Anorectal Malformations(ARM) : The Hospital Universiti Sains Malaysia (HUSM) experience from 1999 to 2006.

By

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VI. ABBREVIATIONS

A&E	Accident and emergency department
ARM	Anorectal Malformation
ASD	Atrial septal defect
CT	Computerized tomography
CBD	Continuous bladder drainage
CNS	Central nervous system
DRE	Digital rectal examination
DTPA	Diethylenetriamine pentaacetic acid
EUA	Examination under anaesthesia
GA	General anaesthesia
GIT	Gastro intestinal tract
G6PD	Glucose-6 –Phosphate Dehydrogenase
HKL	Hospital Kuala Lumpur
HKT	Hospital Kuala Terengganu
HRPZ	Hospital Raja Perempuan Zainab II, Kota Bharu
HTAA	Hospital Tengku Ampuan Afzan, Kuantan
HUSM	Hospital Universiti Sains Malaysia
IVP	Intravenous pyelography
KG	Kilogramme
LA	Local anaesthesia
MA	Malformasi anorektum

MCU	Micturating cystourethrogram
MRI	Magnetic resonance imaging
NEC	Necrotizing Enterocolitis
O&G	Obstetrics and Gynaecology
PFO	Patent foramen ovale
PS	Pulmonary stenosis
PSARP	Posterior Sagittal Anorectoplasty
PUJ	Pelviureteric junction
R&D	Research and development
SOPD	Surgical Out Patient Department
TEF	Tracheo-esophageal fistula
TOF	Tetrology of Fallot
US	Ultrasound
UTI	Urinary tract infection
VSD	Ventricular septal defect
VUR	Vesicoureteric reflux

VII. ABSTRAK

PENGENALAN

Malformasi Anorektum (MA) ialah satu malformasi kmpleks yang disebabkan oleh ketiadaan atau kedudukan ektopik anus. Ia biasanya didiagnosa pada hari bayi dilahirkan. Laporan menunjukkan ia biasanya berlaku antara 1 dalam 1500 dan 1 dalam 5000 kelahiran di mana lazimnya ia berlaku pada bayi lelaki berbanding bayi perempuan. Kadar kejadian anomali organ yang berkaitan dengan MA dilaporkan berlaku pada kadar 20% - 70% yang mana sesetengahnya adalah anomali yang minor tetapi yang lain boleh melibatkan nyawa. Selebihnya ia bergantung kepada jantina pesakit, jenis malformasi iaitu samada rendah atau tinggi dan anomali yang berkaitan. Semua prosedur pembedahan untuk memperbaiki MA bertujuan untuk menyediakan tempat yang sesuai bagi pengeluaran najis dari perineum dan juga wewujudkan satu hubungkait antara usus dan sfinkter.

OBJEKTIF

Matlamat kajian ini ialah untuk menilai semua pesakit yang mengalami malformasi anorektum di Hospital Universiti Sains Malaysia (HUSM) dari Januari 1999 sehingga Januari 2006. Ia juga bertujuan untuk menghurai demografi dan kesan rawatan mengikut jenis MA.

METODOLOGI

Satu kajian retrospektif telah dijalankan di Unit Surgeri Pediatrik, Jabatan Surgeri, Hospital Universiti Sains Malaysia (HUSM) dengan mengumpul nama-nama pesakit dalam buku pembedahan pediatrik di antara Januari 1999 dan Januari 2006. Fail rawatan pesakit disaring untuk data epidemiologi dan data yang berkenaan untuk kajian ini. Semua data dimasukkan dan dianalisis menggunakan pakej sains sosial dan statistik versi 12 (SPSS 12) yang diberi lesen kepada USM. Nilai p kurang dari **0.05** dianggap sebagai signifikan secara statistik.

KEPUTUSAN

Seramai 98 pesakit yang memenuhi kriteria penyertaan dimasukkan dalam kajian ini. Sembilan puluh tujuh peratus pesakit adalah Melayu dan nisbah lelaki dengan perempuan ialah 2 : 1. Berat pesakit ketika lahir antara 1.3kg ke 4.5kg dan seramai 42.8% pesakit dilaporkan ke HUSM pada hari pertama dilahirkan. Laluan tidak normal mekonium (39.7%), pengembungan perut (15.3%) dan sembelit (5.1%) adalah antara simptom yang lazim bagi penyakit ini. Prosedur penyiasatan yang biasa dilakukan untuk mengesan anomali yang berkaitan adalah dengan menggunakan ultrasound abdomen (94.8%), "X ray" keseluruhan bayi (87.7%) dan ekokardiogram (75.5%). Anomali yang berkaitan pula dikesan pada 56.6% pesakit. Kebanyakan anomali yang berkaitan yang dikesan dalam pesakit MA adalah anomali kardiovaskular (25.5%) diikuti dengan anomali urologi (22.4%) dan anomali kromosomal (17.3%). Seramai 52 orang pesakit menjalani

pembedahan pembetulan untuk MA rendah iaitu PSARP mini (69.2%) dan diikuti oleh anoplasti (25%) dan pemindahan anal (5.8%). Empat puluh tujuh peratus pesakit menjalani pembedahan untuk MA tinggi, yang mana hanya 2% sahaja menjalani pembedahan PSARP secara satu peringkat. Seramai 44 (73.3%) pesakit yang menjalani pembentukan kolostomi didiagnosa dengan jenis MA tinggi. Enam belas orang pesakit yang selebihnya yang menjalani pembentukan kolostomi didiagnosa dengan MA rendah (26.7%). Komplikasi-komplikasi selepas pembedahan adalah sama dengan komplikasi yang dilaporkan dalam kajian- kajian yang lebih awal. Dalam kajian ini, terdapat seramai 46 pesakit (46.9%) jenis MA tinggi dan 52 (53.1%) pesakit dengan jenis MA rendah. Namun begitu hanya 44 orang pesakit sahaja yang dapat dihubungi dan ditemubual unuk menilai keberkesanan rawatan. Penyempitan anus merupakan komplikasi yang signifikan bagi pesakit yang tidak patuh dengan protokol pembesaran anus (p=**0.007**). Pesakit dengan MA rendah di dapati mempunyai berat yang lebih baik berbanding dengan pesakit yang MA tinggi (p=**0.002**). Pesakit MA rendah mengalami keadaan sembelit lebih tinggi dan ini amatlah signifikan (p=**0.000**). Namun begitu kejadian terkincit dan inkontinens lebih tinggi di kalangan pesakit MA tinggi (P=**0.000**). Tujuh orang pesakit mampu mencapai kontinens sepenuhnya semasa lawatan susulan dan kesemuanya lelaki (p=**0.048**). Hanya 7 orang pesakit dalam kajian ini mencapai keputusan yang baik selepas pembedahan, 54 pesakit mencapai keputusan sederhana dan 16 pesakit keputusan lemah. Kadar kematian selepas pembedahan definitif pula sebanyak 3%.

