J. Fetal Med. (June 2018) 5:81–91
https://doi.org/10.1007/s40556-018-0157-3

ORIGINAL ARTICLE

Prenatal Diagnosis of Cloacal Anomalies: An Analysis of Pattern of Presentation with Emphasis in Males

Mamatha Gowda[1] · **Keerthana Kothandaraman**[1] · **Monica Thiagarajan**[1] · **Shanthi Paranthaman**[1]

Received: 19 January 2018 / Accepted: 6 February 2018 / Published online: 26 February 2018

Abstract The objectives of the study were to recognize the ultrasound findings and describe the pattern and spectrum of cloacal anomalies in prenatal life with special attention to pattern recognition in males. This study included a retrospective cohort analysis over 3 years. Of the 14 cases suspected prenatally, the diagnosis was confirmed by fetal autopsy in ten and postnatal surgery in two. There were 8 male fetuses, 3 females and 2 in whom sex couldn't be determined and no information on sex was available in 1 case. The major findings on antenatal ultrasound were abdomino-pelvic mass in 6 (43%), ascites in 6 (43%), oligohydramnios in 12 (86%), hydronephrosis in 3 (21%), renal agenesis in 5 (36%), multi-cystic kidney disease in 4 (29%), non-visualization of bladder in 12 (86%), single umbilical artery in 2 (21%), ambiguity of external genitalia in 5 (36%), cardiac defects in 4 (29%), limb defects in 3 (21%) and spine anomalies in 3 (21%). Prenatal diagnosis of cloacal malformation should be suspected in the presence of a pelvic cystic mass with fluid-debris level and is reinforced when renal anomalies, non-visualization of bladder and anhydramnios accompany. More research is needed to focus on the pattern of presentation in male fetuses.

Keywords Cloacal malformations · Persistent cloacal · Genito-urinary malformations · Uro-rectal malformations · Common cloaca

✉ Mamatha Gowda
drmamathagowda@gmail.com

[1] Genetic Clinic, Department of Obstetrics and Gynaecology, Jawaharlal Institute of Postgraduate Medical Education and Research (JIPMER), E-38, JIPMER Campus, Dhanvantri Nagar, Pondicherry 605006, India

Introduction

Cloacal anomalies, characterized by a single perineal opening for urinary, gastrointestinal and reproductive tracts are very rare malformations with an incidence of 1 per 50,000 live births [1]. The diagnosis is often made exclusively in females with limited reports in males [2, 3]. They are usually associated with anomalies of urogenital and gastrointestinal system and may occur as a component of syndromes such as VACTERL. Prognosis is usually poor and depends upon the severity of malformation and extent of associated anomalies. Prenatal diagnosis of cloacal malformations is a challenge as the spectrum of defects vary widely and visualization is compromised by the severe oligohydramnios that usually accompanies. In this study, we present our analysis of prenatally diagnosed cloacal malformations with special emphasis on the pattern in males.

Methods

We retrospectively reviewed the case files, images, cine loops and available data on all cases with prenatal diagnosis of cloacal malformations during the period 2014 through 2017 at Jawaharlal Institute of Postgraduate Medical Education and Research, Pondicherry, India. The ultrasounds were done on Voluson E8 of General Electronics by the author as a single operator. Extensive image and videos of each case were replayed and analysed to reaffirm the ultrasound findings as was reported on the initial scan. The findings from prenatal ultrasound were then compared and correlated with the postnatal autopsy or surgery findings. All the parents were also contacted over telephone to collect any missing data.

Results

There were 14 cases of cloacal malformations during the period of analysis, the diagnosis was confirmed by fetal autopsy in ten and postnatal surgery in two cases. The diagnosis was presumed by external examination combined with ultrasound findings in one case and in another case, no such reinforcement of diagnosis by postnatal evaluation was available. Of the 10 cases with autopsy confirmation, 7 were male fetuses, 2 were females and sex could not be determined in one case. Among the 2 cases confirmed intraoperatively during surgery, one was male and one female. Sex determination by prenatal karyotyping is not legally allowed in India, hence it was not done. Differentiation of male from female sex was done on the basis of postnatal karyotyping in three cases and histopathology of autopsy specimen in eight cases. The demographic details of all cases are provided in Table 1. Information on the number of perineal openings were available in 13 cases. Nine fetuses had a single perineal opening (69%), one had 2 perineal openings (7.7%) and smooth perineum with no opening was seen in three cases (23%).

All cases were diagnosed during the second trimester, the earliest diagnosis was at 17 weeks of gestation. Three had second trimester termination of pregnancy, five had carried the pregnancy till term and five were delivered preterm. The major findings on antenatal ultrasound were, abdomino-pelvic mass in 6 (43%), ascites in 6 (43%), oligohydramnios in 12 (86%), hydronephrosis in 3 (21%), renal agenesis in 5 (36%), multi-cystic kidney disease in 4 (29%), non-visualization of bladder in 12 (86%), single umbilical artery in 2 (21%), ambiguity of external genitalia in 5 (36%), cardiac defects in 4 (29%), limb defects in 3 (21%), spine anomalies in 3 (21%), abnormal thymus in the form of bifid thymus and hypoplastic thymus in 2 (14%). The ultrasound findings are presented in Table 2 and further details of antenatal diagnosis and autopsy or post-natal surgery are presented in the Tables 3 and 4.

