Imperforate hymen and vaginal atresia and their associated anomalies[1]

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Summary: The presenting features and associated abnormalities of imperforate hymen and vaginal atresia were studied in 24 girls under the age of 16 years. Hydrocolpos or hydrometrocolpos occurred in 8 infants, 13 older girls developed haematocolpos, but 3 of the girls had no distension of the genital tract. Seven of the older girls were diagnosed as having appendicitis. Anorectal anomalies were present in 9 of the children. Intravenous pyelography was performed on 16 girls and was normal in only one. Urinary tract investigations are indicated in all girls with vaginal outlet obstruction, and the vaginal orifice should be inspected in all girls with anorectal abnormalities.

Introduction

Atresia of the lower genital tract is a well known but uncommon congenital abnormality of girls (Ho 1975). Most frequently, presentation occurs at puberty with abdominal pain and amenorrhoea when examination reveals either haematocolpos or haematometrocolpos. However, occasionally there is neonatal presentation in the form of hydrocolpos or hydrometrocolpos if the fetal cervical glands secrete mucoid material in response to maternal hormones. We report a series of 24 girls with vaginal outlet obstruction and describe the associated congenital abnormalities.

Method

A retrospective study was undertaken of a consecutive series of cases of vaginal outlet obstruction presenting at the Alder Hey Children's Hospital, Liverpool, during a 10-year period. The computerized list of classified diseases was cross-checked, all operating theatre record books were examined, and a list of classified diseases kept by the Senior Records Officer was consulted. The case papers of all girls undergoing gynaecological operations were studied; irrelevant cases were then discarded and appropriate notes abstracted. Details of the mode of presentation and the associated anomalies were collected and tabulated. Numerous cases of abnormalities of the urogenital tract were encountered, often in association with anorectal anomalies, but all were discarded unless there was good evidence of vaginal outlet obstruction.

Alder Hey Hospital is a large district paediatric hospital, accepting children from a wide area of East Liverpool. Additionally, the regional neonatal unit is situated in the hospital (Rickham *et al.* 1978), and therefore this series is biased by an unusually large number of neonatal cases. All cases of adrenogenital syndrome were excluded.

Results

Of the 24 girls with vaginal outlet obstruction, this was observed in 10 during the neonatal period although only 8 had hydrocolpos (Table 1). The obstruction was missed in 3 others who were seen as neonates with different anomalies. One 9-year-old girl presented with enuresis and was noticed to have an imperforate hymen, and the remaining 10 girls

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presented at puberty with abdominal pain and were eventually found to have haematocolpos (Table 1).

Thirteen girls had an imperforate hymen, 6 had vaginal atresia and 4 had double genital systems, comprising 2 with unilateral vaginal atresia, and 2 with unilateral imperforate hymen. One girl had a urogenital sinus. There was no genital distension in 3 children, but 6 developed hydrocolpos, one had hydrometrocolpos, one unilateral hydrometrocolpos with hydrosalpinx in a double system, and 7 had haematocolpos, 3 haematometrocolpos, 2 unilateral haematocolpos, and one had unilateral haematometra with haematosalpinx.

Anorectal abnormalities were present in 9 of of the girls, one of whom had a minor, low lesion amenable to anoplasty, and only one girl with high rectal atresia had the common variety with a colovestibular fistula. There was one girl with an anorectal atresia without a fistula. Four had hydrocolpos with the distal end of the large bowel draining into a closed vagina and one had cloacal exstrophy with a double uterus and a vaginal lesion which has not been fully investigated, since she is one of the three girls without distension of the genital tract. One girl had true rectal stenosis.