KESIMPULAN

Keputusan kajian ini hampir sama dengan kajian yang telah diterbitkan dalam terbitan dari negara-negara lain. Diagnosa klinikal tentang jenis MA juga tepat dalam 76.8% pesakit dan diagnosis MA tinggi lebih susah dibuat berbanding MA rendah. Insiden yang tinggi berkenaan anomali yang berkaitan dengan MA dalam kajian kami, menjadikan pemeriksaan klinikal dan evaluasi neonatal dan tempoh awal bayi diwajibkan dalam semua kes MA. Pembesaran anus adalah tatacara yang penting dalam pengurusan selepas pembedahan PSARP untuk mengelak daripada penyempitan bahagian anoplasti. Memandangkan hanya sebilangan pesakit yang mencapai keputusan yang baik, satu pendekatan secara berkumpulan dan program pengurusan usus yang lebih baik perlu di buat supaya lebih ramai pesakit akan datang untuk rawatan susulan dan seterusnya mendapat keputusan yang lebih baik.

VIII. ABSTRACT

INTRODUCTION

Anorectal Malformations (ARM) are a complex group of malformations diagnosed at the time of birth because of absence or an ectopic location of anus. The usual reported incidence is between 1 per 1500 and 1 per 5000 live births and they are more often seen in boys than in girls. The incidence of associated organ anomalies with ARM is variously reported from 20% -70% some being minor anomalies but others being life threatening. Further management depends on the sex of the patient, type of malformation either high or low and the associated anomalies. All operative procedures for the correction of ARM aim at providing portal for the discharge of feces from the perineum and establishing a working relationship between the bowel and sphincter.

OBJECTIVES

The aim of the study is to review the patients presenting with anorectal malformations (ARM) to Hospital Universiti Sains Malaysia (HUSM) and describe the demographics and outcome in relation to the type of ARM.

METHODOLOGY

The study was a retrospective case review which was carried out in the Paediatric Surgery Unit, Department of Surgery, Hospital Universiti Sains Malaysia (HUSM) between January 1999 and January 2006. The case notes and operative notes were screened for epidemiological data and data relevant to the study. Patients diagnosed with ARM but did not undergo surgery, and patients whose case notes could not be traced or incomplete were excluded from the study. All the data entry and analysis were carried out using the social science and statistical packaged (SPSS) version 12 licensed to USM. A p value of less than **0.05** was considered statistically significant.

RESULTS

Ninety eight patients were included into the study after fulfilling the inclusion criteria. The male to female ratio was 2 to 1 and 97% of the patients were Malays. The birth weight of the patients in this study ranged from 1.3kg to 4.5kg. Forty two point eight percent of the patients presented to HUSM within the first day of life. The most common presenting symptoms were abnormal passage of meconium (39.7%), abdominal distension (15.3%) and constipation (5.1%). The investigative procedures done to detect associated anomalies were ultrasound abdomen (94.8%), babygram (87.7%) and echocardiogram (75.5%). Associated anomalies were detected in 56.6% of the patients. The most number of associated anomalies detected in patients with ARM was cardiovascular anomalies (25.5%) followed by urological anomalies (22.4%) and

chromosomal anomalies (17.3%). Fifty two patients (53%) underwent surgical repair for low ARM which were mini PSARP (69.2%) followed by anoplasty (25%) and anal shift (5.8%). Fofty six patients (46.9%) underwent surgery for high ARM of which only 2% underwent single stage PSARP repair. Forty four patients (73.3%) who had colostomy formed were later diagnosed with high type of ARM. The remaining 16 patients who had colostomy formed were diagnosed with low ARM (26.7%). Post operative complications were similar to those reported in earlier studies. In this study, there were 46 patients (46.9%) with high type of ARM and 52 patients (53.1%) with low type of ARM. Only 44 patients were able to be contacted and interviewed to assess their functional outcomes. Anal stricture was a significant complication in patients who were not compliant with the anal dilatation protocol (p = **0.007**). Patients with low ARM were noted to have more adequate weight gain as compared to high ARM patients (p=**0.002**). The incidence of constipation was higher among patients with low ARM and this was highly significant (p= **0.000**). However, the incidence of soiling and incontinence was higher among patients with high ARM (p=**0.000**). Seven patients achieved full continence at follow up and they were all males (p=**0.048**). Only 7 patients in our study achieved "good" outcome following surgery, 54 patients achieved "fair" outcome and 16 patients had "poor" outcome. The mortality rate after definitive surgery in this study was 4%.

CONCLUSION

The demographic finding in the study is quite similar to those published in the literature from other parts of the world. The clinical diagnosis of type of ARM was accurate in 76.8% of the patients and it was more difficult in making a diagnosis of high ARM clinically, compared to low ARM. The high incidence of associated anomalies in our study makes careful clinical examination and evaluation during the neonatal and early infantile period mandatory in all cases of ARM. Anal dilatations are a vital part of the postoperative management to avoid stricture at the anoplasty site. The low number of patients with good outcome in our study suggests that more attempts must be made to keep these patients on follow up, with the development of a proper bowel management program and an integrated team approach to achieve better outcomes.

1. INTRODUCTION TO ANORECTAL MALFORMATION (ARM)

Anorectal malformations (ARM) are one of the commonest causes of intestinal obstruction in the newborn and hence a topic of concern for paediatric surgeons all over the world. Anorectal Malformations (ARM) is a complex group of malformations diagnosed at the time of birth because of absence or an ectopic location of anus (Alford BA, Mcllhenney J. 1997). The usual reported incidence is between 1 per 1500 and 1 per 5000 live births and they are more often seen in boys than in girls (Gupta DK et al. 2002, Pena A. 1992, Saaul DB, Harrison EA. 1997, Stringer D. 1994). Some studies reported the incidence of high ARM to be twice as frequent as low ARM and to be more in boys (Cho S et al. 2001, Kiely EM, Pena.A. 1998, Mittal A et al. 2004).

The incidence of associated organ anomalies with ARM depends upon the meticulousness with which they have been searched for by clinical examination and various investigative procedures. The incidence is variously reported from 20% -70% some being minor anomalies but others being life threatening. These associated congenital anomalies not only lead to overall mortality but also contribute to significant morbidity (Cho S et al. 2001, M. Endo et al.1999, Hassink EA et al. 1996, Nazer J et al. 2000).

Early resuscitation and treatment for neonates born with anorectal malformation is crucial. Further management depends on the sex of the patient, type of malformation, either high, intermediate or low and the associated anomalies. Over the last half century, the treatment of ARM has evolved from a simple cutback to abdominoperineal pull

through, and its modifications to the currently practiced procedure of Posterior sagittal anorectoplasty (PSARP). All operative procedures for the correction of ARM aim at providing portal for the discharge of feces from the perineum and establishing a working relationship between the bowel and sphincter (Pena A, Hong A. 2000).

2.LITERATURE REVIEW

2.1 Embryology

The anus and rectum develop from the dorsal potion of the hindgut or cloacal cavity when lateral ingrowth of the mesenchyme forms the urorectal septum in the midline. This septum separates the rectum and anal canal dorsally from the bladder and urethra. The cloacal duct is a small communication between the 2 portions of the hindgut; downgrowth of the urorectal septum closes this duct by the seventh week of gestation. During this time, the ventral urogenital portion acquires an external opening; the dorsal anal membrane opens later. The anus develops by a fusion of the anal tubercles and an external invagination, known as the proctodeum, which deepens toward the rectum but is separated from it by the anal membrane. This separating membrane should disintegrate during the eighth week of gestation (TW Sadler. 1995).