Discussion

Cloaca is a transient structure during embryonic development and is formed by the confluence of the allantois and hindgut from the tail fold of the developing embryo at 3 weeks of gestation. By 6 weeks, the cloaca is divided, into urogenital sinus anteriorly and a separate hindgut posteriorly. The urogenital sinus then becomes the urinary bladder and the cloacal membrane ultimately ruptures at 10 weeks. Failure of division of the cloaca or urogenital

Table 1 Demographic information of cases analysed

No.	Age	OBI	Co-morbidities	GA at diagnosis	GA at delivery	Outcome of pregnancy	Outcome for baby	Autopsy/surgery
Case 1	27	G3P1L1A1	None	27	28	Induced	SB	Autopsy done
Case 2	21	G1	None	19	19	Termination	Abortus	Autopsy done
Case 3	24	G1	None	18	28	Induced	SB	Autopsy done
Case 4	30	G3P1A1	None	23	40	Vaginal term	SB	Autopsy done
Case 5	22	G1	None	26	28	–	–	Autopsy not done
Case 6	20	G1	None	17	18	Termination	Abortus	Autopsy done
Case 7	26	G2L1	None	26	37	SVD	SB	Autopsy done
Case 8	23	G1	None	27	29	Induced	MSB	External examination
Case 9	32	G3P2L2	None	23	32	SVD	SB	Autopsy done
Case 10	20	G1	None	34	36	SVD	SB	Autopsy done
Case 11	28	G2P1L1	None	37	37	LSCS	LB, expired on POD2	Surgery done
Case 12	19	G1	None	17	18	Terminated	MSB	Autopsy done
Case 13	24	G1	None	37	37	LSCS	LB, expired in post-op period	Surgery done
Case 14	34	G3P2L2	Type IV PP/seizure disorder	31	32	LSCS	SB, M, 1.2 kg	Autopsy done

Table 2 Various ultrasound signs among the study group

Findings on USG	No. of cases	Percentage (%)
Abdominal/pelvic cystic/mass	6	43
Oligohydramnios	12	86
Ascites	6	43
2 vessel cord	3	21
Bladder not seen	12	86
Absent kidney	5	36
Hydronephrosis	3	21
Multicystic kidney	4	29
Ambiguous genitalia	5	36
Hydrocolpos	2	14
Solid pear shaped projection	2	14
Echogenic bowel	2	14
Abnormal spine	3	21
Cardiac anomalies	4	29
Limb anomalies	3	21
Spine anomalies	3	21
Syndromic	4	29
Elfin right ear	2	21

sinus results in either convergence of the genital, urinary, and intestinal contents into a single common channel (cloaca) or the formation of two channels: a common genitourinary channel (urogenital sinus) separate from the hindgut [1]. Development may be arrested at any stage resulting in a wide spectrum of cloacal anomalies. Defective homeobox and sonic hedgehog signaling or hormonal influence have been implicated as the cause of arrest [9].

The categorization of cloacal anomalies is confusing due the various terminologies used in the literature such as urorectal septum malformation sequence, anorectal malformations etc., as also the wide range of possible connections further complicating the nomenclature [4, 5]. Persistent cloaca is an uncommon and most severe type of congenital anorectal malformation characterized by a single perineal opening for the urinary, gastrointestinal, and reproductive tracts. In cloacal dysgenesis, the genitalia can be either absent or ambiguous and the perineum is smooth and devoid of orifices from the urethra, vagina, or anus [3, 6]. The variants of cloacal anomalies are illustrated along with representative images from the study group in Fig. 1. There were 3 cases with 0 perineal opening, 1 case with 2 openings and 9 cases with a single perineal opening among the 13 cases where information on perineal opening was available.

The overall incidence of persistent cloaca is estimated to be 1 in 50,000 accounting for up to 10% of all anorectal malformations [7]. Most of the published reports on cloacal anomalies are in the postnatal group with limited literature on prenatal findings. It is often described as a diagnosis exclusively made in females and a combination of ultrasound findings, together with the confirmation of female karyotype, has been the basis for prenatal diagnosis of cloacal anomaly [8, 9]. In our study, there were 8 males, 3 females and 1 fetus where sex could not be determined. Among the 8 male fetuses, 7 had well formed penis but undeveloped scrotum and in 1 case, both scrotum and penis were well formed. The presentation was also more severe among the male fetuses evident by gross ascites, obstruction to outflow of both urinary and gastrointestinal tracts, anhydramnios and severe pulmonary hypoplasia. The deviation in the sex predilection seen in our study could be attributed to the fact that these were diagnosed in prenatal period in contrast to most other studies involving postnatal group. It is possible that cloacal anomalies are more severe among males in prenatal life. We propose that the sex of the fetus may not form the basis for diagnosis of a cloacal anomaly in prenatal life, as the defect is not limited only to females but can also occur in males.

