Dr. D.Y. PATIL VIDYAPEETH, PUNE
(DEEMED UNIVERSITY)

CURRARINO SYNDROME – A NOTEWORTHY DIFFERENTIAL IN ACQUIRED MEGACOLON

UNIT 5

DEPARTMENT OF GENERAL SURGERY

DR DY PATIL MEDICAL COLLEGE AND HOSPITAL, PUNE

INTRODUCTION

- **Currarino syndrome (CS) is a congenital condition encompassing the presence of an anorectal malformation, a pre-sacral tumour and a sacral deformity.**
- The triad was named after an American radiologist named "Guido Currarino,"
- **The condition is rarely observed in adults and higher frequencies were reported in females.**
- **Incidence : 9 cases of Classic CS have been recorded till date with 8 cases being children and only 1 case reported in adult Age group.**

INTRODUCTION

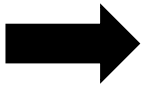
- Usually indolent course but may have a more dramatic presentation especially in cases with :
  - **Anorectal malformations**
  - **Complications e.g.: Meningitis.**

- **We hereby present a case of a young female presented with chronic constipation, symptoms mimicking ultra-short segment Hirschsprung's disease and the challenges in arriving at its true diagnosis**

CASE REPORT

- A **16Y/Female** presented to our center with complaints of **chronic constipation** since **8 years**.

- The treatment modalities offered over the 8 years varied included:
  - Use of Laxatives
  - Dietary modifications
  - Multiple enema instillations



Temporary resolution of symptoms

- **H/O weakness, fatigue and loss of appetite since 3 years.**

- **H/O crampy - lower abdominal pain** – Resolved with medication.

- **H/O distension of abdomen** since **3 years** - **Relieved by finger evacuation of feces**.

CASE REPORT

- **No H/O vomiting**

- **H/O increased urinary frequency** since **1 year** – No burning micturition.
- No H/O recurrent UTI/ fever.

- No H/O PR bleed.
- No H/O tenesmus.

- No H/O trauma.
- No H/O lower limb weakness / tingling.

- No menstrual abnormalities.

CASE REPORT - EXAMINATION

- **General Examination:**
  - Average built
  - Pulse: 90/min
  - BP: 130/70 mmHg
  - **Pallor** ++
  - No icterus/clubbing/generalized lymphadenopathy/ Pedal edema

CASE REPORT - EXAMINATION

- **Per abdomen examination :**
  - Soft, non-tender abdomen
  - **Mild distension +**
  - **Vague lump ~ 5*6 cm – Left iliac fossa – Firm - No visible peristalsis.**
  - Bowel sounds - **Hyperperistaltic**

- **Digital Rectal Examination:**
  - **Decreased resting anal tone** – No fissure/hemorrhoid
  - **Anal squeeze – Reduced asymmetrically**

CASE REPORT - INVESTIGATIONS

- **Nervous system examination:**
  - Motor system – Normal
  - Sensory system
    - **Diminished perianal sensations**
    - **Decreased sensations over S2-S4 dermatomes (Mainly fine touch)**
    - No other focal neurological deficits
  - Spine – Normal (No pit/swelling/tuft of hair)

Biochemical investigations

Severe iron deficiency anemia

(Hb: 4.8g/dl)

