTRACT

Renal anomaly with Urorectal septum malformation (URSM) with persistent cloaca is a very rare congenital anomaly. During dissection of 22 weeks old spontaneously aborted male fetus in the department of Anatomy, We found fused multicystic kidney, ureter anomaly, Persistent cloaca with Urorectal septum malformation. This is due to defective caudal end of mesoderm.

KEYWORDS: Fused multicystic kidneys – ureter anomaly – cloaca – caudal mesoderm – Urorectal septum malformation (URSM)

INTRODUCTION

A very rare congenital anomaly – Fused multicystic kidneys with Urorectal malformation was observed. This presentation has never been reported in the available Journals. Various structures of genitourinary system and lower part of GIT develop from caudal end of mesoderm. Defective caudal end of mesoderm leads to spectrum of malformations which are called Urorectal septum malformation (URSM). This consists of the absence of the perineal and anal openings in association with ambiguous genitalia, urogenital and lumbosacral anomalies[1] .The incidence of URSM sequence is one in 50,000 to 2, 50,000 in neonates. Urorectal septum malformation sequence is associated with many other anomalies and produces many syndromes. Commonest syndrome associated with this is the VACTERL syndrome. This constitutes Vertebral, Anorectal, Cardiac, Thoraco – esophageal, Renal and Limb anomalies[2].

The development of Kidney begins in the fourth week of intrauterine life from intraembryonic mesoderm. Nephrogenic cord derived from intermediate mesoderm forms a longitudinal ridge on posterior abdominal wall on each side of the dorsal aorta. The Neprogenic cord forms three successive kidneys: Pronephros, Mesonephros and Metanephros – succeeding each other in time and space. The last to develop retained as permanent Kidney.

The Pronephros forms at the beginning of fourth week in the cervical region. It is nonfunctional and completely regresses. The Mesonephros forms at the end of fourth week in thoracolumbar region. It is functional for short period and completely regresses. Metanephros forms at the beginning of third month in the sacral region. It persists permanently in humans. It drains into Ureter. Each Kidney develops from two distinct sources: 1.Metanephros, 2.Ureteric bud. Metanephros forms Secretary System and the Ureteric bud which arises from Mesonephric duct, forms collecting system of Kidney. Later communication between the two systems takes place[3].

During development, ventrally the hindgut communicates with the Allantoic diverticulum. This diverticulum divides the hindgut into preallantoic and postallantoic parts. The preallantoic part communicates with the midgut. The postallantoic part forms a dilated sac known as Endodermal cloaca. By coronally oriented partition, the urorectal septum, cloaca is divided into ventral part – Urogenital sinus and dorsal part – Anorectal canal. The division of cloaca begins to divide by 4th week and is completed by 6th week when urorectal septum fuses with the cloacal membrane by fusion of one vertical fold of Tourneux projecting caudally from the ventral wall of hindgut just cephalic to allantoic canal and two lateral folds of Rathke projecting into the Endodermal cloaca. Fusion of one vertical fold of Tourneux and two lateral folds of Rathke forms complete Urorectal

septum. Each lateral fold contains in its interior a mesonephric and paramesonephric ducts[4].

CASE REPORT

During dissection of 22 weeks old male fetus in the department of Anatomy, PSG Institute of Medical Sciences & Research, Coimbatore, a rare Congenital anomaly – Fused multicystic kidney with urethra anomaly with Persistant cloaca with incomplete urorectal sepal malformation with Anorectal anomaly was observed and studied in detail. Prenatal ultrasound of the fetus showed Left renal agenesis with right multicystic, dysplastic kidney.

Figure: 1 Fused, multicystic kidneys and fused lower ends of ureters and then they separate and not opened into the cloaca

