FACE THE EXAMINER

Anorectal Malformations (Part 1)

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(This section is meant for residents to check their understanding regarding a particular topic)

QUESTIONS

1. What are the various types of anorectal malformations (ARM)?
2. What is the pathophysiology of ARM?
3. What are the clinical features of new-born with anorectal malformation?
4. What are the radiological investigations required for diagnostic evaluation?
5. What are its associated anomalies?

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ANSWERS

Answer 1:

Anorectal malformations comprise a wide spectrum of anomalies of the anorectal system, urogenital system, sacral spine and perineal musculature. The extent of anomalies in these four components decides the type of anorectal malformation.

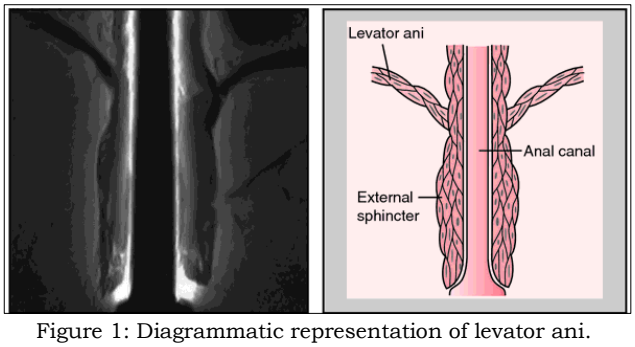

Figure 1: Diagrammatic representation of levator ani.

Gender variations in the type of malformations must also be clearly defined before primary workup and management plan is drawn.

Based on the anatomy, various classifications have been proposed to define the pathology of these anorectal anomalies. The earliest classification dates back to 1953 when Gross proposed a simple differentiation based on the levator muscle (Fig.1), i.e. supralevator – for those above the levator ani or infralevator anomalies, for those below the levator ani. [1]

With advancement in the understanding of the pathology of the malformations, a need was felt to define these lesions more appropriately. During a meeting to celebrate the centenary of the Melbourne Royal Children's Hospital, a new International classification was proposed in 1970 [2,3] as shown in Table 1. This classification utilized the concept of levator ani wherein anomalies above the levator were termed as high and those below were termed as low anomalies, but it also introduced intermediate anomalies which were known as translevator anomalies.

In 1984, during a conference on Ano-rectal malformations organized by Prof. D.Stephens and Prof. D.Smith Wingspread, Wisconsin, another classification was proposed. [4,5] This classification also included the special groups in cloacal and rare malformations as shown in Table 2.

Table 1: International classification of Anorectal anomalies

TYPE	MALE			FEMALE	
High (Supra-levator)	Anorectal agenesis	Without fistula		Without fistula	
		With fistula	Rectovesical Rectourethral	With fistula	Rectovesical Rectocloacal Rectovaginal
	Rectal atresia				
Intermediate	Anal agenesis	Without fistula		Without fistula	
		With fistula	Rectobulbar	With fistula	Rectovaginal - low
					Rectovestibular
Low (Trans-levator)	At normal site	Covered anus - complete Anal stenosis			
	At perineal site	Anterior perineal anus Anocutaneous fistula – covered anus (incomplete)			
	At vulvar site			Vulvar anus Anovulvar fistula Anovestibular fistula	

Table 2: Wingspread classification

	Boys	Girls
High	Anorectal agenesis • With Rectovesical fistula • Without fistula • Rectal atresia	Anorectal agenesis • With rectovaginal fistula • Without fistula • Rectal atresia
Intermediate	• Rectobulbar urethral fistula • Anal agenesis without fistula	• Rectovestibular fistula • Rectovaginal fistula • Anal agenesis without fistula
Low	• Anocutaneous fistula • Anal stenosis	• Anovestibular fistula • Anocuatneous fistula • Anal stenosis
Rare malformations		

Table 3: Pena's classification of ARM [6]

