Malrotation

Heterotaxia and Congenital Heart Disease

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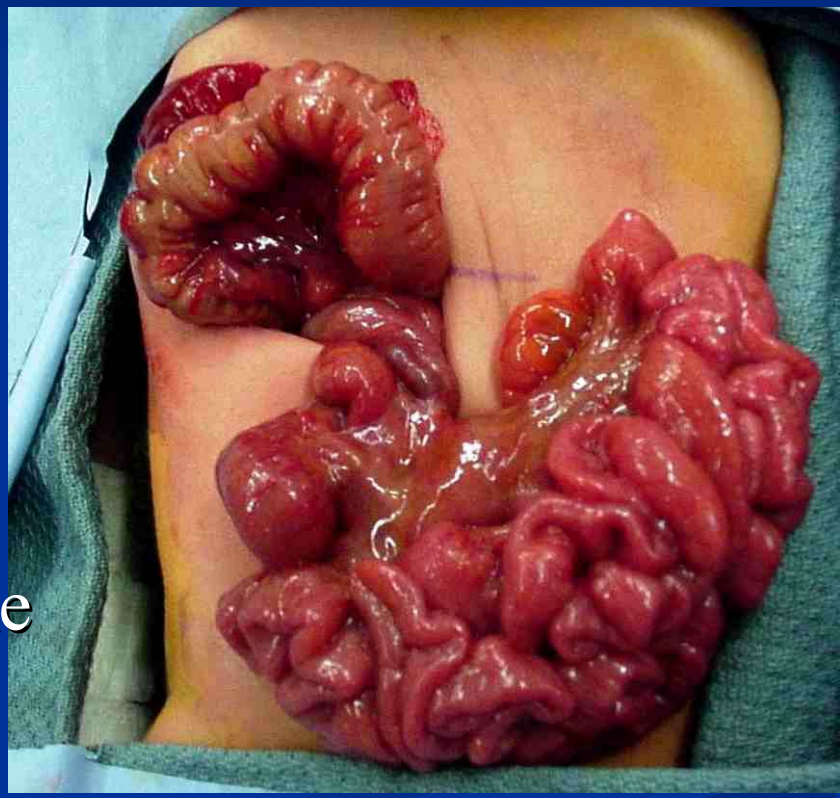
Disclosure of Relevant Financial Relationships

I have no relevant financial relationships with commercial interests to disclose.

The planning committee for this event has no relevant financial relationships with commercial interests to disclose.

William Ladd

- 1932
  - William Ladd describes 10 cases of malrotation and volvulus and treatment by counterclockwise detorsion
- 1936
  - Describes 21 additional cases, emphasizing the division of the bands over the duodenum and placing the cecum in the left upper quadrant



Incidence

- Autopsy prevalence is as high as 0.5 to 1%
- Incidence of symptoms leading to clinical discovery is 1 per 4000 live births
- Males > Females
- 50 to 75% of those who become symptomatic do so in the first month of life
- 90% of clinical symptoms occur in children under one year of age

First Stage: Herniation

- Middle portion of growing intestine begins to herniate into the body stalk at 6 weeks
- As it herniates, it undergoes a 180 degree counterclockwise twist around the SMA
- The proximal limb elongates faster than the distal limb
- The colon remains relatively straight.

Second Stage: Return to the Abdomen

- Undergoes a further 90 degree rotation counterclockwise to make the total rotation 270 degrees counterclockwise
- The “pre-arterial” limb enters first
- The terminal ileum and cecum last

Third Stage: Fixation

- Fixation occurs from 12 weeks until after birth
- Portions of the mesentery fuse with the posterior peritoneum

Normal Anatomy

Abnormal Rotation

Stage I

Stage II (Classic Malrotation)

Stage III

Second Stage Anomalies: Return to the Abdomen

- Nonrotation
  - Rotates through 90 degrees only, instead of 270 degrees
  - Distal limb (colon) enters first instead of last , colon on the left; small bowel on the right
  - 0.5% of autopsies; twice as frequently in males than females
  - Clockwise volvulus is the main danger

Nonrotation

- In “non-rotation,” the cecum is in the left lower quadrant and the base of the small bowel mesentery is broad and not predisposed to volvulus. These children can be safely observed. While volvulus can occur, surgery does not decrease risk. Unfortunately, it is often impossible to determine non-rotation without direct visualization of the bowel.