We have no information about the urinary tract in 8 of these girls, of whom 7 had imperforate hymen and one had vaginal atresia. The intravenous pyelogram (IVP) was normal in only one girl who, surprisingly, was the one with rectal atresia and hydrocolpos without a colovaginal fistula. A micturating cystogram (MCU) has not been performed. Urinary tract abnormalities were found in all of the remaining 15 patients in whom intravenous pyelography was performed. Unilateral hypoplastic kidneys were found in 2 girls, and 3 had unilateral renal agenesis. Duplex kidneys were found in 3 girls. Ectopic ureters were discovered in 2 infants, and one neonate had a non-functioning dysplastic kidney without a ureter. Hydronephrosis or hydroureter occurred in 8 children. One girl had a pelvic kidney.

Megacystis was present in 4 infants, 3 of whom had the urethra passing through a small phallus. One is really a female pseudohermaphrodite with proven ovaries on biopsy, and is in renal failure, having had two unsatisfactory renal transplants (case II). Another had 'prune belly' syndrome, and has been described elsewhere (Patel & Carty 1980). Two of the infants with megacystis were among a group of 3 girls each of whom had a tracheo-oesophageal fistula and a patent ductus arteriosus, with or without other congenital heart defects. In all 3 the hydrocolpos was due to an imperforate hymen. All developed severe respiratory distress in the neonatal period and none survived.

Thirteen patients presented at puberty, 11 with abdominal pain and primary amenorrhoea. Seven of these were diagnosed as having appendicitis and 5 were submitted to appendicectomy. Macroscopically the appendix was normal in each case but, somewhat surprisingly, histological examination revealed acute inflammation in 4 of them. The other 2 girls had had recurrent abdominal pain, suspected as being due to chronic appendicitis for several years prior to the diagnosis of haematocolpos.

Skeletal anomalies were not always recorded satisfactorily, and are omitted from this paper.

Discussion

At about the 10 mm crown-rump length stage of embryonic development, a linear invagination occurs in the coelomic epithelium lateral to the mesonephric duct (MND). This invagination forms the paramesonephric duct (PMND), which will subsequently develop into the upper vagina, uterus and Fallopian tubes. The cranial tip of the PMND will form the fimbrae of the tube, whilst the caudal tip migrates within the mesenchyme of the pelvic floor medial to the MND to meet, and fuse, with the contralateral PMND. The fused PMNDs then advance in the developing urorectal septum to the Müllerian tubercle at the urogenital sinus. Meantime, the PMND is canalizing. The fused part forms the uterovaginal canal. Endodermal proliferation occurs at the Müllerian tubercle and the sinovaginal bulbs of the urogenital sinus to form the definitive vagina (Hamilton *et al.* 1972). The hymen is a partition, persisting as a sandwich of mesoderm between the epithelium of the dilated,

Table 1. Presentation and associated anomalies in 24 girls with vaginal outlet obstruction

Case no.	Age and presentation	Genital tract	Urinary tract: Kidney/ureter	Urinary tract: Bladder	Gastro-intestinal tract	Cardio-vascular system	Other
1	NN Distended abdomen, respiratory distress	IH, hydrocolpos	Bilateral hydronephrosis				Hypoplastic lungs
2	NN Anorectal atresia	IH, hydrometrocolpos	Normal		Anorectal atresia		
3	NN Multiple anomalies	VA, hydrocolpos	R absent kidney L duplex ureter + ureterocele (crossed ectopia)		Covered anus		Rudimentary R thumb, bilateral inquinal hernia
4	NN Abdominal mass, anal anomaly	VA, hydrocolpos	R duplex ureter L pyelonephritis Bilateral hydroureter with reflux	Urogenital sinus	Rectal stenosis		Accessory auricles
5	NN Cardio-respiratory arrest at birth, multiple anomalies	RVA, double system R hydrometrocolpos R hydrosalpinx R absent ovary L vagina→cloaca	R hypoplastic kidney, bilateral hydronephrosis	Thick-wall bladder, vesico-vaginal fistula	Cloaca with fused labia		Bilateral intra-alveolar haemorrhages, talipes
6	NN Multiple anomalies	VA, no distension Bicornuate uterus Streak ovaries	R hydronephrosis	Megacystis, urethra-in-phallus	Anorectal atresia, colovaginal fistula draining to phallus		Prune belly (46XX), malrotation, sacral agenesis, split pubic symphysis
7	NN Multiple anomalies	IH, hydrocolpos Cloaca	L hydronephrosis	Megacystis, urethra-in-phallus	Cloaca	PDA ASD MA	TOF
8	NN Multiple anomalies	IH, hydrocolpos	Bilateral hydronephrosis	Megacystis		PDA	TOF
9	NN Multiple anomalies	IH, hydrocolpos Bicornuate uterus	L ectopic hydroureter		Anorectal atresia, colovaginal fistula	PDA COA	TOF, sacral hemi-agenesis
10	NN Cloacal exstrophy	VA, no distension Bicornuate uterus Bifid clitoris	R pelvic kidney L vesicoureteric reflux (after op)	Extopia vesicae	Exomphalos, colovesical fistula		Split pubic symphysis
11	NN Abnormal perineum 13 Pelvic mass, amenorrhoea	VA, haematometrocolpos	R hydronephrosis L hypoplastic kidney, bilateral hydroureter	Megacystis, urethra-in-phallus			