Interference with anorectal structure development at varying stages leads to various anomalies, ranging from anal stenosis, incomplete rupture of the anal membrane (covered anus) or anal agenesis (a low lesion) to complete failure of the upper portion of the cloaca

to descend and failure of the proctodeum to invaginate (a high lesion). Continued communication between the urinary tract and rectal portions of the cloacal plate causes rectourethral fistulas or rectovaginal fistulas.

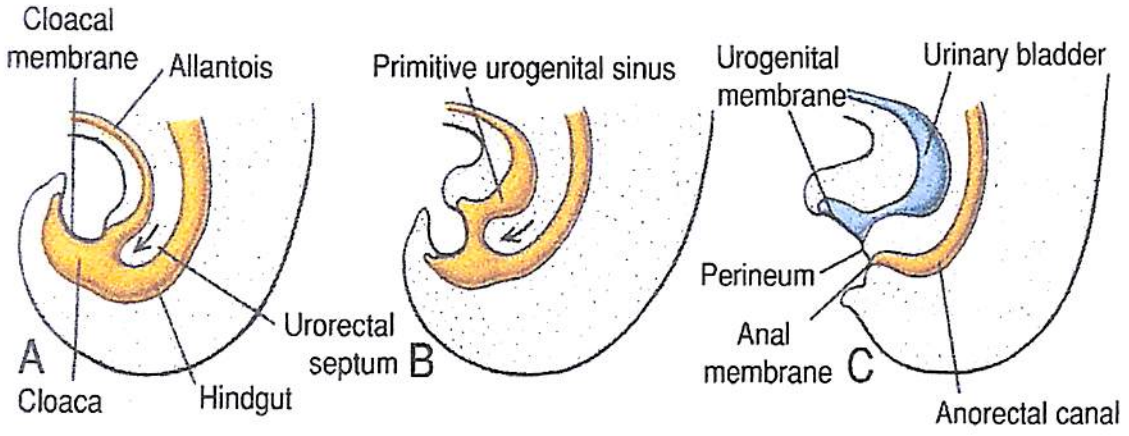

Figure 1 The cloacal region in embryos at successive stages of development (A,B,C). Arrow indicates the route of descent of the urorectal septum. Note the anorectic canal and perineum (adapted from TW Sadler (1995), Langmans Medical Embryology, 7th ed).

The external anal sphincter, derived from the exterior mesoderm, usually is intact and is uninvolved with obstructive anal or rectal lesions. This sphincter marks the prospective anal opening when formation occurs normally. Careful identification of this sphincter is important in planning operative repair of these lesions (Pena A et al. 2004). High anomalies (ie, those above the puborectalis muscle) occur much more commonly in males. A rectourethral fistula usually develops between the rectum and prostatic urethra. Maldevelopment of the sacrum hampers innervation of both anal and urethral musculature (Nazer J et al. 2000).

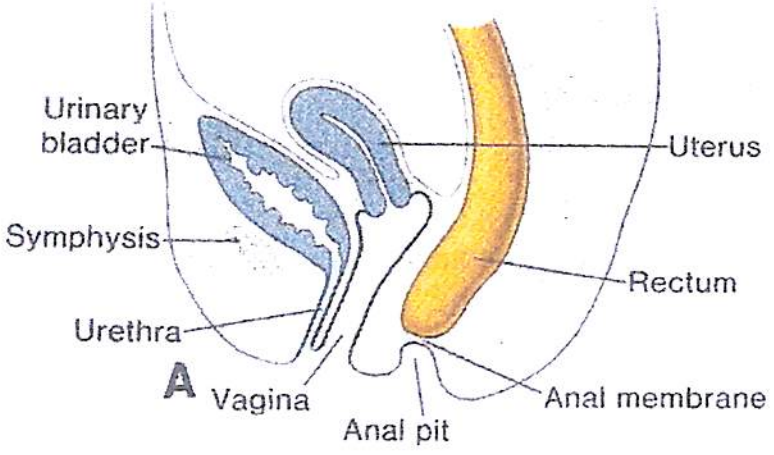

Figure 2. Drawing of an imperforate anus. The anal membrane persists as a diaphragm between the upper and lower portions of the anal canal. (Low type - adapted from TW Sadler (1995), Langmans Medical Embryology, 7th ed)

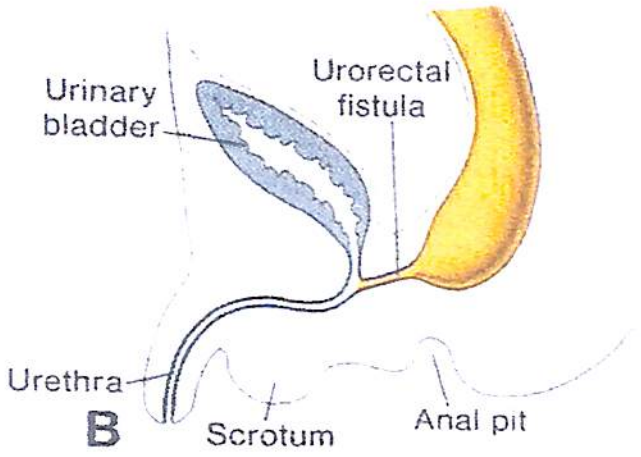

Figure 3. Drawing of urorectal fistula combined with rectal atresia due to a high defect in formation of urorectal septum. (High type - adapted from TW Sadler (1995), Langmans Medical Embryology, 7th ed)

2.2 Anatomy

The rectum is about 12cm long in an adult and is continuous with the sigmoid colon at the level of the third piece of the sacrum. At this junctional region the sigmoid mesocolon ends and the rectum has no mesentry. The taenia of the sigmoid colon gradually broaden to form wide anterior and posterior muscular bands which meet laterally to give the rectum a complete outer layer of longitudinal muscle. There are no appendices epiploicae in the rectum. The rectum turns downwards and backwards as the anal canal 2 to 3 cm in front of the tip of the coccyx. Although the rectum has no mesentry, the visceral pelvic fascia around the rectum is referred to by surgeons as the mesorectum.

Peritoneum covers the upper third of the rectum at the front and sides, and the middle third only at the front. The lower third is below the level of the peritoneum which is

reflected forwards on to the upper part of the bladder (in the male) or upper vagina to form the rectovesical pouch or rectouterine pouch of Douglas. Below the level of the pouch are the rest of the bladder base and seminal vesicles, the prostate, and the ends of the ureter and ductus deferens. In front of the rectouterine pouch is the uppermost part of the vagina (the fornix with the cervix of the uterus projecting into it) while below the peritoneal reflection is more of the vagina, with the thin rectovaginal fascia intervening.

Blood supply of the rectum is principally from the superior rectal artery with contributions from the middle and inferior rectal artery and median sacral vessels. The lower end of the inferior mesenteric artery enters the sigmoid mesocolon and changes its name to superior rectal artery on crossing the pelvic brim. These vessels pierce the muscular wall and supply the whole thickness of the rectal wall including the mucous membrane. They continue submucosally into the anal where they anastomose with the branches of the inferior rectal artery.

The middle rectal arteries reach the lower rectum from the side along the lateral rectal ligaments. The median sacral artery may make an unimportant contribution to the posterior wall in the region of the anorectal junction. Veins correspond to the arteries but anastomose freely with one another, forming an internal rectal plexus in the submucosa and an external rectal plexus outside the muscular wall. The lower end of the internal plexus is continuous with the vascular cushions of the anal canal. The superior and inferior rectal veins are the main veins and closely follow their arteries so that drainage is to both the portal and systemic systems.