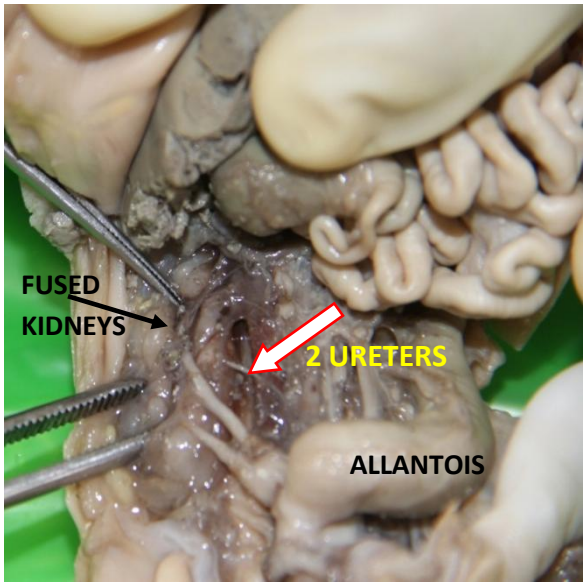
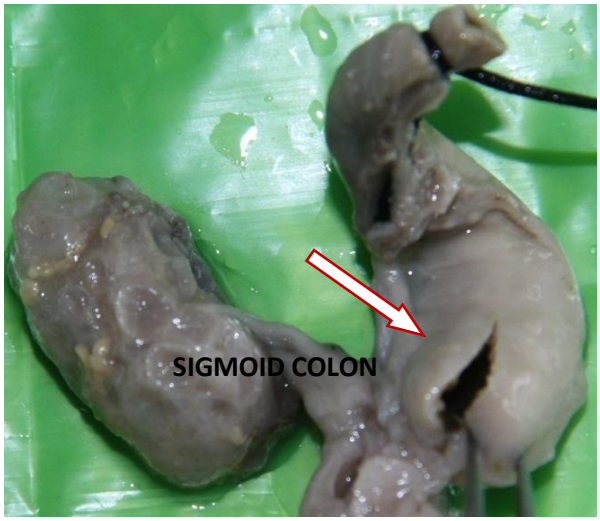


Figure: 2Dilated sigmoid colon Without development of Rectum



OBSERVATIONS

Fused, multicystic kidneys present on the right side of the abdomen. The lower ends of two ureters were fused to form a mass and then they separate and end near the upper part of cloaca posteriorly (Not opened into the cloaca).

Allantois attached to cloaca anteriorly. Dilated sigmoid colon filled with meconium and fused with the upper part of cloaca. Rectum – not developed. Urorectal septum is found to be developing (three parts of it, coronal septum and two lateral folds were seen). No anal orifice or Proctodeum was present.

Figure 3: Developing Urorectal spectrum

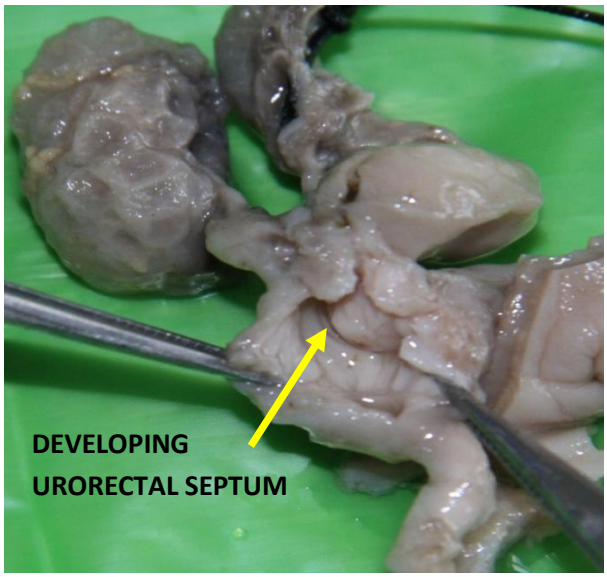
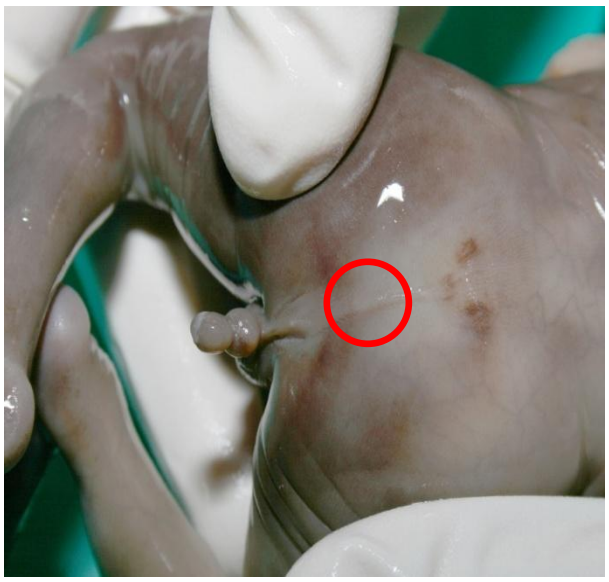