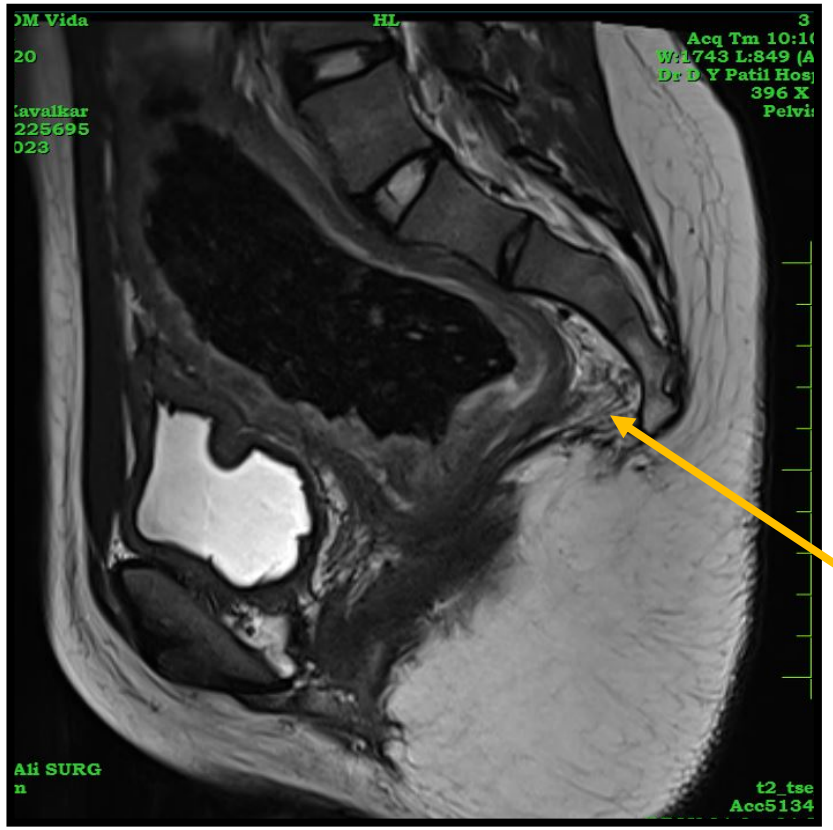
Ultrasonography (USG) - Abdomen/Pelvis

Distended large bowel loops

No free fluid

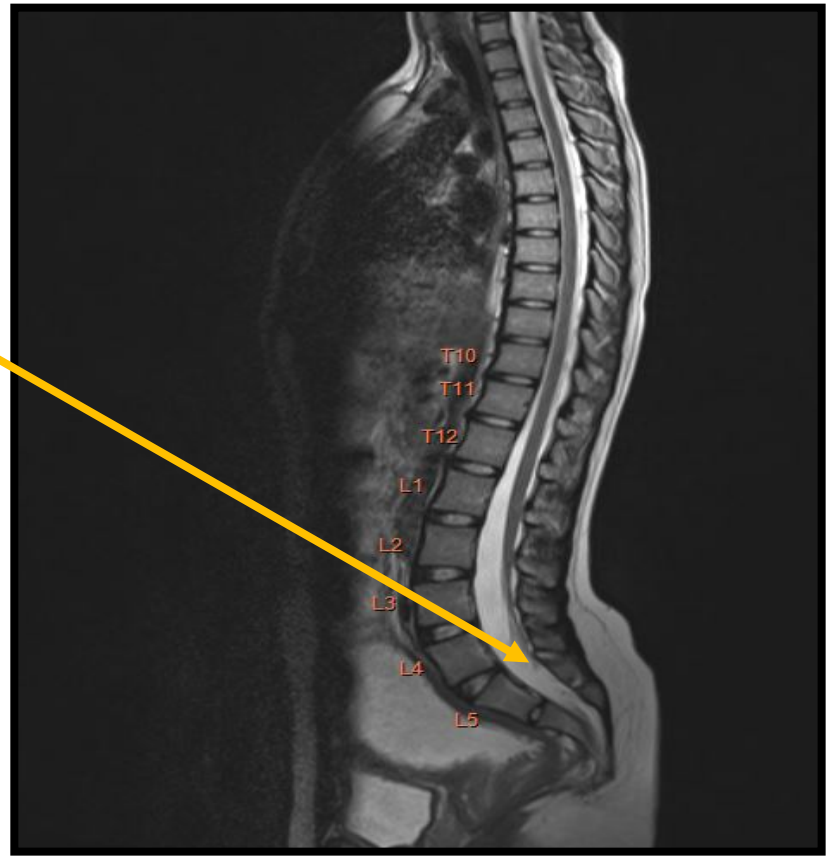
Magnetic Resonance Imaging (MRI) Pelvis + Myelography

- **Left hemi-sacral agenesis with preservation of S1**
- **Small heterogeneous, non-contrast enhancing mass anterior to L5-S1** with caliber of 1.5 × 1.5cm with no diffusion restriction on diffusion weighted image **-? Anterior meningocele**
- **Significantly dilated sigmoid + Descending colon with fecal impaction.**
- **Tethering of cord at L5-S1**