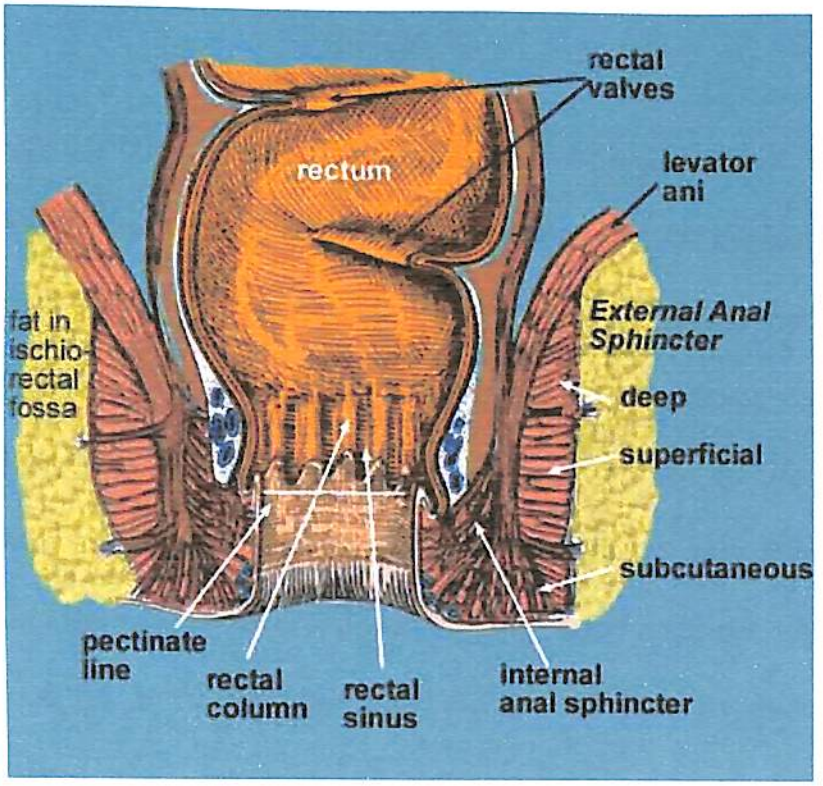

Figure 4. Anatomy of the rectum and anal canal. (adapted from Grant's Atlas of Anatomy)

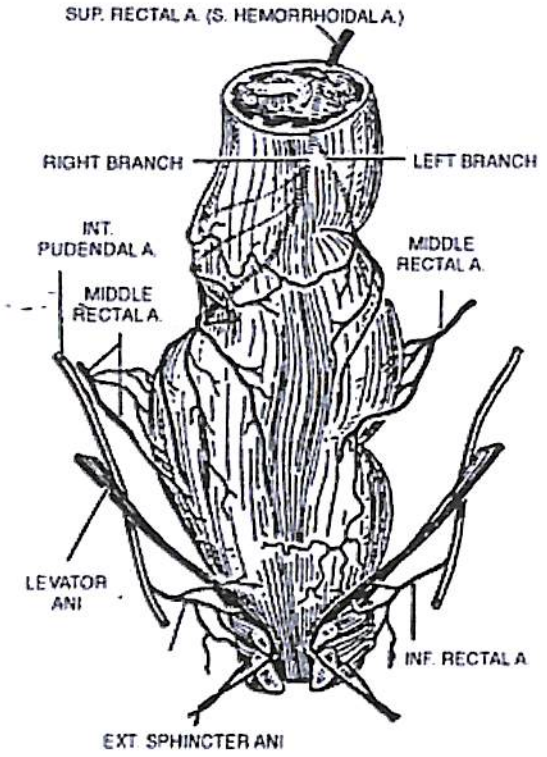

Figure 5. Blood supply to the rectum. (adapted from Grant's Atlas of Anatomy)

Lymphatic drainage from the rectum is mainly upwards. Lymphoid follicles in the mucous membrane drain to the pararectal nodes on the surface of the rectum and thence upwards via nodes along the inferior mesenteric artery to preaortic nodes. Lymphatic drainage from the lower rectum may reach internal iliac nodes along middle rectal and inferior rectal vessels. The sympathetic nerve supply is derived by branches from the hypogastric plexuses and by fibres which accompany the inferior mesenteric and superior rectal arteries from the celiac plexus and ganglia. The parasympathetic supply is from S2 and S3 (or S3 and S4) by the pelvic splanchnic nerves which are motor to the rectal muscle.

2.3 Physiology

Distension of the rectum with feces initiates reflex contractions of its musculature and the desire to defecate. In humans, the sympathetic nerve supply to the internal (involuntary) anal sphincter is excitatory, whereas the parasympathetic supply is inhibitory. This sphincter relaxes when the rectum is distended. The nerve supply to the external anal sphincter, a skeletal muscle comes from the pudendal nerve. The urge to defecate first occurs when rectal pressure increases to about 18 mm Hg. When this pressure reaches 55 mm Hg, the external as well as the internal sphincter relaxes and the contents of the rectum are expelled.

Before the pressure that relaxes the external anal sphincter is reached, voluntary defecation can be initiated by voluntarily relaxing the external sphincter and contracting the abdominal muscles thus aiding the reflex emptying of the distended rectum. Defecation is therefore a spinal reflex that can be voluntarily inhibited by keeping the external sphincter contracted or facilitated by relaxing the sphincter and contracting the abdominal muscles. Defecation after meals is the rule in children due to the gastrocolic reflex. In adults, habit and cultural factors play a large role in determining when defecation occurs.

2.4 Epidemiology

The incidence of ARM is between 1 per 1500 and 1 per 5000 live births and they are more often seen in boys than in girls (Gupta DK et al. 2002, Pena A. 1992, Saaul DB et al. 1997, Stringer D. 1994). Some studies reported the incidence of high ARM to be twice as frequent as low ARM and to be more in boys (Cho S et al. 2001, Kiely EM, Pena A. 1998, Mittal A et al. 1998). However, Niedzielski J (2000) reported higher incidence of females with ARM having low anomalies in Lodz province, Poland. There were no racial differences in disease incidence or severity of disease documented, although the disease seemed more common in some areas. Chatterjee cited an incidence in Calcutta of 1 case in 1862 live births. Some families have a genetic predisposition and successive generations have anorectal malformations (Chatterjee SK, Das S. 1979).

The underlying etiology of ARM's is not well documented. There is no single etiological cause to account for them, a situation that seems to fit with the spectrum of anomalies encountered. Other suggested etiologies include environmental factors such as fetal alcohol syndrome and infectious factors such as toxoplasmosis and cytomegalovirus. The remaining cases of ARM with associated anomalies fall into a miscellaneous group of multiple congenital anomalies, which account for 60% (Cuschieri A. 2002).