12	NN 13	Ectopia vesicae Pelvic mass	VA, haematometrocolpos	Bilateral pyelonephritis + calculi, staghorn calculus	Ectopia vesicae		
13	NN 13	Anorectal atresia Pelvic mass, abdominal pain	IH, haematocolpos	R cystic dysplastic kidney R absent ureter L ectopic ureter		Anorectal atresia, colovestibular fistula	Absent L radius
14	9	Enuresis	IH, no distension	No IVP, MCU normal			
15	14	Abdominal pain and mass	VA, haematocolpos	No IVP			
16	13	Abdominal pain and PB	IH, haematocolpos	No IVP			
17	13	Abdominal pain and PB	IH, haematocolpos	No IVP			
18	13	Abdominal pain and PB	IH, haematocolpos	No IVP			
19	14	Abdominal pain and PB	IH, haematocolpos	No IVP			
20	15	Appendicitis	IH, haematocolpos	No IVP			
21	13	Appendicitis	IH, haematometrocolpos Bilateral haematosalpinx	Bilateral duplex ureter			
22	14	Appendicitis	LIH, double system L haematocolpos R system normal	No IVP			
23	14	Abdominal pain and mass	LVA, double system L haematometra L haematosalpinx R system normal	L absent kidney R enlarged kidney			
24	11	Abdominal pain and mass	RIH, double system R haematocolpos L system normal	R absent kidney			

ASD atrial septal defect; COA coarctation of the aorta; IH imperforate hymen; L left; MA mitral atresia; NN neonate; PB perineal bulge; PDA patent ductus arteriosus; R right; TOF oesophageal atresia with tracheo-oesophageal fistula; VA vaginal atresia

canalized sinovaginal bulbs above, and the urogenital sinus, now the vulva, below (Koff 1933).

A cloaca is said to be caused if the urorectal septum does not migrate adequately. The PMND is then prevented from migrating to the perineum. Hence the urinary, genital and alimentary tracts have a communal opening. This is a severe anomaly and all 3 of our affected infants (cases 5, 6, 7) died early in the neonatal period. All died from cardiorespiratory distress and most had other severe anomalies. If such an infant survives, then a complex abdominoperineal pull-through of both vagina and colon may be required with simultaneous creation of a urethra from the urogenital sinus (Hendren 1977).

The urogenital sinus may persist as a common channel for the urinary and genital tracts. This is thought to represent developmental arrest after the PMND has fused with the urogenital sinus at about 37 mm crown-rump stage, but before the urogenital septum has migrated to divide the urinary and genital tracts (Marshall *et al.* 1979). In this series, we did not discover a case in association with membranous vaginal outlet obstruction, but a girl with the rarely encountered rectal stenosis (case 4) has the vagina draining into the urethra above the level of the external urinary sphincter. She presented in the neonatal period with gross hydrocolpos which was drained with a catheter. Now that menstruation has started, she has the unusual symptom of menouria. Nowadays, one would recommend early separation of the urinary and genital tracts, and this may have prevented pyelonephritis (Hendren 1977, Ein & Stephens 1971).