Second Stage Anomalies: Return to the Abdomen

- Malrotation
  - Rotates only 180 degrees, instead of 270 degrees
  - Terminal ileum enter abdomen first
  - Cecum is in a subhepatic location on the right

Second Stage Anomalies: Return to the Abdomen

- Malrotation
  - Ladd bands (peritoneal bands) attach the cecocolic loop to the posterior abdominal wall

Second Stage Anomalies: Return to the Abdomen

- Ladd's bands may compress the second part of the non-rotated duodenum
- Entire midgut being suspended on the superior mesenteric vessels on a narrow stalk
  - This mesentery is prone to volvulus

Second Stage Anomalies: Return to the Abdomen

2 Other Rare Variants: reversed rotation and hyper-rotation

- Reversed rotation
  - In stage 1 the intestine rotates 90 degrees counterclockwise (instead of 180)
  - In stage 2 the intestine rotates 180 degrees clockwise (i.e., the final 180 degrees of rotation is clockwise instead of counterclockwise)

Second Stage Anomalies: Return to the Abdomen

- Reversed rotation (2 subtypes)
  - If the colon enters first, the colon is posterior to the SMA
    - This can cause obstruction of the transverse colon and is usually presents in adulthood
  - If the small bowel enters first, the small bowel is on the left and anterior to the artery and the colon is on the right
    - Opposite of nonrotation

Second Stage Anomalies: Return to the Abdomen

- Hyperrotation
  - Rotation continues through 360 or 450 degrees
    - Instead of stopping at 270 degrees
  - Cecum in pelvis or in area of splenic flexure of the colon in the LUQ

Third Stage: Failure of Fixation

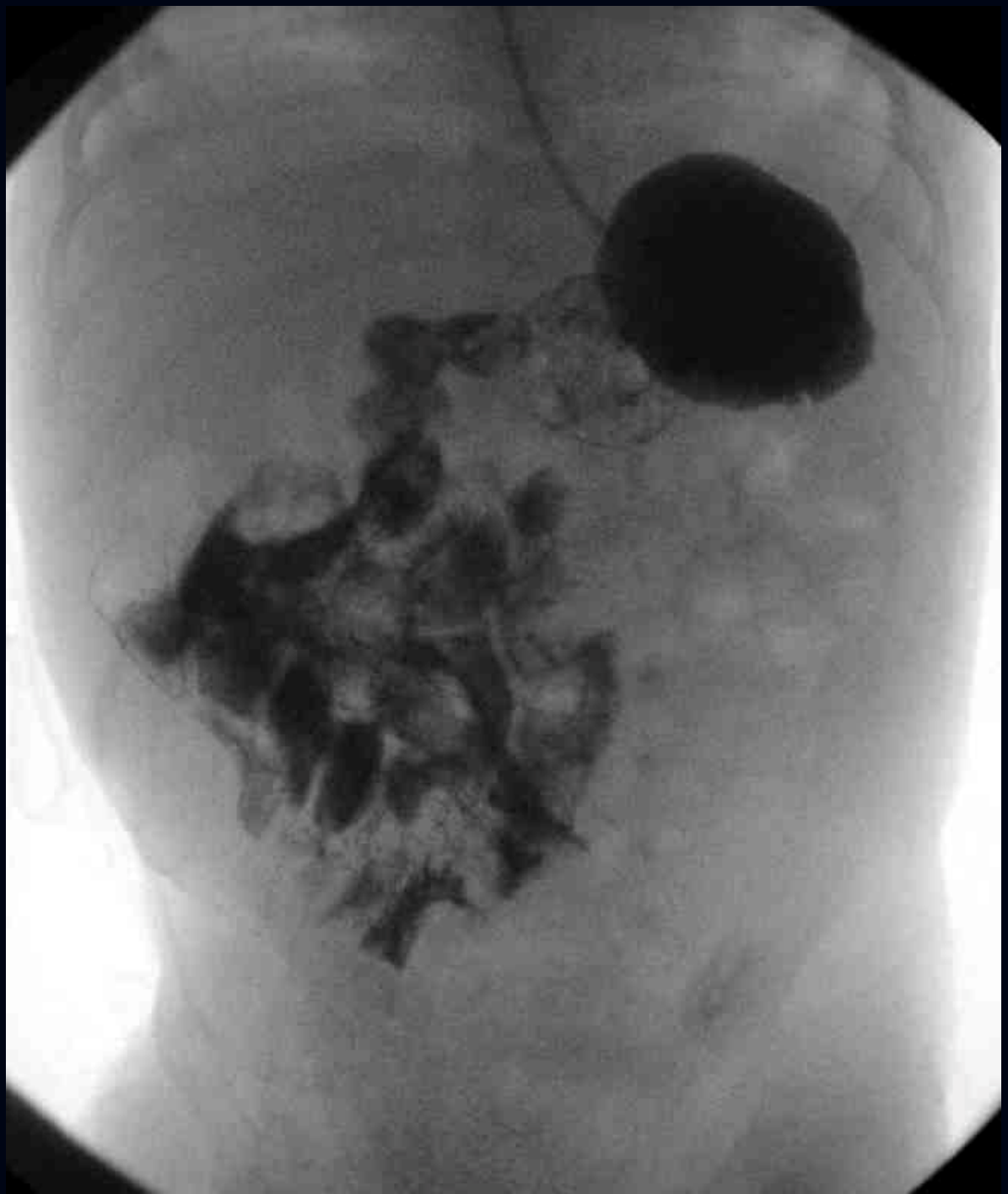
- Normal rotation, but cecum and ascending colon are not fixed
- Present in as many as 10% of asymptomatic individuals,
- more common in females (mobile cecum)
- May allow cecal volvulus
  - Associated with Cornelia de Lange Syndrome
  - Other syndromes