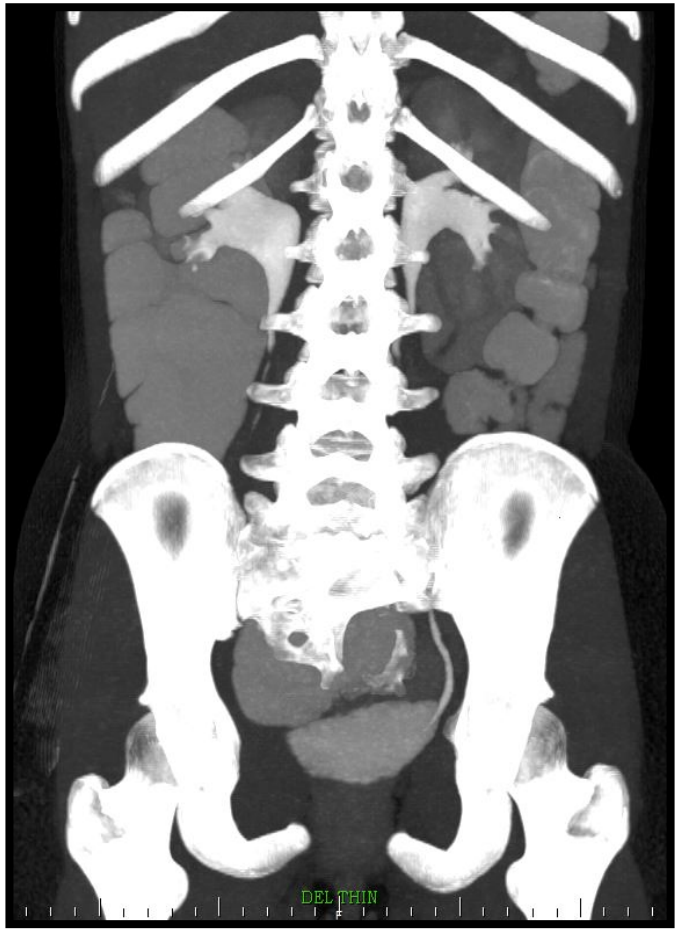
MR Pelvis showing pre-sacral mass (? Anterior meningocele) with upstream dilatation of colon

MR Myelography showing tethering of cord at L5-S1

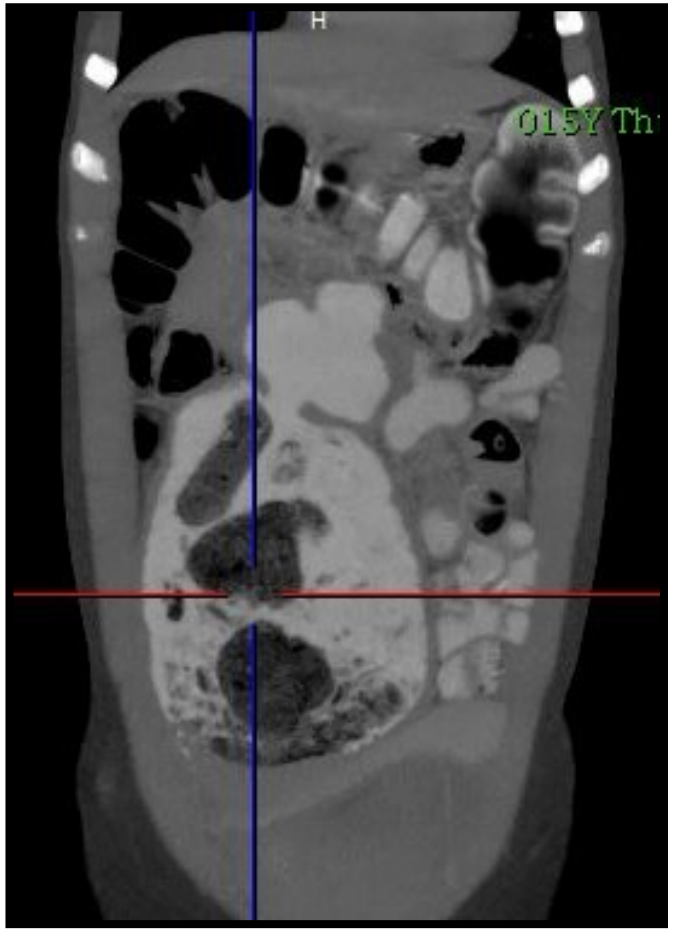


Contrast Enhanced Computed Tomography (CECT)- Abdomen/Pelvis

- **Marked dilatation of the sigmoid colon and rectum with fecal impaction**
- **Pre-sacral mass lesion ~ 1.5*0.8 cm opposite L5-S1 vertebrae with no post-contrast enhancement.**