Accurate prenatal diagnosis of persistent cloaca is difficult because of the rarity, complexity, extent and severity of multisystem involvement [10]. The literature contains only case reports with description of the anomaly reported as early as 12 weeks and very small series [11–15]. A retrospective review of 489 patients born with cloaca found that the diagnosis was suspected prenatally in only 6% of patients, despite the presence of suspicious sonographic findings in almost 62% on reviewing the prenatal imaging reports [16]. Prenatal diagnosis has implications not only for perinatal management of the fetus but also for parental counseling about complex social consequences. Thus there is a need to improve the pattern recognition for better prenatal diagnosis and management. Presence of 3 common findings on antenatal ultrasound, a cystic pelvic mass, urinary tract abnormalities, and dilated bowel loops should raise the possibility of a persistent cloaca. Serial ultrasound may reveal a sequence of fetal ascites, a bilobed cystic structure with debris arising from the fetal pelvis, poor visualization of bladder, hydronephrosis, oligohydramnios, growth restriction and vertebral anomalies [1]. The most prominent findings on ultrasound in our study group were abdomino-pelvic mass, non-visualisation of bladder, renal anomalies and ascites (Table 2). Few ultrasound images in correlation with the autopsy findings are presented in Fig. 2.

There may be variable degrees of obstruction to drainage at the perineum, although the common outflow channel of cloaca is usually patent [17]. In 50% of cases, there may be no obstruction and no cystic pelvic mass thus making the prenatal diagnosis difficult. Urinary tract malformations are present in 90% of cloacal anomalies and resemble the bladder outlet obstruction with megacystis,

Table 3 Details of antenatal ultrasound findings and additional information on perinatal autopsy or postnatal evaluation

CASE		Kidneys	Bladder	Liquor	GIT	Perineal openings		Pelvic anatomy	Other malformations/findings	Diagnosis
						Uro-genital	Anal			
1	USG	B/L HUN	Not seen	↓↓	–	–	–	Large multiseptate cystic mass in abdomen and pelvis	Severe lung hypoplasia, heart pushed to left	Cloacal malformation
	Autopsy	B/L HDUN	Not seen	–	Common cloaca	Long edematous phallus, absent scrotal sacs	–	Common cloaca receiving ureters and dilated large intestine	Abdominal wall edema, hypoplastic lungs	Common cloaca, SUA
2	USG	Not seen	Not seen	Nil	–	–	–	–	Mild ventriculomegaly, gross ascites.	Renal agenesis
	Autopsy	B/L absent	Not seen	–	Rectum atretic	Penis without scrotal sacs	Imperforate	Persistent UGS with atretic rectum attached posteriorly	–	Persistent UGS, B/L mild ventriculomegaly
3	USG	B/L HDUN	Not seen	↓↓	Small SB	–	–	Huge cyst with septations	Ascites	Cloacal malformation
	Autopsy	B/L HDUN	Not seen	–	Distended Sigmoid	Male type	Imperforate	Common cloaca receiving colon and ureters. Thick mound anterior to colon	–	Common cloaca
4	USG	B/L small	Small bladder	Normal	–	–	–	B/L Cystic structure in pelvis	Septated Cystic hygroma, Large left CDH with herniation of left lobe of liver. HLV with CAT	Cloacal malformation
	Autopsy	Persistent Metanephric ducts	Not seen	–	–	Male	Patent	B/L Large and elongated hollow sacs filled with fluid, persistent UGS with partially formed cavity	Short/wide neck, widely spaced nipples, absent left diaphragm. Stomach, intestines, spleen and left lobe of liver to the left of heart. Hypoplastic LV, CAT	Common cloaca, agenesis of Left Hemidiaphragm, HLV and CAT
5	USG	Seen	Lost contour	↓↓	–	–	–	–	Ascites	Urinoma/Cloacal malformation with ascites
6	USG	B/L renal agenesis	–	Nil	Absent SB	–	–	–	–	Renal agenesis
	Autopsy	B/L absent	Atretic	–	–	Female	Imperforate	Colonic atresia	–	Persistent UGS with colonic atresia

Table 3 continued

CASE		Kidneys	Bladder	Liquor	GIT	Perineal openings		Pelvic anatomy	Other malformations/findings	Diagnosis
						Uro-genital	Anal			
7	USG	Not seen	Not seen	Nil	–	–	–	–	Gross ascites	?Cloacal malformation
	Autopsy	B/L normal	Common cloaca	–	–	Female	Imperforate	Ureters and Ileal loops ending blindly into the cloacal sac	–	Common cloaca
8	USG	B/L small	? Small	Nil	–	–	–	–	Right limb not well seen Gross ascites	Cloacal malformation with right lower limb aplasia of proximal segment
	External exam	–	–	–	–	Ambiguous ? Male	–	–	Dysmorphic facies, right lower limb aplasia of proximal segment	
9	USG	B/L MCDK	Not seen	Nil	–	–	–	Retort shaped cystic mass	VSD, thick long bones	Colon obstruction, CHD
	Autopsy	B/L MCDK	Not seen	–	–	Only Skin tags ? sex	No anogenital opening	–	Sirenomelia, PS VACTERL association	Common cloaca, sirenomelia, PS, VACTERL association
10	USG	L absent R dysplasia	–	Nil	–	–	–	–	TOF Thick placenta	TOF/Renal dysplasia
	Autopsy	L absent R small	–	–	–	Male	Imperforate	Common cloaca	Triangular head, absent right tibia/fibula/great toe, placenta calcified, short-thick cord	Common cloaca, TOF, PS, open spina bifida, SUA
11	USG	B/L not seen	Not seen	Normal	–	Dilated bowel loops	–	Large round pelvic mass with sedimentation/fluid level	Thoracic hypoplasia	Complex pelvic mass
	Surgery	B/L dysplastic	Small	–	–	Female	Imperforate	Ruptured mass with meconium peritonitis	Baby expired after 2 days	Common cloaca with rectal obstruction, dysplastic kidneys
12	USG	B/L dysplastic	Not seen	↓↓	–	–	–	–	AVSD, DORV	AVSD, DORV
	Autopsy	B/L MCDK	Solid mound	–	–	Male with small penile projection	–	Common cloaca, large bowel dilated, rectum short, atretic and blind ending	Low set ears, B/L non lobulated lungs, hypertrophied myocardium, AVSD	Common cloaca, AVSD, DORV