2.5 History of treatment

Throughout the centuries, doctors have seen and have tried to treat babies born with imperforate anus. Paulus Aegineta wrote the earliest account of a survivor of surgery for imperforate anus in the 7^{th} century. He suggested rupturing an obstructing membrane with the finger or point of a knife and then dilating the tract until healing was complete. This approach was used for many years. Almost 1000 years later, in 1660, Scultet treated an infant with anal stenosis with dilatation. Formal perineal proctoplasty (ie, mobilization of the bowel through a perineal incision with suturing of it to the skin) was described by Amussat in 1835, and this technique gained rapid acceptance. Imaging to delineate the abnormality was first advocated by Wangensteen and Rice in 1930. Single-stage abdominoperineal procedures became widely used after reports by Rhoads, Pipes, Randall, Norris, Brophy, and Brayton (1948-1949). Stephens (1953) described this procedure and emphasized preservation of the puborectalis muscle. This surgery and its modifications were the standard approach until 1980.

In 1980, the surgical approach to repairing anorectal malformations changed dramatically with the introduction of the posterior sagittal approach (Pena A. 1992). This approach allowed pediatric surgeons to view the anatomy of anorectal malformations clearly and to repair them under direct vision, with better visualization and understanding of the anatomy than previous approaches. Surgeons were able to understand the complex anatomic arrangement of the junction of rectum and genitourinary tract.

2.6 Classification for anorectal anomalies

The most common international classification for anorectal malformations was the Wingspread classification which was elaborated in Wingspread, Wisconsin in 1984 (FD Stephens et al. 1998) as seen in Appendix 1.This classification distinguished between high, intermediate, and low anomalies in the male and female, with special groups established for cloacal and rare malformations. High-type anorectal malformations were subdivided into anorectal agenesis with and without fistula, and rectal atresia. The intermediate malformations were classified as rectovestibular and rectovaginal fistula in the female and rectobulbar fistula in the male. The low-type malformations were classified as anovestibular fistula in the female and, in both sexes as anocutaneous fistula and anal stenosis. This classification was widely accepted over the years and was based on detailed embryological and anatomic studies performed especially by Stephens and Kelly on anatomic sections and radiographic investigations (FD Stephens et al. 1998).

They recognized that the pubococcygeal line extending from the upper border of the os pubis to the os coccyx corresponds with the attachment of levator ani muscles to the pelvic wall, separating high-type malformations lying above the levator muscle and intermediate and low forms of anorectal agenesis lying below this anatomic line. Furthermore, in healthy individuals, the lowest point of the ischial tuberosity, the I-point, represents the deepest point of the funnel of the levator ani muscles. Therefore, every blind rectal pouch, lying between the pubococcygeal line and the I-point, was classified as an intermediate anomaly and could be treated by posterior sagittal anorectoplasty (PSARP) according to DeVries and Pena A (1982). Low lesions below the I-point could be easily managed from a perineal approach. Because of these anatomic relations, the Wingspread classification had a significant impact on the choice of surgical approach.

However, some details of the Wingspread classification remained questionable (Pena A, Hong A. 2000). Some types of anorectal malformations such as rectovaginal fistulas are very rare, and from the surgical point of view, using PSARP in about two thirds of all anorectal malformations, the sex of the patient did not seem important in the choice of the surgical approach. Therefore in 1995, Pena proposed a classification based on the type of the fistula present as shown in Appendix 2 (Pena A, Hong A. 2000).

Pena and Hong presented a critical and detailed analysis of 245 patients from a single centre examined postoperatively (Pena A, Hong A. 2000). His follow-up criteria were also new and included voluntary bowel movement, fecal soiling, constipation, and complete fecal continence. This descriptive and fistula-related grouping became widely

accepted over the past decade. The advantage of the classification by Pena is that the type of the fistula provides information not only about localization of the blind pouch but also on the anticipated extent of mobilization of the atretic rectal segment necessary to perform a sacro or abdominosacroperineal pull-through (Pena A, Hong A. 2000).

It is important to remember that the course of the fistula may vary from one individual to another, can be ascending or descending and of shorter or longer length so that the confluence of the fistula with the urogenital tract or perineum may differ from the lowest point of the blind pouch. This is especially true if the fistula arises from a higher level of the blind-ending rectum and not from its lower most point. Therefore, the classification of Pena does not distinguish between rectovestibular and anovestibular fistulas. However, when performing a PSARP procedure this differentiation did not seem important (Pena A, Hong A. 2000).

By closely comparing both classifications, that is, the Wingspread classification and the suggestions of Pena, it becomes clear that there is no real contradiction between them (Pena A, Levitt M et al. 2004). Perineal and vestibular fistulas could be regarded as low malformations, bulbar fistulas, and imperforate anus without fistula, and most of the vaginal fistulas may be regarded as intermediate-type anomalies, and prostatic and bladder neck fistulas are considered high-type imperforate anus. The same is true for rectal agenesis or stenosis.

2.7 Approach to ARM management

When the diagnosis of anorectal malformation is established at birth, the most important question to be answered is whether or not the baby has an associated defect that is life threatening and requires immediate treatment. Intravenous access is required for fluids and antibiotics. The child is fasted and a nasogastric tube is inserted to keep the stomach decompressed to avoid risk of vomiting and aspiration. This also rules out the presence of oesophageal atresia which occurs in 5% of these cases (Agarwala S et al. 1999).

Figure 14, shows the decision making algorithm for the management of newborn male infants. In 80-90% of boys, perineal inspection and urine analysis provide enough information for the surgeon to decide whether the baby requires a divided colostomy. All those defects which are considered low are treated with a perineal anoplasty or minimal posterior sagittal anorectoplasty. These low defects include perineal fistula with or without a midline raphe subepithelial component, "bucket handle" malformation below which an instrument can be passed, anal stenosis, and anal membrane.

Perineal inspection usually provides more information in female than male patients (Pena A. 2000). The presence of a single perineal orifice means that the infant has a cloaca. The patient is then subjected to a colostomy and / or vaginostomy and vesicostomy and /or any other urinary diversion, when necessary. Figure 15 shows the decision making algorithm for the management of newborn female infants with anorectal malformations.