CECT Pelvis – Left scimitar sacrum with preservation of S1 vertebra



CECT Pelvis showing dilated sigmoid colon with fecal impaction

CASE REPORT - INVESTIGATIONS

Colonoscopy

- **Normal mucosa - No mass lesion**
- No extrinsic compression
- **? Transient narrowing at the distal rectum – Full Thickness biopsies obtained (No e/o Hirschsprung's disease)**

Anorectal manometry

- **Type II Dyssynergia**
- **Absence of urge at 500ml**
- **No e/o Hirschsprung's disease**

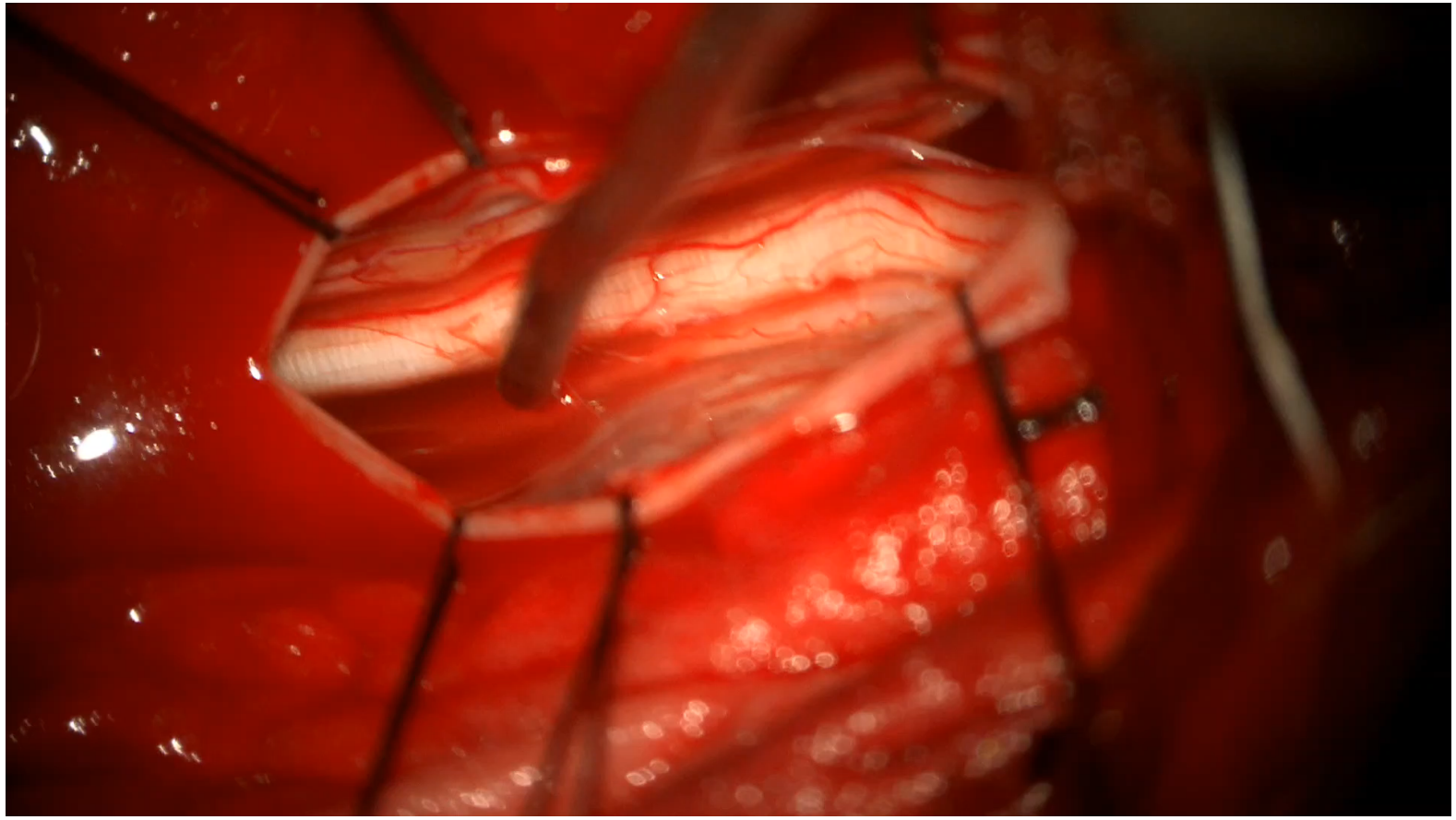
CASE REPORT - MANAGEMENT

- Based on all investigations performed the diagnosis of **Mild Currarino Syndrome**
- In conjunction with our Neurosurgery department – A staggered approach was preferred

STEP 1 : Release of the cord tethering