Table 3 continued

CASE		Kidneys	Bladder	Liquor	GIT	Perineal openings		Pelvic anatomy	Other malformations/findings	Diagnosis
						Uro-genital	Anal			
13	USG	B/L not seen	Not seen	Nil	–	–	–	Large retort shaped cystic mass, echogenic content	Thoracic hypoplasia, structurally normal heart with poor contractility	Complex pelvic mass
	Surgery	R dysplasia L absent	No bladder	–	–	Male	Absent	Colon draining to common cloaca	Baby expired the next day	Common cloaca
14	USG	B/L not seen	Solid mound	Nil	–	–	–	–	Hemivertebrae in lumbar region, vertebral segmentation defects	Complex pelvic mass
	Autopsy	Kidneys and ureters absent	Solid mound	–	–	Undifferentiated 3 tubercles ?Primitive genital tubercles, ?Male	Absent	Small mound of solid tissue in front of bowel, meconium filled distended large bowel	Hypoplastic right mandible,split thymus, small lungs, thick pterygeum from external genitalia to left foot, right CTEV	Popliteal pterygium/ Pallister–Hall variant

SUA single umbilical artery, *HUN/HDUN* hydronephrosis/hydroureteronephrosis, *UGS* urogenital sinus, *HLV* hypoplastic left ventricle, *CAT* common arterial trunk, *MCDK* multicystic-dysplastic kidneys, *CTEV* congenital talepus-equino varus, *SB* stomach bubble, *CHD* congenital heart disease, *CDH* congenital diaphragmatic hernia, *PS* pulmonary stenosis, *AVSD* atrio-ventricular septal defect, *DORV* double outlet right ventricle, *TOF* tetrology of fallot

Table 4 Compiled ultrasound and postnatal findings

Case no.	Pelvic mass	Ascites	AFI	Renal	Bladder	Cord	Ext. genital	Associated malformation				Syndromic	Perineal opening
								Heart	Limbs	Spine	Others		
1	+	+	↓↓	B/L HUN	−	N	M	−	−	−	−	−	1
2	−	++	Nil	B/L agenesis	−	N	M	−	−	−	Hydrocephalus	−	1
3	+	+	↓	B/L HDUN	−	N	M	−	−	−	−	−	1
4	+	−	N	−	Small	N	M	HLV/ CAT	−	−	CDH/short neck, hypoplastic thymus	Di-George	2
5	−	++	N	N	Lost contour	N	−	−	−	−	−	−	−
6	−	−	Nil	B/L agenesis	−	N	F	−	−	−	−	−	1
7	−	++	↓	Mild HDN	−	N	F	−	−	−	−	−	1
8	−	++	Nil	?	−	N	A	−	−	−	−	−	1
9	+	−	Nil	B/L MCDK	−	SUA	?	+PS VSD	Sirenomelia, absent thumb	VSD	−	Sirenomelia	0
10	−	−	Nil	R-dysplastic L-absent	−	Large vein SUA	M	TOF	LRD	Open NTD	−	VACTERL	1
11	++	−	↓	Dysplastic	−	N	F	−	−	−	−	−	1
12	−	−	↓	B/L MCDK	−	N	M	AVSD	−	−	−	−	0
13	++	−	Nil	B/L agenesis	−	N	M	−	−	−	−	−	0
14	−	−	Nil	B/L agenesis	−	N	M	−	Pterygeum, CTEV	Hemivertebra	−	?Pallister–Hall variant	1