	WITH FISTULA		WITHOUT FISTULA
NON-SYNDROMIC ARM	Rectoperineal malformations	Imperforate anus with recto-urethral fistula • Recto-urethral bulbar fistula • Rectourethral prostatic fistula • Bladder neck fistula	Cloacal malformations with a short common channel (<3cm)
	Imperforate anus in female	Rectovestibual fistula Rectovaginal fistula Cloacal malformation	Cloacal malformations with a long common channel (>3cm)
			H-shaped fistula (rectovaginal)
			Rectal duplication
SYNDROMIC ARM	VACTERL	Pallister-Hall syndrome	Townes-Brock syndrome
	MURCS	Lowe syndrome	Ulnar –mammary syndrome
	OEIS	Heterotaxia	Okihiro syndrome
	Axial mesodermal dysplasia	FG syndrome	Reiger syndrome
	Klippel Feil syndrome	X Linked mental retardation	Hirschsprung's disease
	Sirenomelia-caudal regression	Ciliopathies	Feingold syndrome
	Trisomy 21,13,18	Fraser syndrome	Kabuki syndrome
	Pallister-Killian syndrome	MIDAS syndrome	Optitz BBB/G syndrome
	Cat-eye syndrome	Christian syndrome	Johanson-Blizzard syndrome
	Parental unidisomy 16	Currarino syndrome	Spondylocostal dysostosis
	Deletion 22q11 syndrome	Baller-Gerold syndrome	Short rib-polydactyly syndrome

Table 4: Krickenbeck classification of ARM [7]

MAJOR CLINICAL GROUPS	RARE /REGIONAL VARIANTS
Perineal (cutaneous) fistula	Pouch colon atresia/stenosis
Rectourethral fistula/atresia/stenosis	Rectal atresia/stenosis
Bulbar fistula	Rectovaginal fistula
Prostatic fistula	H-type fistula
Rectovesical fistula	others
Vestibular fistula	
Cloaca	
ARM's with no fistula	
Anal stenosis	

By the early 1980's, several other rare anomalies such as perineal groove, H type of anorectal anomalies, pouch colon, rectal ectasia, rectal atresia, etc. were introduced and documented which were not included in the Wingspread's classification. Also, neither the surgical procedures nor the protocols for assessing post-operative outcome were standardized. Thus, in 1995 Pena introduced a disparate classification system as shown in Table 3.

May 2005, 21 years after the Wingsrpead classification saw the Krickenbeck meeting organized by Professor Alex Holschneider from Cologne, Germany.[7] The goal of the meeting was to develop international criteria for treatment and develop a uniform scoring system for comparable follow-ups. The Pena's classification was modified as per the type of fistula and included rare variants as shown in table 4.

Answer 2:

Embryologically, interference in the development of anorectal and genitourinary organs at various stages upto 7 to 8 weeks of gestation gives rise to a range of anomalies from mild to severe abnormalities involving even the musculoskeletal system of the hindgut. Continued communication between the urogenital tract and rectal portions of the cloacal plate causes rectourethral fistulas or rectovestibular fistulas. [8]

Till date, the accurate embryologic defect causing anorectal malformations still remains undetermined. Nevertheless what is known is that defects in formation or shape of cloacal membrane formation and subsequent breakdown into urogenital and anal openings, which occurs by 8 weeks of gestation, are responsible for the numerous abnormalities of the anorectum. The incorporation of Mullerian ducts, which are formed later, into the anomalous development is also not clear. The pelvic floor as well as the external anal sphincter, derived from exterior mesoderm, is usually present but has varying degrees of anomalies which range from normal musculature to absent muscle complex. The higher the rectal pouch, more are the chances of mal-development of the pelvic floor.

With recent researches in the pathogenesis of anorectal malformations, the previous theories have been discarded. While in the past, defects in lateral fusion were thought to be causative, there is evidence from animal models and from detailed study of human fetuses with major anomalies that a deficiency in the dorsal component of the cloacal membrane and the adjacent dorsal cloaca is causative. A subsequent malfunction of the primitive streak and tail bud in the early development phase around 3-4 weeks has been proposed (yet to be clearly defined) as causation for associated anomalies of the pelvic floor.

The histological analysis of specimens from human fetuses with non-viable malformations revealed the following findings: [9]

1. Primarily, the mal-development affects the anal canal and rectum is secondarily affected.
2. There is ventral displacement of anal canal which opens either on the perineum or forms a fistula to urogenital tract.