STEP 2 : Excision of pre - sacral mass SOS
Colostomy
STEP 3 : Reversal of colostomy

CASE REPORT - MANAGEMENT

- **Intra-operative findings:**
  - Posterior midline approach.
  - **Sacrum was exposed and Laminectomy done for S2,3,4 vertebrae**
  - **Release of cord tether was performed**
  - Water tight closure of dural tube was achieved at the terminal end.



CASE REPORT - MANAGEMENT

- There was no neurological deficit in the immediate postoperative period.
- The post-operative period was uneventful and **patient passed stools freely** on POD 3
- **Patient was discharged on POD 15** after removal of sutures

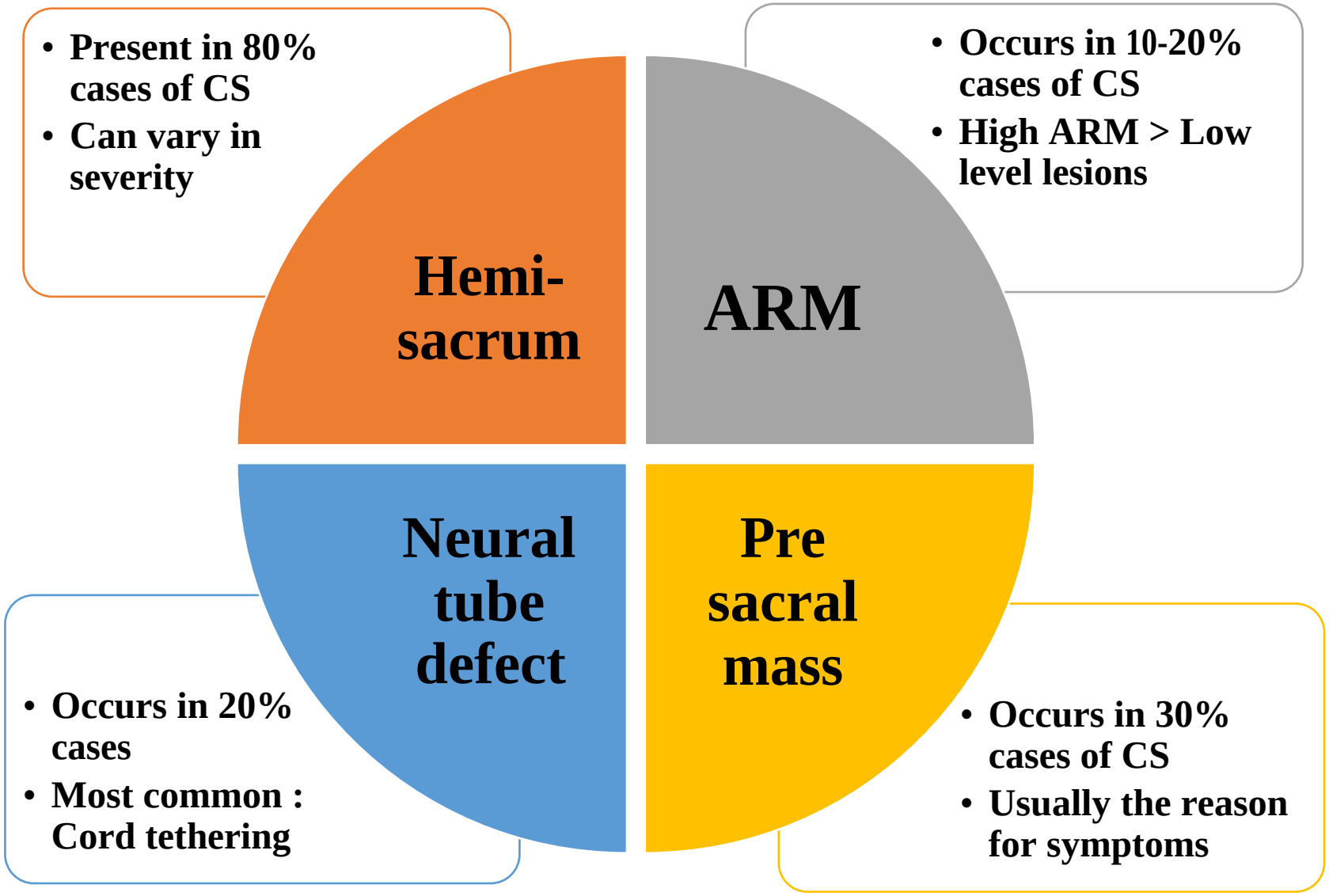
- **Further surgical management is planned for later sitting - Patient has maintained regular follow up**