Figure 4: No anal orifice



DISCUSSION

Sunita Arvind Athavale[1] described OEIS complex in 20 weeks fetus. She observed Ompholocele, Exstrophy of bladder; imperforate anus and Spinal defects were due to caudal mesoderm defects. This does not fit into urorectal malformation sequence. She made hypothesis suggested that a local internal environmental imbalance at the site of implantation can cause nutritional insult to the embryo during Gastrulation during the third and early fourth week of embryonic life.

Soumya pathra and radheshyan purkai [2] found URSM sequence in 34 weeks fetus with VACTERL association. The fetus had congenital Varus deformity of lower limbs (may be due to vertebral defect), anal atresia, cardiac defect and tracheoesophageal fistula, bilateral hydronephrosis and polydactyly – features of VACTERL association. The patient also had absent perineal and anal opening with ambiguous genitalia which were under URSM sequence.

The term 'Urorectal septum malformation sequence' was coined by Escobar et al [5] 1987 to describe a combination of anomalies arising from failure of migration of Urorectal septum and/or fusion of urorectal septum with the cloacal membrane. Anomalies include absent perineal and anal openings, Ambiguous genitalia, urogenital, colonic and lumbosacral anomalies. Turkmen M, Inan G et al[6]found urorectal septum malformation sequence in a 32 weeks old fetus that died because of Oligohydramnios and pulmonary hypoplasia. Other findings in that fetus were Potter face, lower limb deformities, no urethral and perineal openings, lower gastrointestinal anomalies, bilateral renal agenesis and female pseudohermaphroditism. These anomalies are due to abnormalities of cloacal septation.

According to Deepali Jain, Mehar C.Sharma[7] et al, Urorectal septum malformation sequence (URSM) was identified in seven cases.They stated that absent perineal and anal openings with ambiguous genitalia as 'Full URSM sequence' and a single perineal or anal opening draining a common cloaca with an imperforate anus as 'Partial URSM sequence'. They found colon, ureters and uterus were opened into common cloaca. They found association of external genitalia, renal and sacral abnormalities. They suggested that the full URSM sequence develops due to complete lack of breakdown of the cloacal membrane with a deficiency in urorectal septal development. In partial URSM sequence, partial breakdown of the cloacal membrane leads to the presence of a single perineal/ anal opening. Lack of caudal mesoderm formation contributes to association of other abnormalities.

Patricia G.Wheeler and David D.Weaver[8]identified twenty five cases of partial urorectal septum malformation sequences. Associated vertebral, renal, cardiac, CNS anomalies were found to be due to Persistant cloaca. They encountered URSM disorders are secondary to caudal mesodermal and endodermal deficiencies, abnormal development of urorectal septum and/or lack of cloacal membrane breakdown.

Shazia sharif, Martyn Williams et al[9] encountered Wenstrup syndrome in 36 weeks old fetus. The syndrome comprises of Microphthalmia, absent sacrum, vertebral abnormalities, extra ribs, truncus arteriosus, ventricular septal defect, abdominal muscle hypoplasia/ aplasia, Meckel's diverticulum, tracheoesophageal fistula, ambiguous/ absent genitalia, female pseudohermaphroditism, absent/ malformed uterus, vaginal atresia, large bladder, renal abnormalities, absent/hypoplastic thumbs and club foot. This is similar to URSM along with many other abnormalities. Shreeprasad P.Patankar, Vijay Kalrao[10] et al found renal, skeletal, spine abnormalities with anorectal malformation along with vaginal atresia due to Mullerian agenesis in 7 years old female child and MRKH syndrome (Mayer Rokitansky Kuster Houser syndrome). Primary amenorrhea is the commonest presentation. Urorectal septum formation and division are normal. Amongst Anorectal malformation, Mullerian duct agenesis is frequently found common cloacal anomaly.