Vaginal agenesis is due to failure of the caudal migration of the PMND. The exact cause is unknown, but is often associated with abnormalities of the MND, kidneys and rectum (Bryan *et al.* 1949). Anomalies of the axial skeleton have also been reported by Chawla *et al.* (1966). Not surprisingly, the uterus and tubes are often poorly developed, but the ovaries are usually normal. The syndrome of vaginal agenesis, uterine abnormality, renal and skeletal anomalies is known as the Mayer-Rokitansky-Kuster-Hauser syndrome; some 500 cases have been recorded (Welch 1979). As a rule there is no urgency to treat girls with vaginal agenesis since menstrual products do not usually emanate from the hypoplastic uterus. Vaginal reconstruction is best left until the girl is willing to cooperate in the tedious treatment because of her desire for sexual intercourse (Johnston 1982, Bryan *et al.* 1949). Various approaches have been advocated for the creation of a cavity between the urethra and bladder in front and the rectum behind. McIndoe (1950) suggested a split skin graft and Pratt & Smith (1966) suggested the use of an isolated segment of sigmoid colon. However, in cases associated with rectal atresia with a colovestibular fistula, it may be more appropriate to convert the distal bowel into a vagina at the time of peforming an abdominoperineal colonic pull-through operation (Ein & Stephens 1971).

Vaginal atresia is clinically indistinguishable from vaginal agenesis, but is said to be due to failure of recanalization of the vaginal cord at the 150 mm crown-rump stage. The atresia may take the form of a septum – in any plane and at any level, including the site of the hymen – or it may take the form of a stenosis.

In this series, the vaginal outlet obstruction has been classified by the description in the case sheets. It appeared that the term 'vaginal atresia' was used in all cases in which there was more than just a simple, thin, imperforate hymen. In retrospect, it is probable that these cases were correctly diagnosed. The distinction between agenesis and atresia is of some importance, since in the former group sterility is common because of the severe hypoplasia of the uterus and tubes, whereas pregnancy is possible in the latter group following surgery. Pregnancy occurred in one of our patients (case 15) following a modified McIndoe's operation. Serious urinary tract anomalies were present in all of the girls undergoing investigation in this series, although the incidence of renal anomalies has been put at between 30 and 50% (Bryan *et al.* 1949, Phelan *et al.* 1953, Chawla *et al.* 1966). Treatment for vaginal atresia becomes necessary at puberty as the genital tract becomes painfully distended at each attempt at menstruation. A combined abdominoperineal procedure is necessary, and it is not safe to approach the distended upper vagina by blind dissection from the perineum alone (Ramenofsky & Raffensperger 1971).

Imperforate hymen is the commonest obstructive lesion which may present with hydrocolpos in the neonate or with haematocolpos at puberty. The incidence is said to be one in 16 000 female births (Westerhout *et al.* 1964). The prognosis for the neonate is poor, and 4 of our 6 patients died, mainly from respiratory failure in association with gross abdominal distension (Reed & Griscom 1973). The distension may be so severe as to cause abdominal dystocia, urinary retention (MacLachlan *et al.* 1980), upper urinary tract dilatation (Cook & Marshall 1964) or bowel perforation (Gupta & Barson 1980). Hydrocolpos is known to be associated with imperforate anus (Young 1977) and with polydactyly, especially in the familial type of hydrocolpos (MacLachlan *et al.* 1980, Gupta & Barson 1980). Three of our infants had oesophageal atresia with tracheo-oesophageal fistula and patent ductus arteriosus. This is an unusual combination of anomalies and it is possible that the resulting disorders of circulation of the amniotic fluid could lead to additional pulmonary complications. Hydrometra and hydrosalpinx are uncommon (Ho 1975), but one of our patients (case 7) developed plastic peritonitis from leakage of the mucinous fluid into the peritoneal cavity (Gupta & Barson 1980). Occasionally peritoneal calcification occurs (Reed & Griscom 1973). Only one of our 6 infants had two normal kidneys. Very often, the infant presents with an abdominal mass rather than a perineal bulge. If the obstructing membrane is thin and low, a cruciate incision under antibiotic cover is all that is required. However, the presence of a thick or high membrane is an indication for laparotomy, evacuation of the fluid by suction, followed by an abdominoperineal procedure to relieve the obstruction.