Clinical Aspects

- Presentation
  - Bilious Emesis (95%)
  - Volvulus
    - May present acutely (usually in first year of life) or as chronic intermittent or partial obstruction
  - Duodenal obstruction
  - Internal herniation

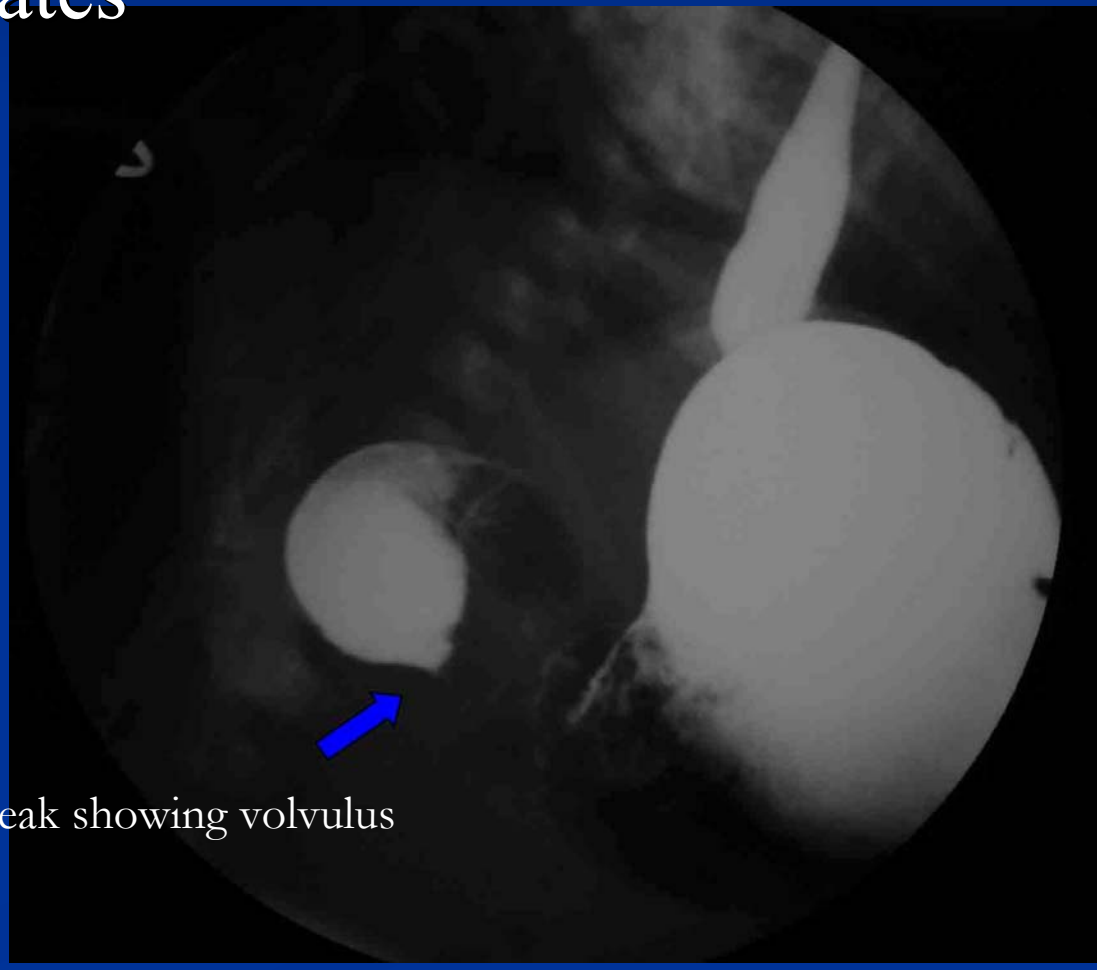
Clinical Aspects

- UGI
  - Abnormal position of ligament of Treitz
  - Duodenal obstruction
- BE
  - Abnormally placed cecum
  - Though normal placement does not Rule out malrotation)
  - Transverse colon obstruction if volvulus