- **Improvement in neurological deficit**
    - **Return of rectal sensation.**
    - **Easier evacuation of feces with no need for finger evacuation.**

DISCUSSION

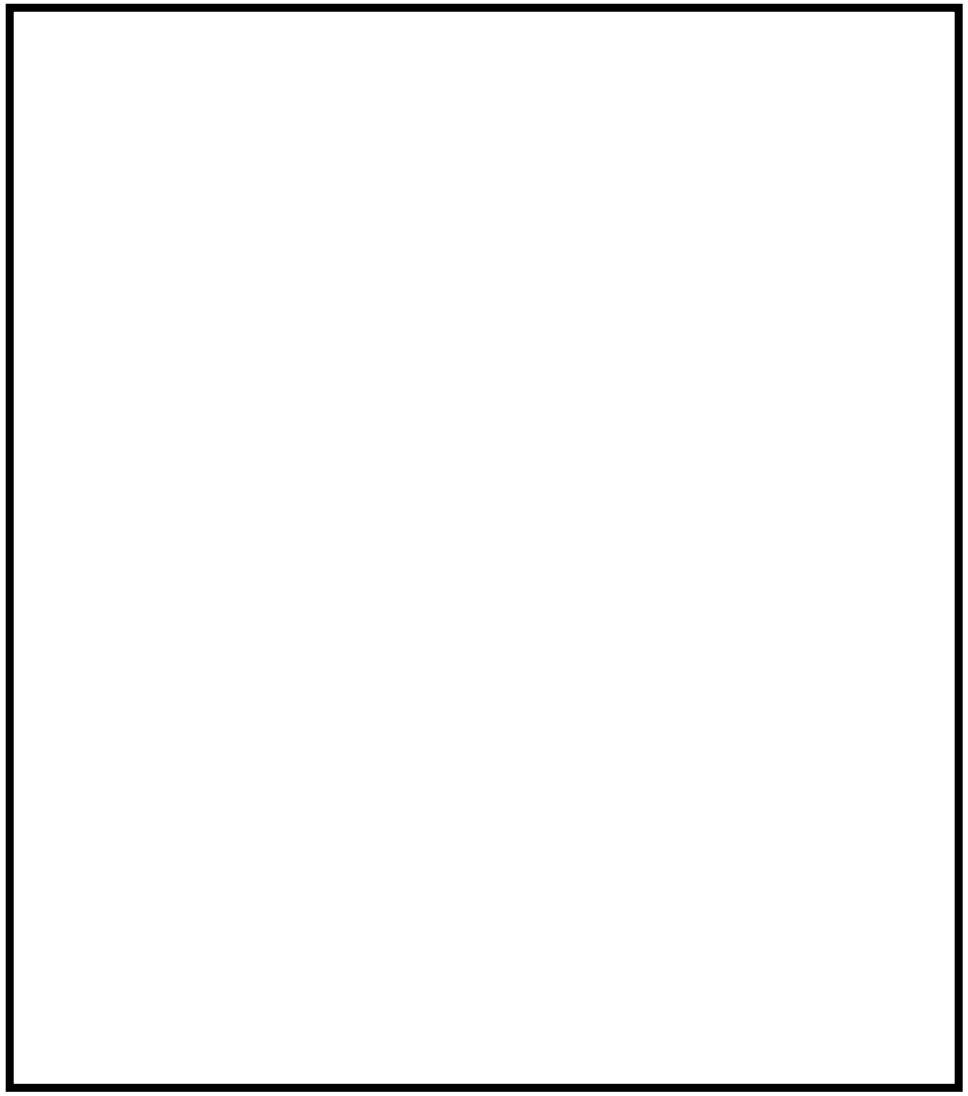
- **Currarino syndrome - First described by Currarino in 1981 - Triad of sacral defect, anorectal malformation and a presacral mass**
- Target gene : **HLXB9 homeobox gene**
- **Variable gene penetrance - Plausible rationale for the presence of incomplete manifestations of a disease, wherein certain clinical features may be absent or not fully expressed**
- **The patient of Currarino syndrome may be diagnosed in the neonatal period** - But the lesion may remain asymptomatic or be manifested **diagnosis is usually made late in childhood.**