Table 5: Clinical features in ARM in newborn at birth

		BOYS	GIRLS
History	Perineum – Pelvic floor	Absent/present anus	
		Specks of meconium in anal region	
		Failure to pass meconium	
		Meconuria	Passing meconium through introitus
		Flaturia	
	External Genitalia	Normal or abnormal	
	Abdomen	Vomiting	
		Distension	
		Distension with visible peristalsis	
	Others	Any other abnormality	
		Family history	
Examination	Pelvic floor	Absence or presence of anal opening	
		Position of anus – normal or anteposed	
		Bulge in perineum on crying or straining	
		Anal dimple	
		Shape of buttocks	
		Anal reflex	
		Perineal groove	
		Bucket handle deformity	
		Meconium or mucus running up the median scrotal raphe	
	Genitals	Phallus - Normal or hypospadiac	Appearance of external genitalia + labia normal or shortened (cloaca)
		Meconium staining at urethral meatus	Number of openings in vestibule • Single opening – cloaca • 2 openings – rectovaginal fistula/rectovestibular fistula with absent vagina • 3 openings – anovestibular fistula
		Testis descended/undescended	
		Any other abnormality	
	Abdomen	Large visible loop occupying more than half of abdomen	
		Palpable kidney/any other palpable lump – solid or cystic	
			Hydrocolpos – palpable lump in lower abdomen
	Lumbo-sacral spine	Occult or obvious spinal dysraphism	
		Absent sacral vertebrae of variable levels	
	Other associated anomalies – as described below		

3. Those malformations in which a fistula is not demonstrated, a rudimentary partly regressed connection is found on histology.

4. In those with fistula from rectum to urogenital structures, there is a gradual transition of the anal mucosa to urogenital mucosa.

5. In proximal fistulae, the development of trigone of bladder, the upper urethra and the urethral sphincter is also abnormal in males whereas in females, vaginal development is inappropriate causing a urogenital sinus caudal to mesonephric ducts (as seen in persistent cloaca).

6. With deficient anal canal, the striated muscles of the perineum often have abnormal configuration. The longitudinal fibers of external anal sphincter are concentrated medially, the bulbospongiosus muscle is displaced medially in high lesions and the puborectalis sling, the external urethral sphincter and ischiocavernous muscles are variably affected depending on the severity of the lesion.

7. The likelihood of associated abnormalities in the development of pelvis, perineum, bladder, ureters, phallus etc. were proportional to the length of agenesis as measured from the actual anal site.

Answer 3:

A thorough clinical assessment (substantiated with radiological assessment when needed) is essential for accurately classifying the malformation as the choice of surgical treatment is largely dependent on the extent of the anomaly.

The important aspects in history and clinical examination are listed in tabular form as shown in Table 5.

Table 6: Radiological assessment in newborns with Anorectal malformations

TYPE OF TEST	TIMING		INTERPRETATION
	BOYS	**GIRLS**	
Plain X-Ray Abdomen Erect	At birth/at the time of prone cross table lateral (PCTL) xray [10]	If massive distension of abdomen and failure to pass meconium	• Multiple dilated bowel loops with air-fluid levels and absent rectal gas • Large dilated loop with A-F level (pouch colon)
X-ray lumbo-sacral spine -	At time of PCTL	At birth/at time of pre-operative work-up	• Measure sacral ratios • Sacral defects • Hemivertebrae • Presacral masses
PCTL X-ray	12-18 hours after birth or later if presentation is after 18 hours	Not done	Presence of rectal gas shadow : • Low anomalies – below the M line • Intermediate anomalies – above the M line and below the PC line • High anomalies – above the PC line Other features : • Air in bladder • Beaking of terminal rectal pouch (fistula)
Invertogram [11]	Obsolete	Not done	
Ultrasonography	Abdomen	Abdomen and Pelvis	• Urological anomalies especially hydroureteronephrosis (VUR), Hydronephrosis, absent kidney, etc. • Presence or absence of uterus/ovaries in females as well as e/o hydro/hydrometrocolpos
	Cardiac – 2D Echo - Congenital Cardiac anomalies		
	Spine – screening for occult spinal malformations		
MRI – abdomen and pelvis [12,13,14]	• Delineates the level of anomaly • Provides information about the fistula • Pelvic floor musculature – puborectalis sling, external anal sphincter anatomy is clearly defined • Anomalies of spine, spinal cord, urogenital system can be simultaneously diagnosed		