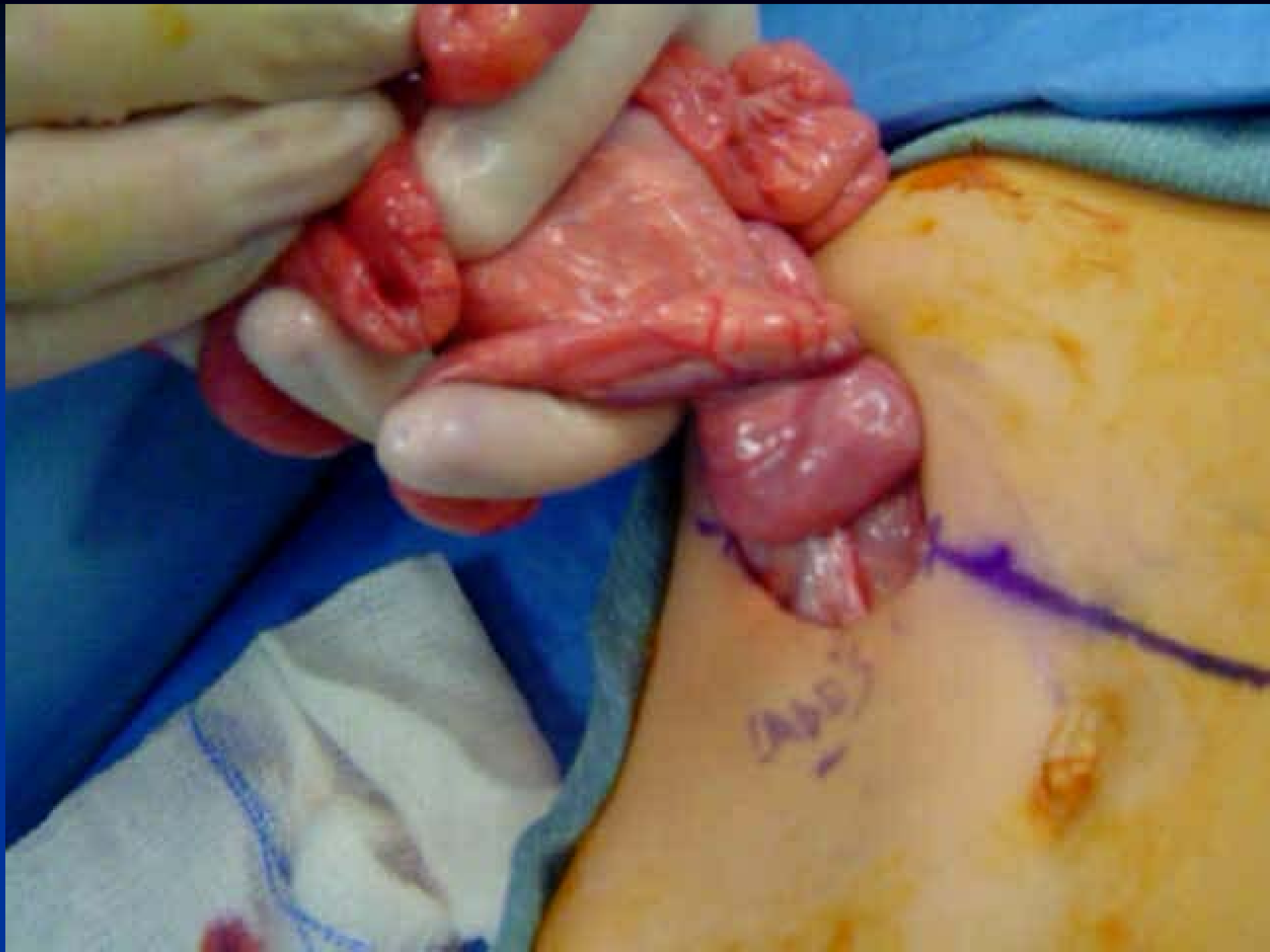
Volvulus

- 60 to 80% of neonates with Malrotation develop Volvulus



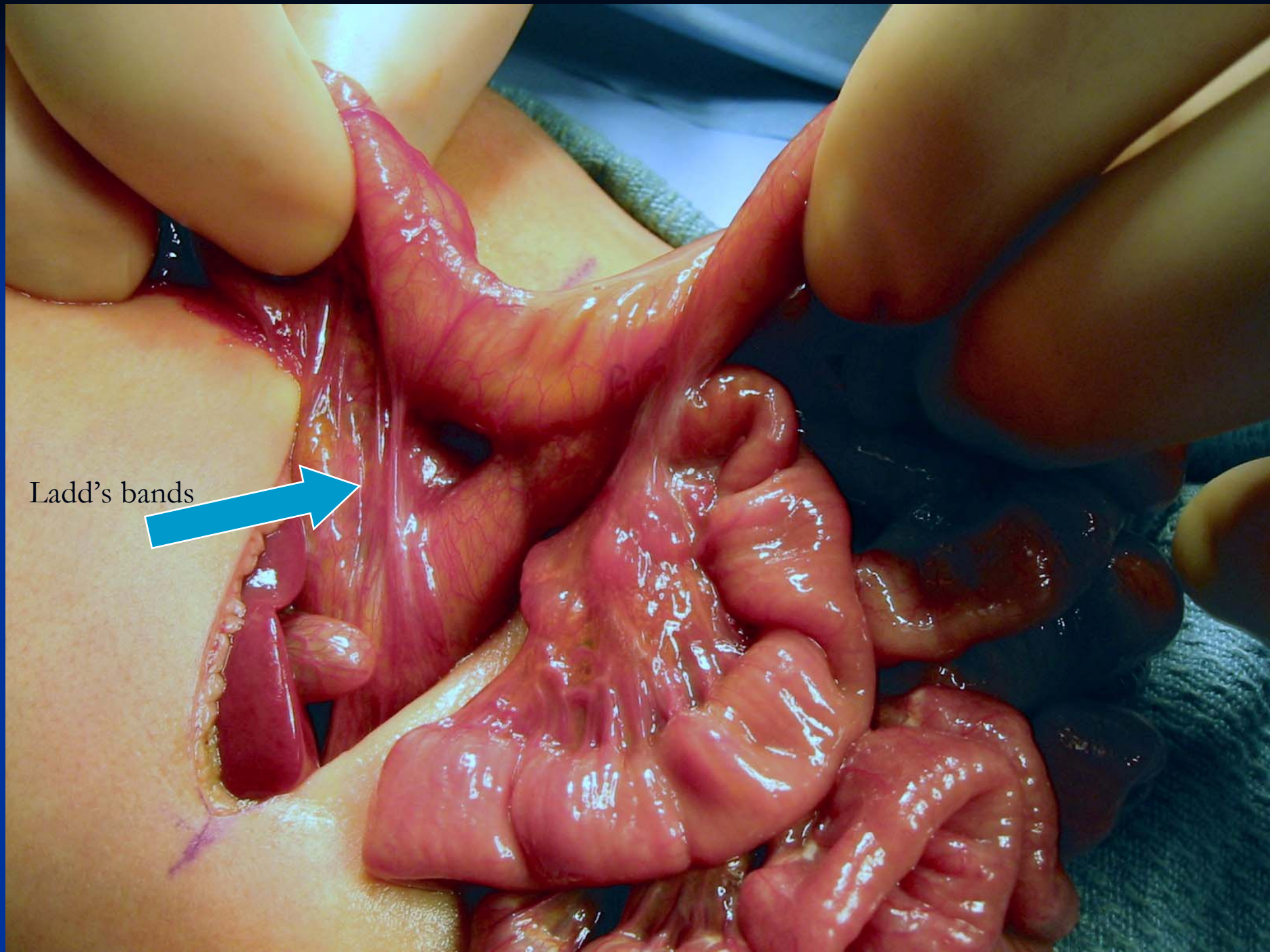
Bird's beak showing volvulus

Video of Volvulus



Management

- Ladd's procedure
  - Bands obstructing the duodenum are divided
  - Bowel placed in position of nonrotation (small intestine on right, large bowel on the left, cecum in the left hypochondrium
  - Appendectomy (not described by Ladd)
  - No fixation


Ladd's bands

How common do patients with Malrotation have CHD?