Complete CS (Classic)	**Mild CS :** **Hemi-sacrum + Presacral mass +/- NTD**
Complete CS	**Minimal CS :**


Hemi-sacrum
ARM
Neural tube defect
Pre sacral mass
• Present in 80% cases of CS
• Can vary in severity
• Occurs in 10-20% cases of CS
• High ARM > Low level lesions
• Occurs in 20% cases
• Most common : Cord tethering
• Occurs in 30% cases of CS
• Usually the reason for symptoms

Hemi-sacrum

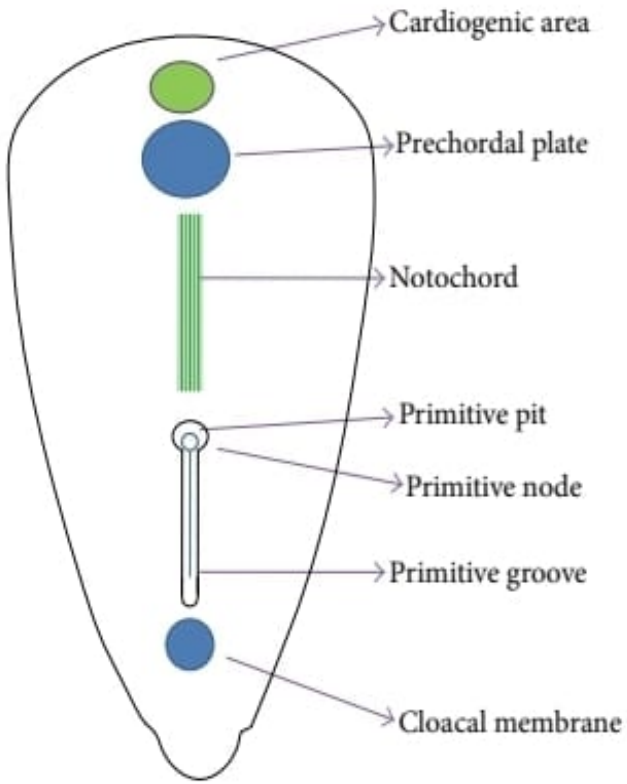
- Sacral defects - Sacral agenesis with the peculiarity - **Sparing of the first sacral vertebra.**
- **However, the sacral scimitar remains the characteristic sacral anomaly associated with CS**
- **CT – Investigation of choice for sacral defect delineation**



Types of sacral anomalies in CS – Type IV in our case

DISCUSSION

EMBRYOLOGICAL BASIS



HLXB9 gene on 7q36 (chromosome 7q36) - Major causative gene in Currarino syndrome

Positive in approximately 30% of the cases

Theories proposed by Lui et.al. and Currarino:

