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 newborn with anorectal malformation?
4. What are the radiological investigations required for diagnostic evaluation?
5. What are its associated anomalies?

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**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.

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

Figure 1: Diagrammatic representation of levator ani.
### Table 2: Wingspread classification

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

**Rare malformations**

### Table 3: Pena’s classification of ARM [6]

| NON-SYNDROMIC ARM | WITH FISTULA | WITHOUT FISTULA |
|-------------------|-------------|-----------------|
| 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 | Rectovestibular 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 unidi-sony 16 | Curarrino syndrome  | Spondylocostal dysostosis  |
| | Deletion 22q11 syndrome | Baller-Gerold syndrome  | Short rib-polydactyly syndrome  |
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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

| History | BOYS | GIRLS |
|---------|------|-------|
| Perineum – Pelvic floor | Absent/present anus | Specks of meconium in anal region |
| | Failure to pass meconium | Passing meconium through introitus |
| | Meconuria | Flaturia |
| External Genitalia | Normal or abnormal | |
| Abdomen | Vomiting | Distension |
| | Distension with visible peristalsis | |
| Others | Any other abnormality | |
| | Family history | |

| Pelvic floor | BOYS | GIRLS |
|--------------|------|-------|
| Absence or presence of anal opening | Appearance of external genitalia + labia normal or shortened (cloaca) |
| Position of anus – normal or anteposed | Number of openings in vestibule |
| Bulge in perineum on crying or straining | • Single opening – cloaca |
| Anal dimple | • 2 openings – rectovaginal fistula/rectovestibular fistula with absent vagina |
| Shape of buttocks | • 3 openings – anovestibular fistula |
| Anal reflex | |
| Perineal groove | |
| Bucket handle deformity | |
| Meconium or mucus running up the median scrotal raphe | |

| Genitals | BOYS | GIRLS |
|----------|------|-------|
| 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 | BOYS | GIRLS |
|---------|------|-------|
| 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 | BOYS | GIRLS |
|-------------------|------|-------|
| 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 medi- ally 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                                                                 |
|-------------------------------|---------------------------------------------|-------------------------------------------------------------------------------|
| Plain X-Ray Abdomen Erect     | At birth/at the time of prone cross table   | Multiple dilated bowel loops with air-fluid levels and absent rectal gas      |
|                               | lateral (PCTL) xray [10]                    | Large dilated loop with A-F level (pouch colon)                               |
| X-ray lumbo-sacral spine      | At time of PCTL                             | Measure sacral ratios                                                        |
|                               |                                             | Sacral defects                                                                |
|                               |                                             | Hemivertebrae                                                                 |
|                               |                                             | Presacral masses                                                              |
| PCTL X-ray                    | 12-18 hours after birth or later if         | Presence of rectal gas shadow:                                               |
|                               | presentation is after 18 hours              | 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                                    | Urological anomalies especially hydronephrosis (VUR), Hydronephrosis, absent  |
|                               |                                             | kidney, etc.                                                                  |
| Ultrasonography               | Abdomen and Pelvis                          | Presence or absence of uterus/ovaries in females as well as e/o hydrometro    |
|                               |                                             | colpos                                                                      |
| Cardiac – 2D Echo             |                                             | Congenital Cardiac anomalies                                                 |
| Spine – screening for occult  |                                             |                                                                             |
| spinal malformations          |                                             |                                                                             |
| MRI – abdomen and pelvis      | Delineates the level of anomaly             |                                                                             |
| [12,13,14]                    | 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-ureteral reflux                               | 50%                  |
|               | Hydronephrosis                                       |                      |
|               | Renal agenesis                                       |                      |
|               | Renal dysplasia                                      |                      |
| Genital       | Vaginal septum                                       | 50%                  |
|               | Uterine didelphys/Bicornuate uterus                  | 35%                  |
|               | Cryptorchidism                                       | 3-19%                |
|               | Vaginal duplication/vaginal agenesis/absent ovary    |                      |
| Vertebral [15]| Lumbosacral anomalies                                | 30-35%               |
|               | Tethered cord                                         |                      |
|               | Cord lipomas                                          |                      |
|               | Syringohydromyelia                                   |                      |
| Cardiovascular| VSD                                                  | 12-22%               |
|               | Tetralogy of Fallot                                   |                      |
|               | Transposition of great vessels                        |                      |
|               | Hypoplastic left heart syndrome                       |                      |
| 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.

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