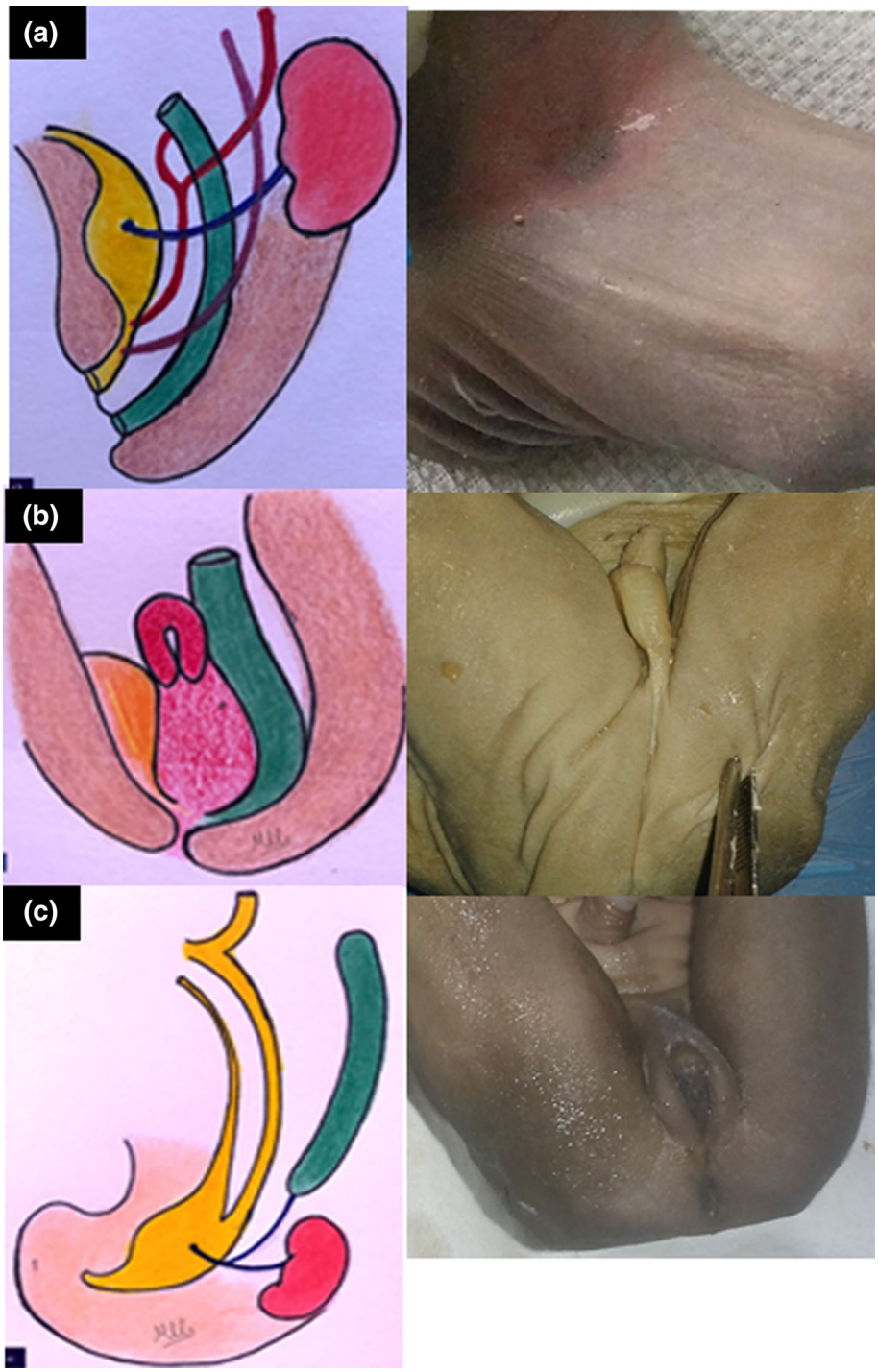

Fig. 1 Illustration of spectrum of cloacal malformations with representative images from the study group. **a** Smooth perineum with 0 opening, **b** Single perineal opening, **c** Two perineal openings

hydroureter, and hydronephrosis being the dominant features [18]. Multicystic dysplastic kidney, pelvic kidney, horse-shoe kidney, and crossed fused ectopia, renal agenesis can also accompany. The obstruction to outlet combined with poor renal function results in severe oligohydramnios leading to Potter sequence and also limits the visibility on ultrasound. Other associated abnormalities are ambiguous genitalia, a 2-vessel cord, tethered cord, hypoplastic sacrum, caudal dysgenesis, hemivertebrae etc. Common associated anomalies in our study were, renal in 12 (86%), cardiac in 4 (29%), vertebral and limb anomalies in 3 cases (21%) (Table 2 and Fig. 3). Bowel obstruction, evident as dilated bowel loops and meconium peritonitis due to reflux of meconium mixed urine into the peritoneal cavity via the fallopian tubes can also be occur. In the milder variant known as a persistent urogenital sinus