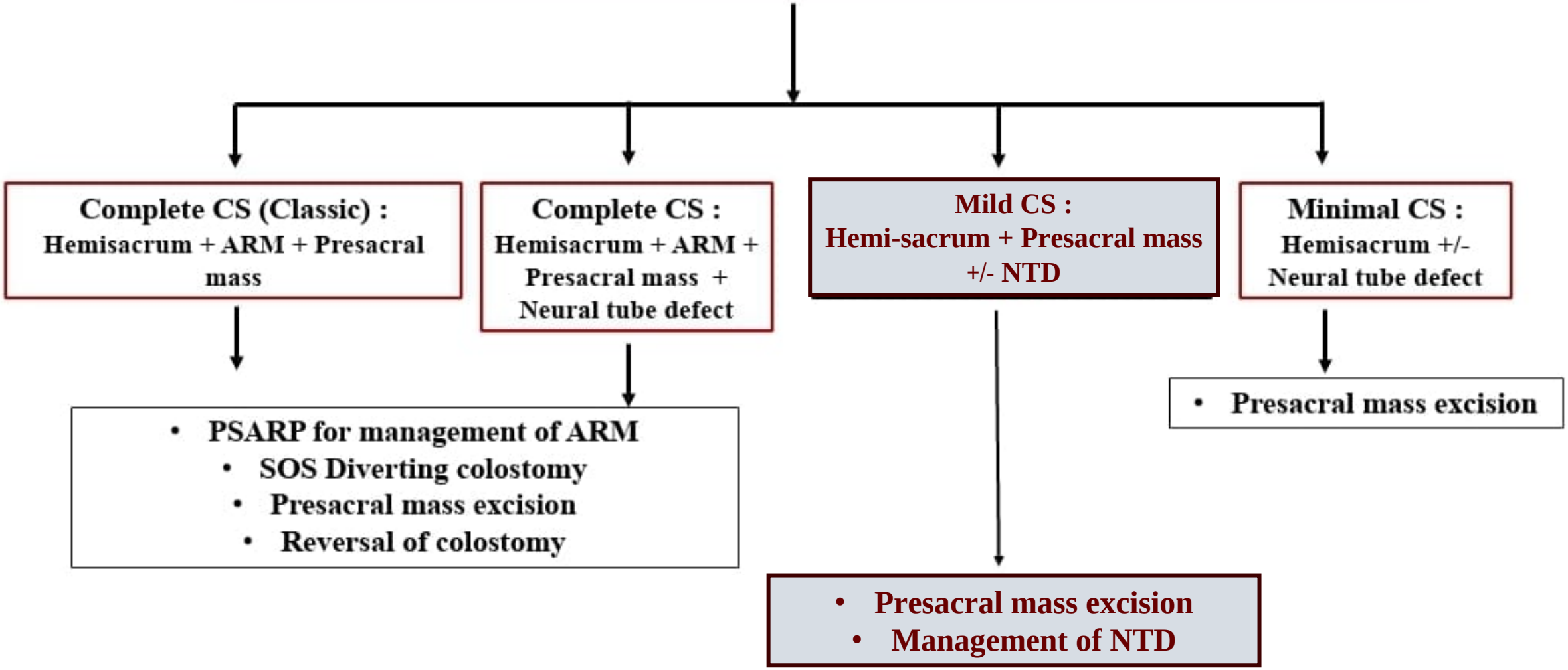
- **Failure of migration of epiblasts from primitive node during notochord formation**
- **Adhesions between the primitive gut and notochord can give rise to leak of CSF and anorectal malformations**

DISCUSSION

- In order to mitigate the diagnostic challenge associated with identifying surgical causes of constipation, **it is imperative to consistently adhere to the established methodology for examination.**
-
- **The management of Currarino syndrome involves :**
  - **Surgical removal of the presacral mass**
  - **Correction of any associated anorectal deformity using the posterior sagittal ano-rectoplasty (PSARP) technique.**

- **The implementation of staged surgery is recommended as a preventive measure to mitigate the potential risk of meningitis.**

MANAGEMENT PROTOCOL FOR CURRARINO SYNDROME



CONCLUSION

- Currarino Syndrome constitutes a rare and notorious trifecta deeming itself a **worthy differential whilst handling cases with chronic constipation in pediatric as well as adolescent age groups.**

- With dominant differentials like Hirschsprung's disease for chronic constipation, physicians and surgeons alike should keep other rarer causes like CS in mind.

- **Despite having an armamentarium of investigation a detailed history and thorough physical examination holds the key to diagnosis of rarest of rare cases.**

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THANK YOU