- One study:
  - 27.1% of all the patients with malrotation to have a major or minor CHD.
  - Major CHD was seen in almost half
- 284 patients with malrotation
  - 93 of the 284 (33%) patients had CHD
    - 15 patients had (PPHN), 14/15 with CDH
    - Closer to 27%
- Stewart et al. found only 12% with CHD

Type of CHD

Table 1 Types of major and minor congenital cardiovascular defects

Major CCVD	*n* = 37 (100%)	Minor CCVD	*n* = 40 (100%)
Complex cardiovascular defects	*n* = 18 (48.6%)	Ventricular septal defect	*n* = 16 (40.0%)
Tetralogy of Fallot	*n* = 7 (18.9%)	Atrium septal defect	*n* = 7 (17.5%)
Hypoplastic left heart syndrome	*n* = 3 (8.1%)	Peripheral pulmonary stenosis	*n* = 4 (10.0%)
Atrioventricular septal defect	*n* = 3 (8.1%)	Persistent ductus arteriosus	*n* = 3 (7.5%)
Pulmonary valve atresia	*n* = 1 (2.7%)	Pulmonary valve stenosis	*n* = 3 (7.5%)
Transposition of the great arteries	*n* = 1 (2.7%)	Aortic valve stenosis	*n* = 2 (5.0%)
Tricuspid valve atresia	*n* = 1 (2.7%)	Aortic coarctation	*n* = 1 (2.5%)
Common arterial trunk	*n* = 1 (2.7%)	Hypertrophic cardiomyopathy	*n* = 1 (2.5%)
Aortic arch hypoplasia	*n* = 1 (2.7%)	Subvalvular aortic valve stenosis	*n* = 1 (2.5%)
Dextrocardia	*n* = 1 (2.7%)	Left superior caval vein	*n* = 1 (2.5%)
		Arteria lusoria	*n* = 1 (2.5%)

CCVD Congenital cardiovascular defects

What is the Risk of Volvulus?

- 72 pts with CHD and volvulus
- 28 operated for acute surgery for malrotation, at a median age of 4.0 days (range 0–455).
- 5 patients had ischemic bowel disease at time of acute IM intervention;
  - 3 of these patients died, 1 due to massive intestinal necrosis
- Risk: 38% volvulus, 7% Ischemic

M. Kouwenberg Æ R. S. V. M. Severijnen Æ L. Kapusta Congenital cardiovascular defects in children with intestinal Malrotation Pediatr Surg Int (2008) 24:257–263

Post-op Complications in Patients with CHD and Ladd's

- Post malrotation complication rate 61%
  - No difference between patients with major and minor CHD.
- Symptomatic CHD signs before malrotation surgery increased post-malrotation morbidity significantly (OR 4.0, 95% CI 1.4–11.0).
- 8 fold increased risk of complication if corrected for age.
- 20% patients died, almost half due to cardiovascular cause.
- Mortality risk was increased by intestinal ischemia

Post-op Complications in Patients with CHD and Ladd's

- Thirteen patients (18%) needed re-laparotomy
  - small bowel obstruction (n=5)
  - Leakage of the bowel anastomosis (n = 4),
  - Wound dehiscence = 2
  - feeding difficulties (n = 1)
  - necrotizing enterocolitis (n = 1).
- Risk of SBO after Ladd's = 19%

Etiology of Association

- The gut and the heart are both organs that show a characteristic left–right asymmetry
- Two signaling molecules for cardiac and gastrointestinal asymmetry
  - nodal
  - pitx2c [1]
- Reduced, diffuse or absent expression of pitx2c in the embryo cause abnormal looping from both heart and gut in animal studies [1]

[1]Logan M, Pagan-Westphal SM, Smith DM, et al (1998) The transcription factor Pitx2c mediates situs-specific morphologenesis in response to left-right asymmetric signals. Cell 94:307–317

Heterotaxia

- Also called “Situs Ambiguous”
- Any anatomical configuration that differs from complete situs solitus and from situs inversus totalis
- Heterotaxia is a continuum of anatomical configurations that extend from isolated dextrocardia with abdominal situs solitus to the total absence of asymmetry along the left-to right body axis that is seen in left isomerism (polysplenia) or right isomerism (asplenia)

Heterotaxia

Right-sided Liver Left-sided Spleen 1 “Right” Lungs on right 1 “Left” Lung on left Levocardia	Midline Liver Asplenia 2 “Right” Lungs ?cardiac anomaly	Midline Liver Polysplenia 2 “Left” Lungs ?cardiac anomaly	Left-sided Liver Right-sided Spleen 1 “Right” Lungs on Left 1 “Left” Lung on Right Dextrocardia