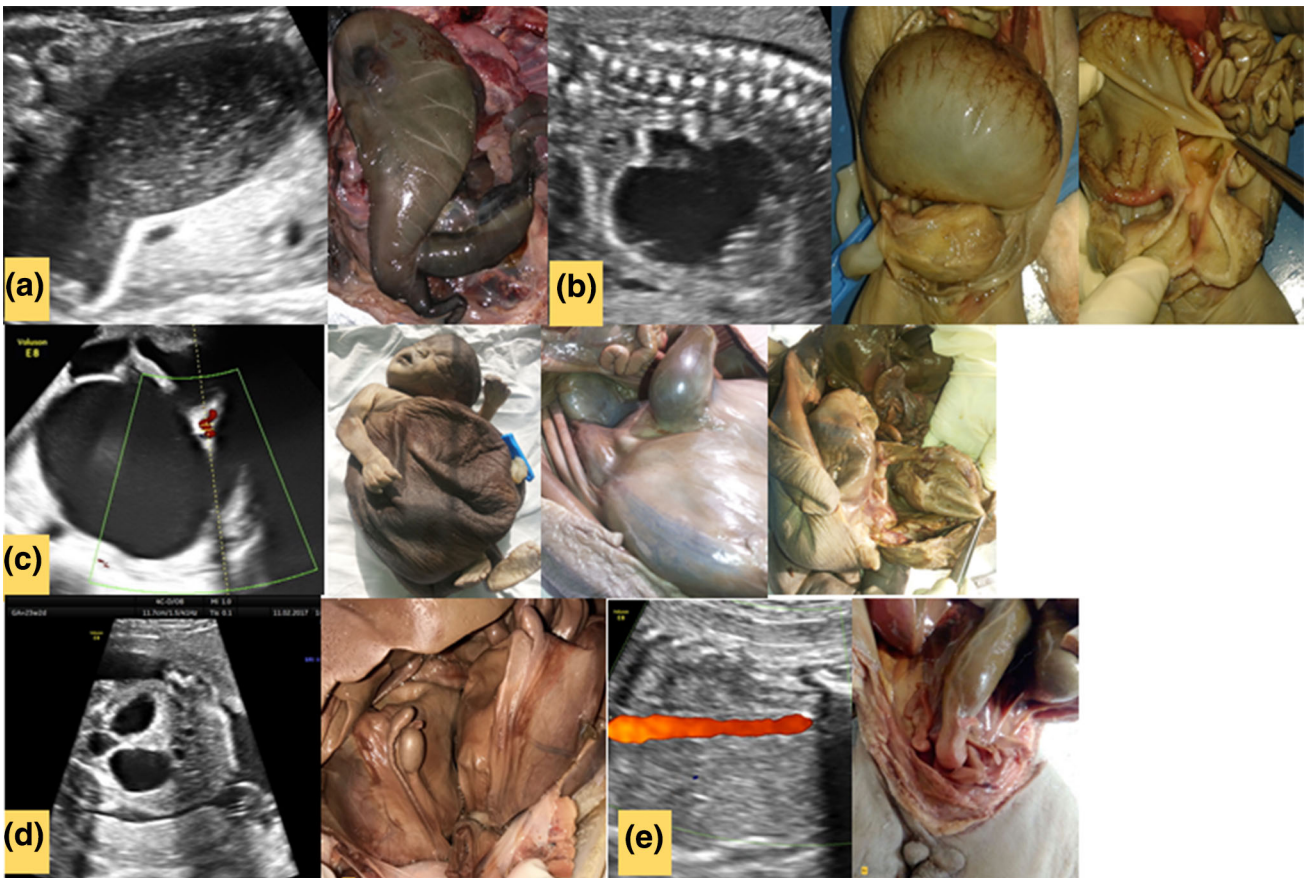

Fig. 2 Autopsy correlation with ultrasound findings. **a** Retort shaped pelvic mass on ultrasound, **b** Fetus had mass with fluid-debris level on ultrasound and a common cloaca with distended large bowel, **c** A large cystic mass suggesting bladder outlet obstruction had an obstructed common cloaca and obstruction at bowel connection, **d** Ultrasound showed a small bladder and two elongated, closely placed cystic structures. On autopsy, there was a common drainage canal and primitive metanephric ducts, **e** There was bilateral renal agenesis on ultrasound and no pelvic mass, however, autopsy showed a persistent urogenital sinus, non canalized rectum, primitive genital tubercles and pterygium of lower limb

without communication to the anorectum, the meconium filled rectum may be normal. Diagnostic modalities like MRI, diagnostic fetoscopy, and vesicocentesis can supplement the USG in cases where the diagnosis is unclear or to demonstrate additional anomalies [15]. It may be difficult to differentiate cloacal malformations from other causes of an abdomino-pelvic cystic mass in fetus as in, bladder outlet obstruction, pelvi-ureteric obstruction, ovarian cyst, enteric duplication cyst, mesenteric cyst, megacystis microcolon hypoperistalsis syndrome or a lymphangioma and cloacal dysgnesis [19, 20]. The mass in cloacal malformation is characteristically septate usually with fluid-debris level. A cloacal anomaly may also be a part of multisystem anomalies, urological, spinal, gastrointestinal and cardiac, thus overlapping with VACTERL association or caudal dysgenesis or caudal regression syndrome [21]. Therefore, an examination of the genitalia, spine and heart must be done in all cases of cloacal anomaly. These defects may be difficult to detect prenatally, if there is oligohydramnios and large mass obscuring the visibility.