Table 7: Associated anomalies in ARM

SYSTEM	TYPE OF ANOMALY	FREQUENCY
Urinary	Vesico-ureteric reflux Hydronephrosis Renal agenesis Renal dysplasia	50%
Genital	Vaginal septum	50%
	Uterine didelphys/Bicornuate uterus	35%
	Cryptorchidism	3-19%
	Vaginal duplication/vaginal agenesis/absent ovary	
Vertebral [15]	Lumbosacral anomalies Tethered cord Cord lipomas Syringohydromyelia	30-35%
Cardiovascular	VSD Tetralogy of Fallot Transposition of great vessels Hypoplastic left heart syndrome	12-22%
Gastrointestinal [16]	Tracheo-esophageal fistula	10%
	Duodenal obstruction	
	Malrotation	
	Hirschsprung's disease	
Curarino triad [17,18]	- Sacral defect + presacral mass + imperforate anus	> 350 cases reported in literature
Other anomalies	As listed in Pena's classification	Rare

Answer 4:

Assessment of the type of anomaly often needs radiological assistance in the form of x-rays or ultrasonography. Few associated anomalies also need to be investigated at the time of birth, especially the genitourinary and cardiac lesions.

The timing and method of radiological investigations are tabulated as in Table 6 as follows.

Answer 5:

Anorectal malformations present with a high incidence of associated anomalies. The anomalies are presented in a tabular form in the decreasing order of frequency as shown in table 7.

REFERENCES

1. Gross RE. Malformations of the anus and rectum. In: Gross RE (ed). The surgery of infancy and childhood. WB Saunders, Philadelphia, 1953: 348-57.
2. Nixon HH. Anorectal anomalies: with an international proposed classification. Postgrad Med J. 1972; 48:465-70.
3. Santulli TV, Kiesewetter WB, Bill AH Jr. Anorectal anomalies: A suggested international classification. J Pediatr Surg. 1970; 5:281-7.
4. Upadhyaya V, Gangopadhyay A, Srivastava P, Hasan Z, Sharma S. Evolution of management of anorectal malformation through the ages. Internet J Surg. 2007; 17:1.
5. Wakhlu AK. Management of Congenital Anorectal malformations. Indian Pediatr 1995; 32:1339-42.
6. Levitt MA, Peña A. Anorectal malformations. Orphanet J Rare Dis. 2007; 2:33.
7. Gupta DK. Anorectal malformations - Wingspread to Krickenbeck. J Indian Assoc of Pediatr Surg. 2005; 10:75-7.
8. Pediatric Imperforate anus. Medscape. Available from http://www.emdeicine.medscape.com /article/929904-overview-accessed on 01/01/15
9. Van der Putte. The development of the perineum in the human. A comprehensive histological study with a special reference to the role of stromal components. Adv Anat Embryol Cell Biol. 2005; 177:1-131.
10. Narasimharao KL, Prasad GR, Katariya S, Yadav K, Mitra SK, Pathak IC. Prone cross-table lateral view: an alternative to the invertogram in imperforate anus. Am J Roentgenol. 1983; 140:227-9.
11. Wangensteen OH, Rice CO. Imperforate anus: a method of determining the surgical approach. Ann Surg. 1930; 92:77-81.
12. Nievelstein RA, Vos A, Valk J. MR imaging of anorectal malformations and associated anomalies. Eur Radiol. 1998; 8:573-81.
13. Mchugh K, Dudley NE, Tam P. Pre-operative MRI of anorectal anomalies in the newborn period. Pediatr Radiol. 1995; 25:33-6.

14. Sato Y, Pringle KC, Bergman RA, et al. Congenital anorectal anomalies. MR imaging. Radiol. 1988; 168:157-62.
15. Stathopoulos E, Muehlethaler V, Rais M, Alamo L, Dushi G, Frey P, et al. Preoperative assessment of neurovesical function in children with anorectal malformation: association with vertebral and spinal malformations. J Urol. 2012; 188:943-7.
16. Rosenbaum DG, Kasdorf E, Renjen P, Brill P, Kovanlikaya A. Sling left pulmonary artery with patent type IIA tracheobronchial anomaly and imperforate anus. Clin Imag. 2014; 38:743-6.
17. Lee SC, Chun YS, Jung SE, et al. Currarino triad: anorectal malformation, sacral bony abnormality, and presacral mass--a review of 11 cases. J Pediatr Surg. 1997; 32:58-61.
18. Versteegh HP, Feitz WF, van Lindert EJ, Marcelis C, de Blaauw I. "This bicycle gives me a headache", a congenital anomaly. BMC Res Notes. 2013; 6:412.

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