Situs Solitus

Situs Inversus

Heterotaxia

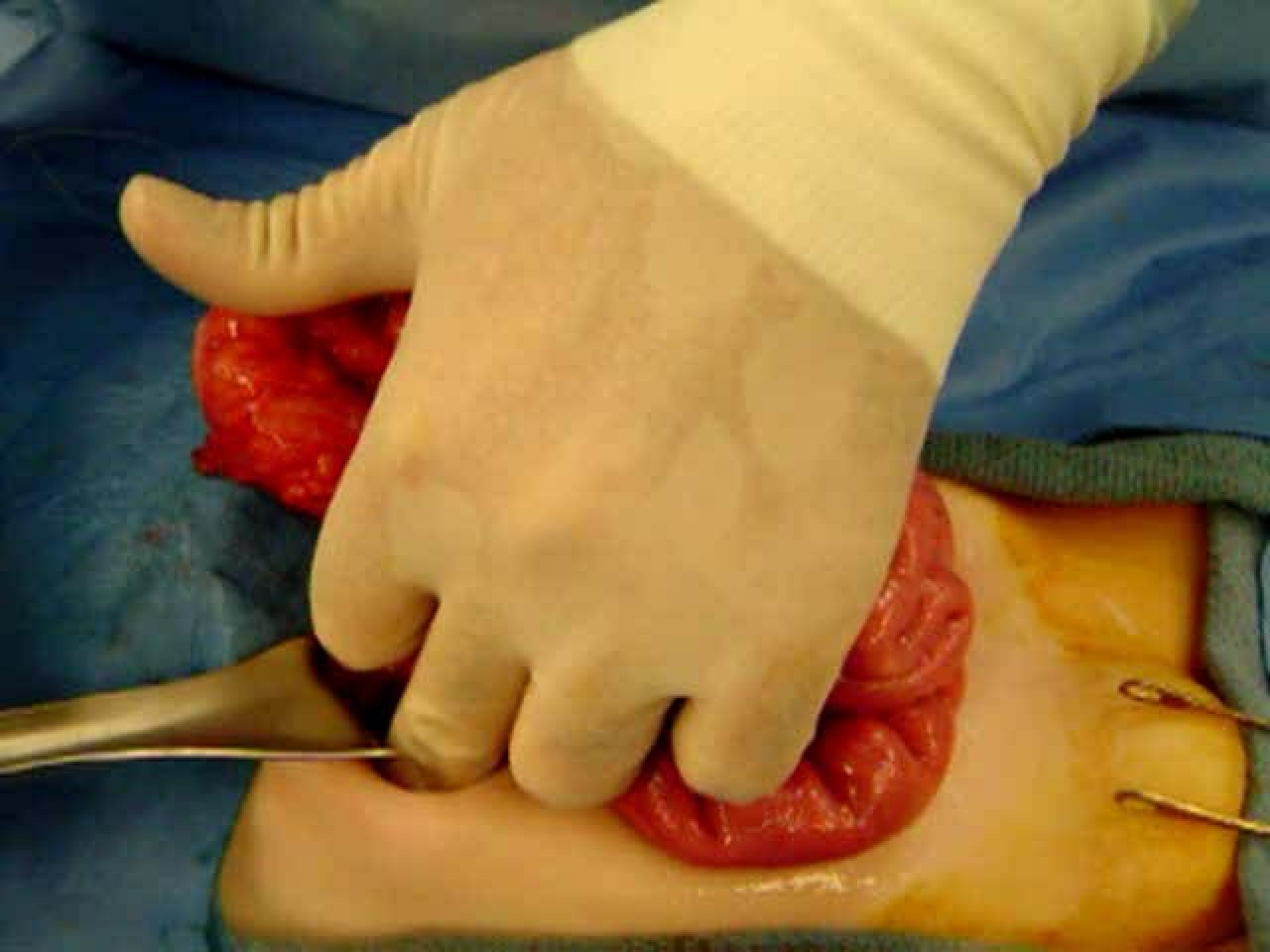
- Congenital heart disease (CHD) is present in 90% or more of patients with heterotaxia,
- Severity of CHD represents a spectrum
  - from isolated simple lesions, such as ASD,
  - complex defects, (functional single ventricles, anomalous systemic and/or pulmonary venous return).
- Patients who typically present with the most severe forms of CHD are those with asplenia and right atrial isomerism.

Case Report

- 4 patients with malrotation and heterotaxia
- All 4 developed midgut volvulus occurred in unsuspected malrotation
- This lead to a recommendation for screening all patients with heterotaxia

Chang J, Brueckner M, Touloukian RJ. Intestinal rotation and fixation abnormalities in heterotaxia: early detection and management. J Pediatr Surg 1993;28:1281- 5.

Video of Heterotaxia with Malrotation



Heterotaxia and Malrotation

- The problem with screening
  - normal rotation and malrotation is a spectrum of rotational abnormalities
- Results
  - operating on all patients with intestinal rotational abnormalities because there are no clear criteria to determine which rotational abnormalities predispose to volvulus.