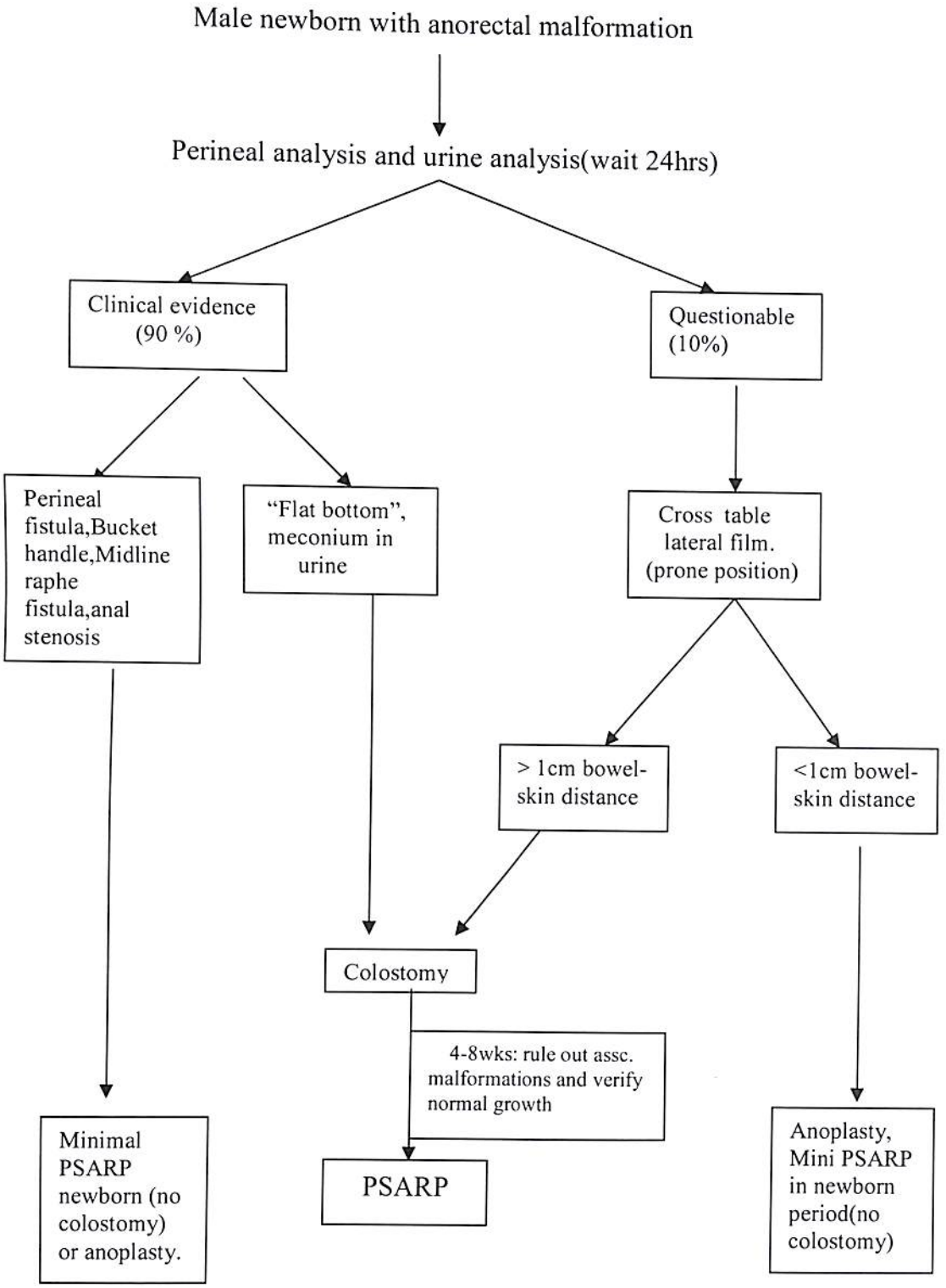

Figure 6: The decision making algorithm for the management of newborn male infants with ARM (adapted from Ashcraft, Pediatric Surgery, 3rd Edition).

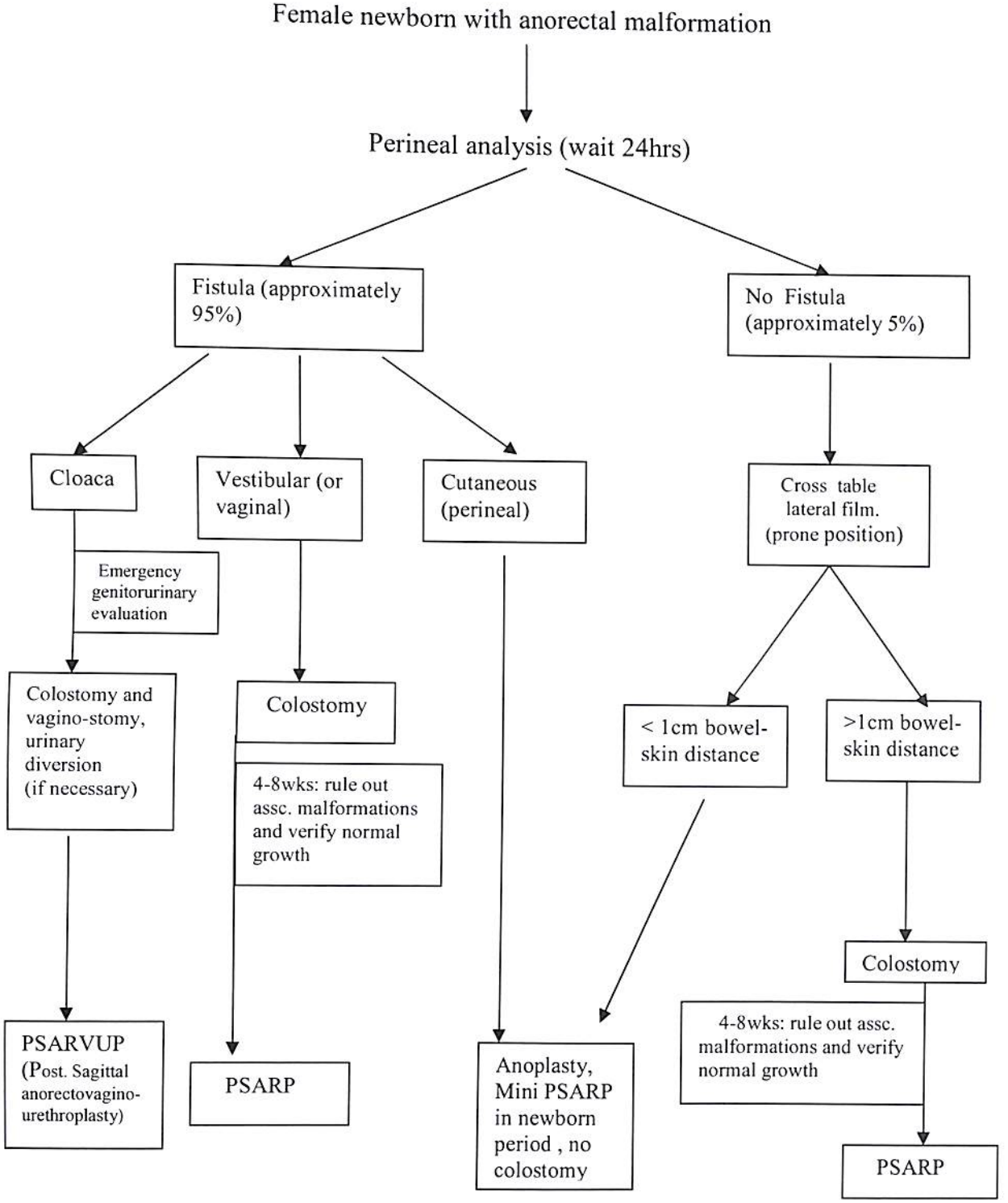

Figure 7: The decision making algorithm for the management of newborn female infants with ARM (adapted from Ashcraft, Pediatric Surgery, 3rd Edition).

2.8 Clinical presentation, diagnosis and initial management

2.8.(i) History of presenting illness

In the labour room, all newborns are subjected to a perineal examination and a thermometer is inserted to check for the patency of the anus. Patients with less severe disease initially may be able to defecate, but over time, constipation, abdominal distension, and occasionally enterocolitis supervene with increasing symptoms. Patients with ectopic anus may present as children or adults. These patients have lifelong histories of constipation and painful defecation and usually have required laxatives or enemas for management. Newborns with high lesions have meconium in the urethra or meconium detected by urine analysis.