Prenatal diagnosis should be followed by counselling to facilitate the parents make an informed choice and outline an optimal perinatal care. The outcome depends on the severity of the spectrum and associated malformations. Complete cloacal dysgenesis, at the severe end of the spectrum is often lethal, while a persistent urogenital sinus at the milder end has favorable prognosis. A poor outcome is expected in cases diagnosed early in pregnancy due to significant risk of lung hypoplasia and renal failure. Fetal interventions such as Vesico-amniotic shunt may be considered when there is severe oligohydramnios and severe hydronephrosis, but the experience with these interventions is limited [7]. The option of termination of pregnancy should always be discussed with the family whenever permissible. Three patients among our study group had second trimester termination of pregnancy, five had carried the pregnancy till term and five were delivered preterm. The decision to terminate pregnancy depended mainly on

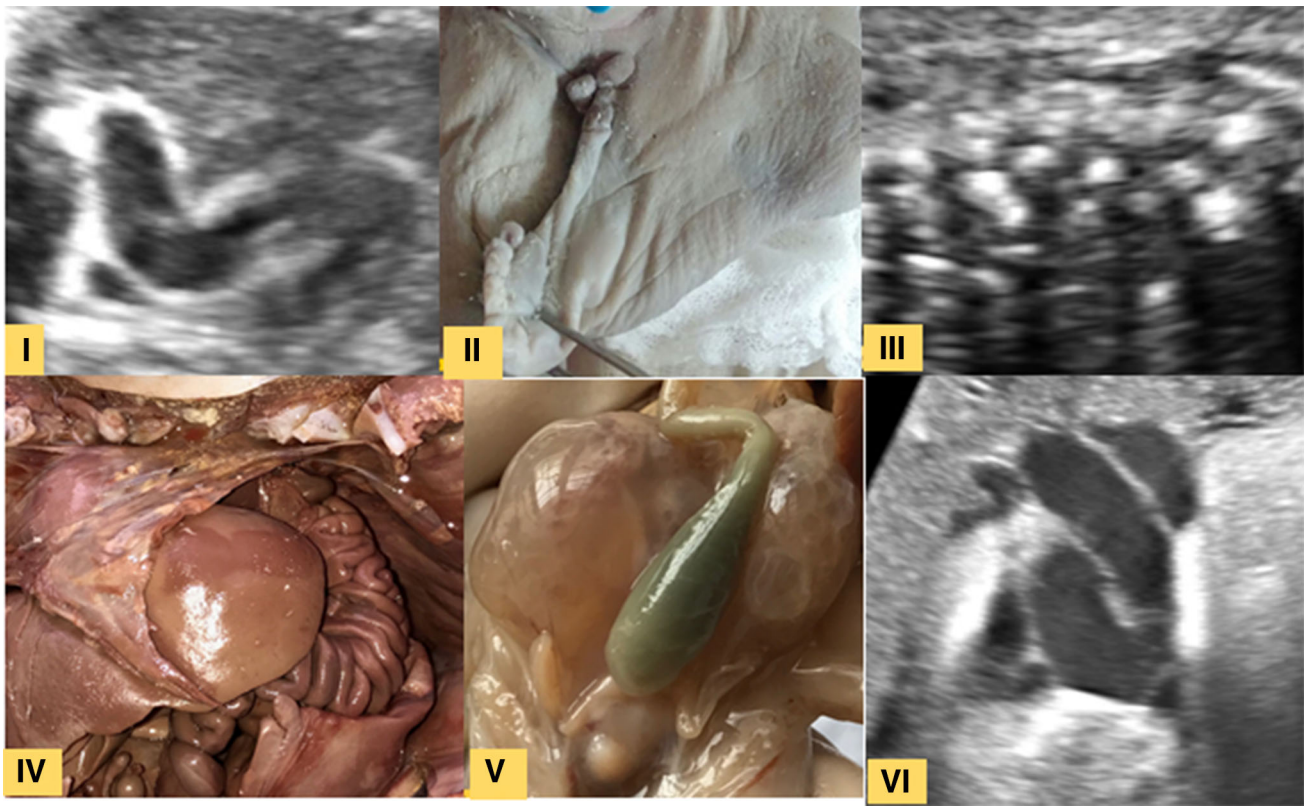

Fig. 3 Associated malformations. **i** Pulmonary atresia, **ii** Pterygeum, **iii** Vertebral defect, **iv** Congenital diaphragmatic hernia, **v** Bilateral multicystic kidneys, **vi** Single umbilical artery and dilated umbilical vein

the legally permissible limit of gestation for abortion, which is currently 20 weeks in India.

Postnatally, the goal of early management is to detect associated anomalies, and achieve diversion of the gastrointestinal tract urinary tract to relieve obstruction [15]. Initial decompression of bowel can be achieved by protective colostomy until definitive surgery is planned later. Definitive repair of persistent cloaca is a major technical challenge requiring correction of urogenital and anorectal tracts with multi-disciplinary approach in specialized centers [22]. Functional outcome depends on the length of the cloaca and it is difficult to repair long cloacas (> 3 cm) than short cloacas [6, 23]. In a large retrospective review of patients with cloacal anomalies, chronic renal failure occurred in childhood in 50% with a mortality rate of 6% from renal failure [24]. The long term follow up and functional outcomes of the survivors are unknown. Postnatal surgery with colostomy was done in two cases in the study group, but both of them expired within few days of surgery. There were no survivors in our study group as 3 had second trimester termination of pregnancy, while 8 expired immediately after birth and 2 expired in the postoperative period.