Anas M.Ajazami, Ranad Shaheen et al [11] reported a second – cousin consanguinous family in which four siblings had bifid nose associated with anorectal and renal abnormalities. This autosomal recessive form of disorder is due to FREM 1 mutation. Riaz Ahmad[12] reported Thrombocytopenia, absent radius with Crossed fused renal ectopia in 24 years old female who admitted with pelvic fracture. TAR syndrome association (Thrombocytopenia, absent radius with presence of Thumbs) was found in crossed fused renal ectopia. Gupta, Pankaj; Goel, Sandeep et al[13] diagnosed crossed renal ectopia with dysplastic ectopic kidney in 20 weeks old antenatal scan. Ultrasound showed cystic mass in the right flank in relation to right kidney. The left kidney was not found in the left flank. Adhan Narci Mevlit Korkmaz et al[14]reported left to right Crossed multicystic dysplastic ectopic kidney associated with right severe hydronephrosis due to right ureterocele in 5 months old infant.

But in the present case there was crossed fused kidneys were found on the right side of abdomen(fig.1). Lower end of ureters were fusing on the posterior part of cloaca, where Rectum has to develop(fig.2). A developing Urorectal septum was seen(fig.3). Sigmoid colon was fusing with the vertical fold of urorectal septum. No anal orifice was seen(fig.4). Urethral opening was seen in the lower end of cloaca. This type of anomaly partially fits into Partial Urorectal septum malformation sequence but fused kidneys and ureteral anomaly cannot be explained. This type of presentation is not reported in any Journals and this anomaly is not associated with any syndromes.

CONCLUSION

The full Urorectal septum Malformation sequence is incompatible with life; however the partial sequence allows for survival if timely urologic/ pediatric surgical interventions are taken. It is important for the Radiologists,

Urologists and Pediatric surgeons to be familiar with and have an understanding of mechanisms and presentations of this condition as it may be useful in the diagnosis and treatment of such condition.

REFERENCES

1. Sunita Arvind Athavale.Deciphering caudal embryonic defects: embryologic analysis and reviewing literature.Rom J Morphil Embryology 2012;53(4)

2.Soumya Pathra,Purkait.URSM sequence in a newborn with VACTERAL association.J.College of Physicians and Surgeons2012;22(2):116 – 17:1013 - 1020 3.Vishram singh. Development of Urinary system. In: Textbook of Clinical Embryology.Vishram singh,editor. 1st edition .Chennai:Elsevier,A division of Reed Elsevier India Private Limited. Olympus Premedia Pvt.Ltd.2013.pp233 - 240

4. Asim Kumar Datta. The Urogenital system.In: Essentials of Human Embryology. Asim Kumar Datta.editor.6th edition. Kolkatta: The Indian Press Pvt.Ltd.2010.pp210 - 211

5.Escobar LF,Weaver DD et al.URSM sequence.Report of six cases and embryological analysis.Am J Dis Child 1987;141:1021 – 1024

6.Turkman M, Inan G et al.Urorectal septum malformation sequence.A case report.East.J.Medicine 2002:7(1):19 - 21

7.Deepali Jain, Mehar C.Sharma et al.URSM sequence: A report of seven cases.Congenital anomalies Dec 2008;(48):174 – 179

8.Patricia G.Wheeler,David D Weaver.Partial URSM sequence, a report of 25 cases.Am J Med Genetics 2001;103:99 - 105

9. Shazia shant, Martyn Williams et al. Wenstrup syndrome: An unusual presentation of a rare condition.J.Pediatric surgical specialities Dec 2013; Vol 7, No.4,1 -52

10. Shreeprasad P.Patankar, Vijay Kalrao et al. Mayer – Rokitansky syndrome and Anorectal malformation. I.J.Pediatrics Dec 2004; Vol.71

11.Anas M.Ajazami, Ranad Shaheen et al. Frem 1 mutations cause bifid nose, Renal agenesis and Anorectal malformations syndrome. Am J Hum Genet Sep 2009; 85(3):414 – 18

12. Riaz Ahamad.A rare association of Crossed fused ectopia. BMC Nephrology 2007; 8:5

13. Gupta, Pankaj; Goel, Sandeep; et al. Case reports: Crossed fused ectopia with multicystic renal dysplasia. BJUI 2011

14. Adnan Narci, Mevlit Kormaz. Cross ectopic multicystic dysplastic kidney with ureterocele in non – ectopic site. Can Urol Asso J Jun 2010; 4(3):E61 – E63

---

*Corresponding author: Dr. A.K.Manicka vasuki
E-Mail: vasukikalyan01@gmail.com