2.8.(ii) Physical examination

In most cases, diagnosis is confirmed by examination of the perineum, although low lesions with a perineal fistula or anterior ectopic anus may be more difficult to discern. It is therefore, imperative for the clinician to look carefully at the perineum of the baby in order to see a tiny fistula. There are some hints that will help to suspect the presence of a perineal defect, and those are the presence of a well-formed midline groove and a prominent anal dimple. In addition, a band of skin tissue known as "bucket handle" is usually an indication of the presence of a perineal fistula. A flat bottom in which there is

no midline groove and no anal dimple is a poor prognostic sign and is usually associated with a very high defect (Pena A. 2000).

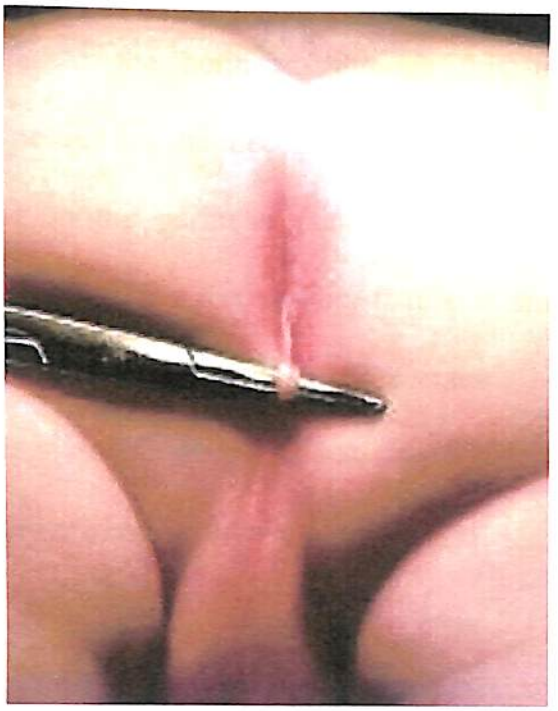

Figure 8 Low ARM with 'bucket handle'deformity.

In either sex, a flat perineum with a short sacrum and little muscle contraction suggests a high anomaly (ie, above the levator muscles). In males, a rectoprostatic or rectourethral fistula may be detected by finding meconium at the urethral meatus or in the urine. Pneumaturia occasionally is observed. In females, a rectal fistula may open at the posterior vestibule or, more rarely, within the vagina. These fistulas almost always are associated with a high lesion. In both males and females, a perineal fistula may be quite small and difficult to detect (Pena A. 2000).

Waiting 24 hours before proceeding with surgical intervention is important to allow gas or meconium to appear at the perineum through this tiny opening. Significant intraluminal pressure is needed for the meconium to be forced through the fistula orifice. If meconium is present on the perineum, that is evidence of a perineal fistula. If meconium is in the urine, there is a rectourinary fistula. A single perineal opening in a female patient implies a cloacal malformation. Concentrate the balance of the physical examination on detecting associated anomalies, which occur in 50-60% of affected children, especially those with high anomalies.

2.9.(i) Imaging studies performed in the newborn period

A patient can be diagnosed with anorectal malformation soon after birth if an adequate perineal examination is done. Occasionally, abdominal radiographs do not show air in the rectum before 24 hours because the rectum is collapsed, and intraluminal pressure has not overcome the muscle tone of the sphincters that surround the lower rectum. A lateral cross table radiograph of the pelvis, the infant in the prone position with the pelvis elevated, and a radiopaque marker placed on the perineum is performed (KA Narasimharao et al. 1983). Rarely, radiography may show the column of air in the distal rectum to be within 1 cm of the perineum. If this is the case, management can be similar to that for rectoperineal fistula and a newborn perineal operation can be performed. If the air column is more than 1 cm from the perineum, a colostomy is indicated. This technique is now used, instead of invertogram in which the infant was held upside down (Pena A. 2000).

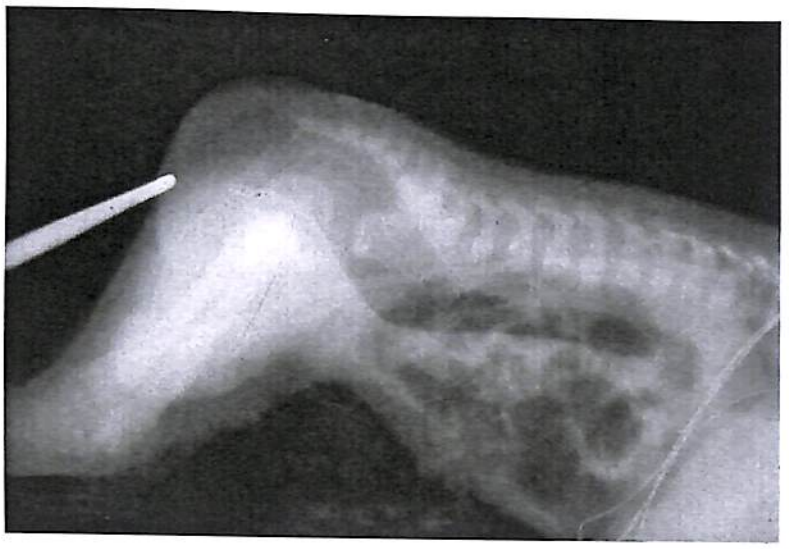

Figure 9. Lateral cross table radiograph with the baby in the prone position to detect high or low type of ARM.

Babygram (X-ray of the whole baby including chest, abdomen and limbs) is done to detect associated anomalies. Plain radiography of the spine can reveal spinal anomalies, such as spina bifida and spinal hemivertebrae. Plain radiography of the sacrum in the anterior-posterior and lateral projections can demonstrate sacral anomalies, such as a hemisacrum and sacral hemivertebrae. Recently, a standardized measurement of the sacral ratio has been proposed (Pena A, Hong A. 2000).This method permits a more objective evaluation of the sacrum and hopefully a better correlation with the final functional prognosis. The sacral length is compared with bony parameters of the pelvis, creating a ratio. A normal ratio in a lateral position is 0.77. Children with anorectal malformations have different degrees of sacral dysplasia that affects this ratio and may run from 0 to 0.77 as shown in Figure 9. Generally children with sacral ratio of less than 0.3 do not develop good bowel control (Pena A. 2000).