Conclusion

Prenatal diagnosis of cloacal malformation should be suspected in the presence of a pelvic cystic mass with septations and fluid-debris level. The diagnosis is reinforced when renal anomalies, non-vsualisation of bladder and anhydramnios accompany. Careful search for associated anomalies must be done and a combination of lumbosacral, genitourinary, and bowel malformations, if present should raise the possibility of a cloacal anomaly. Prenatal diagnosis helps in counselling, planning the management and appropriate referrals to improve the outcome. As this anomaly does not appear to be confined only to females, the presence of suspicious findings should raise the possibility of cloacal anomaly in male fetuses too. More research is needed to focus on the pattern and severity of presentation in male fetuses.

Acknowledgements We thank the Department of Obstetrics and Gynecology for their support for the study. We thank Dr. Manoj Kumar Rangappa. M.S. Mch, Consultant in Surgical Gestro-enterology for his contribution to development of the manuscript and inputs on ano-rectal malformations.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

References

1. Warne S, Chitty L, Wilcox D. Prenatal diagnosis of cloacal anomalies. BJU Int. 2002;89(1):78–81.
2. Qureshi F, Jacques S, Yaron Y, Kramer R, Evans M, Johnson M. Prenatal diagnosis of cloacal dysgenesis sequence: differential diagnosis from other forms of fetal obstructive uropathy. Fetal Diagn Ther. 1998;13(2):69–74.
3. Qureshi F, Jacques S. Cloacal abnormalities in male fetuses. J Ultrasound Med. 2012;31(12):2046–7.
4. Winkler N, Kennedy A, Woodward P. Cloacal malformation. J Ultrasound Med. 2012;31(11):1843–55.
5. Wheeler P, Weaver D. Partial urorectal septum malformation sequence: a report of 25 cases. Am J Med Genet. 2001;103(2):99–105.
6. Liang X, Ioffe O, Sun C. Cloacal dysgenesis sequence: observations in four patients including three fetuses of second trimester gestation. Pediatr Dev Pathol. 1998;1(4):281–8.
7. Morikawa M, Yamada T, Cho K, Yamada H, Minakami H. Prenatal diagnosis and therapy of persistent cloaca: a case report. Fetal Diagn Ther. 2006;21(4):343–7.
8. Cilento B, Benacerraf B, Mandell J. Prenatal diagnosis of cloacal malformation. Urology. 1994;43(3):386–8.
9. Williams D, Fitchev P, Policarpio-Nicolas M, Wang E, Brannigan R, Crawford S. Urorectal septum malformation sequence. Urology. 2005;66(3):657.e5–7.
10. Odibo A, Turner G, Borgida A, Rodis J, Campbell W. Late prenatal ultrasound features of hydrometrocolpos secondary to cloacal anomaly: case reports and review of the literature. Ultrasound Obstet Gynecol. 1997;9(6):419–21.
11. Zaccara A, Gatti C, Silveri M, Rivosecchi M, Bilancioni E, Spina V, et al. Persistent cloaca: are we ready for a correct prenatal diagnosis? Urology. 1999;54(2):367.
12. Liang X, Ioffe O, Sun C. Cloacal dysgenesis sequence: observations in four patients including three fetuses of second trimester gestation. Pediatr Dev Pathol. 1998;1(4):281–8.
13. Rios L, Araujo Júnior E, Nardozza L, Caetano A, Moron A, Martins M. Prenatal diagnosis and postnatal ultrasound findings of cloacal anomaly: a case report. Case Rep Pediatr. 2012;2012:1–3.
14. Livingston J, Elicevik M, Breech L, Crombleholme T, Peña A, Levitt M. Persistent cloaca. J Ultrasound Med. 2012;31(3):403–7.
15. Taipale P, Heinonen K, Kainulainen S, Seuri R, Heinonen S. Cloacal anomaly simulating megalocystis in the first trimester. J Clin Ultrasound. 2004;32(8):419–22.
16. Bischoff A, Levitt M, Lim F, Guimarães C, Peña A. Prenatal diagnosis of cloacal malformations. Pediatr Surg Int. 2010;26(11):1071–5.
17. Levitt M, Peña A. Cloacal malformations: lessons learned from 490 cases. Semin Pediatr Surg. 2010;19(2):128–38.
18. Levitt M, Peña A. Anorectal malformations. Orphanet J Rare Dis. 2007;2(1):33.
19. Petrikovsky BM, et al. Fetal cloacal anomalies: prenatal sonographic findings and differential diagnosis. Obstet Gynecol. 1988;72(9):464–9.
20. Gearhart J, Benchaim J, Jeffs R, Sanders R. Criteria for the prenatal diagnosis of classic bladder exstrophy. Obstet Gynecol. 1995;85(6):961–4.
21. Shaw-Smith C. Oesophageal atresia, tracheo-oesophageal fistula, and the VACTERL association: review of genetics and epidemiology. J Med Genet. 2005;43(7):545–54.
22. Warne SA, Hiorns MP, Curry J, et al. Understanding cloacal anomalies. Arch Dis Child. 2011;96(11):1072–6.
23. Wilcox DT, Warne SA. Urological problems in children with anorectal malformations. In: Holschneider AM, Hutson JM, editors. Anorectal malformations in children: embryology, diagnosis, surgical treatment and follow-up, vol. 276. Berlin: Springer; 2006. p. 269–81.
24. Warne SA, Wilcox DT, Ledermann SE, et al. Renal outcome in patients with cloaca. J Urol. 2002;67:2548–51.