Haematocolpos presents at puberty with primary amenorrhoea and frequently with recurrent abdominal pain, sometimes for several years. Unlike in the neonate, leakage of menstrual fluid into the peritoneal cavity is common, usually through only one Fallopian tube. Frequently, the diagnosis of appendicitis is entertained, as occurred in several of our cases. Simple incision of the hymen may be inadequate since the collection may recur if the edges reseal, as in 2 of our cases. We recommend a cruciate incision with sutures to keep the edges retracted. Antibiotic cover is advised to avoid infective complications and subsequent infertility (Brown & Brews 1930). One of the girls in this series eventually had three successful pregnancies. Unfortunately, the urinary tract was adequately investigated in only one of our adolescent cases, but we suspect that some of these girls may have had renal anomalies.

Genital tract duplication is rare and is said to occur most commonly in association with exstrophy of the cloaca (Johnston 1982). However, there was only one such patient (case 10) in our small series, and 4 other girls had genital duplications. One (case 5) presented in the neonatal period with cloacal abnormality and did not survive. A 14-year-old girl (case 23) caused considerable diagnostic confusion when she presented with recurrent abdominal pain and a left-sided abdominal mass. The vulva was inspected and there was a normal, patent vagina, so haematocolpos was not considered further, since menstruation occurred normally. It was decided that she had a torsion of a left ovarian cyst and ultrasound examination seemed to support that diagnosis, the left ovary being indistinguishable from the mass. At laparotomy, the mass consisted of the left tube and ovary, thought to be a tubo-ovarian abscess, which was removed. It was then realized that there was a separate left-sided cervix which did not communicate with the perineum, although the upper part of the vagina was present. A window was then made from the patent right vagina to the upper part of the left vagina and old menstrual blood discharged. In retrospect, menstruation had occurred only from the patent right vagina and a simpler operation would have cured the girl.

Two other girls (cases 22 and 24) had unilateral imperforate hymen with contralateral perforate hymen and both were thought to have normal menstruation when first questioned. Despite the presence of a blue, left-sided perineal bulge, the mother of one of these girls (case 22) became most indignant when it was suggested that the girl had abnormal menstruation. No urinary tract investigations were performed in one girl, but all of the others had severe hypoplasia or agenesis of the kidney on the same side as the vaginal outlet obstruction.

Awareness of these conditions so as to avoid unnecessary hysterectomy has been emphasized by Dajani (1972). We should like to stress the importance of investigation of the urinary tract in these girls, and recommend ultrasound examination of the kidneys in all cases of genital abnormality. More detailed investigation is required in the case of neonates, and early corrective surgery is now recommended at one major session (Hendren 1977, Ein & Stephens 1971, Ramenofsky & Raffensperger 1971). In 1941, Gruenwald demonstrated that PMND growth and migration is dependent upon the presence of the MND. However, a reciprocal induction is proposed to exist by Marshall & Beisel (1978), between the metanephric blastema (primitive kidney) and ureteric bud which arises from the MND. So if a MND abonormality might be the source of abnormal growth and development of the PMND, and hence the uterus and vagina, it seems logical to suspect that ureteric and metanephric and hence renal abnormalities should also be associated with original MND defects. This explanation is based mainly on observations in rats. However, there has been some evidence in humans (Phelan *et al.* 1953, Johnston 1978, Woolf & Allen 1953), and certainly there is a strong association between vaginal and upper urinary tract abnormalities in our series.

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