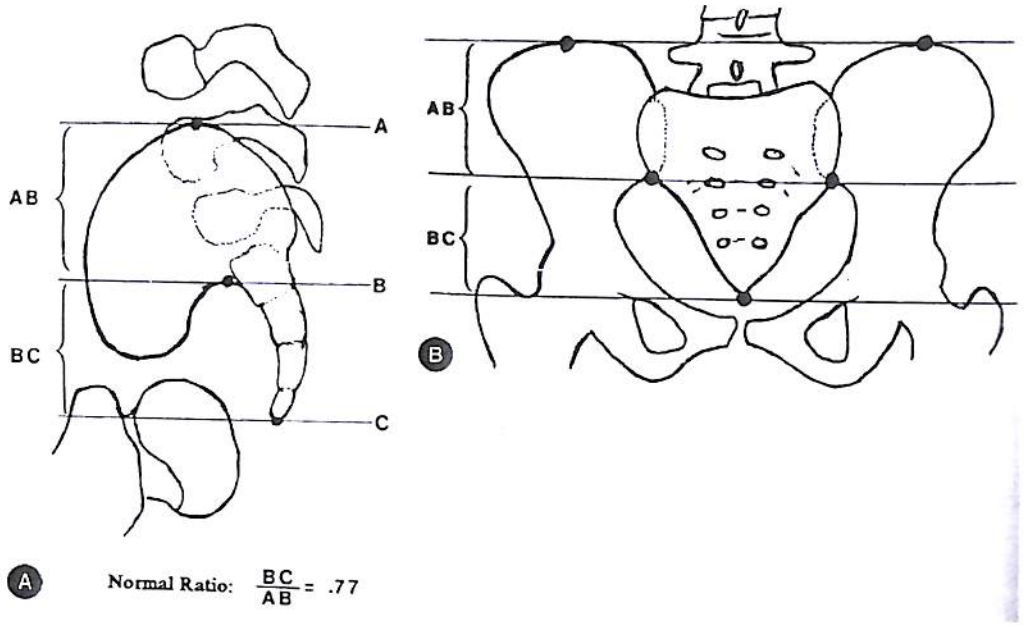

Figure 10. Calculation of sacral ratio, (A) anterior posterior view (B) lateral view (adapted from Rob & Smiths, Operative Surgery, Pediatric surgery, Fifth Edition).

Spinal ultrasonography in the newborn period and up to age 3 months (at which time, the sacrum becomes ossified) can be performed to look for evidence of a tethered spinal cord and other spinal anomalies. An echocardiography should be done to rule out associated cardiac anomalies. The radiologic evaluation of a newborn with imperforate anus includes abdominal ultrasonography to evaluate for urologic anomalies. In cases of persistent cloaca, distended vagina (hydrocolpos) can be identified.

2.9.(ii) Imaging studies performed before definitive surgery

Distal loopogram is performed as an outpatient procedure, after a divided colostomy has been created. Hydrosoluble contrast material is injected into the distal stoma to demonstrate the precise location of the distal rectum and its likely urinary communication. Hydrostatic pressure under fluoroscopic control is required. A Foley catheter is placed in the mucous fistula and the 3-cm^3 balloon is inflated and pulled back to occlude the stoma during contrast injection. The hydrostatic pressure must be high enough by manual syringe injection to overcome the muscle tone of the striated muscle mechanism that surrounds the rectum and keeps it collapsed. This is the best way to demonstrate a rectourinary communication and determine the rectum's true height.

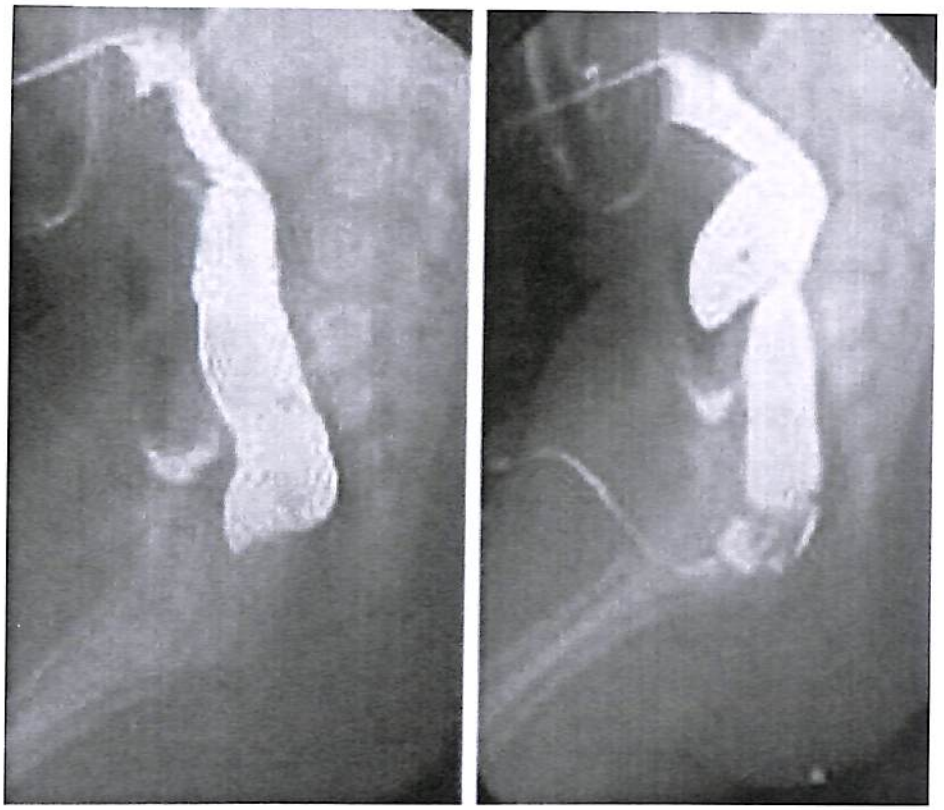

Figure 11. Distal loopogram showing rectourethral fistula.

The contrast material usually fills the proximal urethra and bladder through the fistula. The injection is continued until the child voids, and pictures are taken during micturition to show, in a single picture, the sacrum, height of the rectum, perineum, fistula location, bladder, vesicoureteral reflux (VUR) if present, and urethra. When this study is performed correctly, micturating cystourethrography (MCU) and cystoscopy are not necessary in these babies. MCU is required to determine presence of vesicoureteral reflux.

This study is vital in determining the anatomy so that definitive repair can be planned. In 10% of patients, the fistula is at the level of the bladder neck; in these cases, during the main repair, the surgeon knows that the rectum can be found only through the abdomen, and a combined posterior sagittal and abdominal or laparoscopic approach is used. The anorectal defect of imperforate anus without fistula may also be demonstrated with this radiologic evaluation. This defect occurs in approximately 5% of patients, it has a good functional prognosis, and it is common in individuals with Down syndrome. If the spine was not evaluated with ultrasonography in the newborn period (where clinical stigma of underlying spina bifida eg. hemangioma, hair etc was present) Magnetic resonance imaging (MRI) is necessary after age three months to rule out the presence of tethered cord and other spinal anomalies.

2.10 Associated anomalies

The incidence of associated organ anomalies with ARM depends upon the meticulousness with which they have been searched for by clinical examination and various investigative procedures (Nazer J et al. 2000). The incidence is variously reported from 20% -70% some being minor anomalies but others being life threatening (Hassink EA et al. 1996, Nazer J et al. 2000, Pena A, Hong A. 2000, Niedzielski J. 2000).These associated congenital anomalies not only lead to overall mortality but also contribute to significant morbidity (Cho S et al. 2001, M Endo et al. 1999, Hassink EA et al. 1996, Nazer J et al. 2000). Nievelstein RAJ et al (1998) observed that associated anomalies are present in a high percentage of patients with anorectal malformation and are twice more common in patients with high type of ARM than in patients with low type of ARM.

Though abnormalities of all systems have been described, genitourinary, cardiovascular, gastrointestinal and vertebral anomalies are the ones commonly reported. The well known occurrence of three or more of these anomalies **V**ertebral defects, **A**norectal malformation, **T**racheo-**E**sophageal fistula with oesophageal atresia and **R**enal defects and **R**adial limb dysplasia is not uncommon and has been amalgamated to form the 'VATER' association by Quan and Smith in 1973, later expanded to 'VACTERL' when **C**ardiac and **L**imb defect was added in 1984 by Czeizel et al (Chittmittrapap S et al. 1989).