Malrotation spectrum

- Recently, it has been suggested that not all rotational abnormalities predispose to volvulus
- Mehall: 201 children who underwent surgery for malrotation were classified based on the position of the ligament of Treitz

Management of typical and atypical intestinal malrotation . Journal of Pediatric Surgery , Volume 37 , Issue 8 , Pages 1169 – 1172 J . Mehall

Malrotation spectrum

- Malrotation was classified as ‘typical’ if the ligament of Treitz was to the right of the midline.
- Atypical variants were classified as
  - High: ligament of Treitz was at or left of the midline and above T12
  - Low: ligament was at or left of the midline and below the T12
- 16% of patients with typical malrotation were found to have a volvulus
- 2% of those with either a low or high ligament of Treitz had a volvulus.

Borenstein & Langer

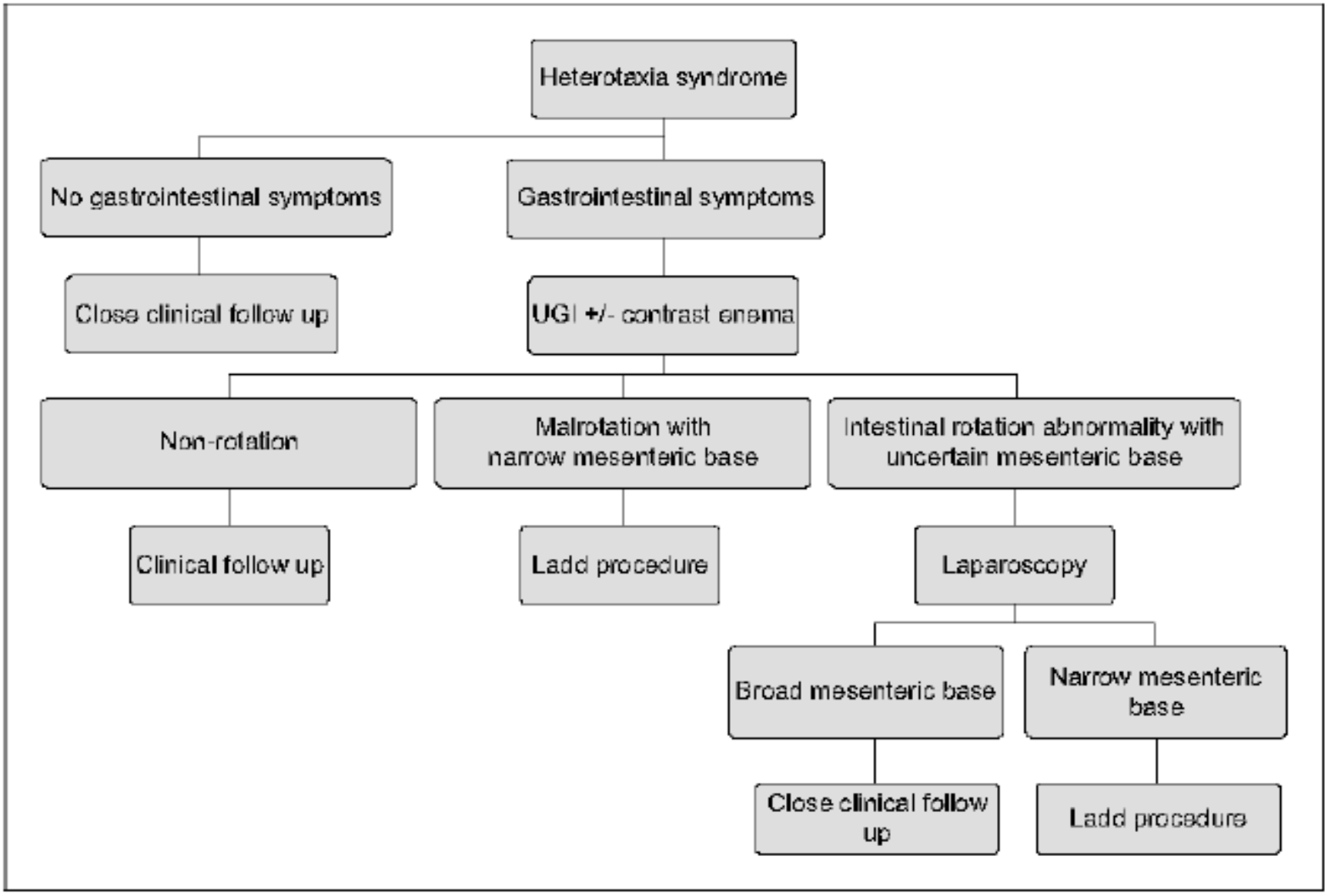
- 177 children with heterotaxia.
- 11 neonates had symptoms suggestive feeding intolerance, bilious vomiting, abdominal distention and
- 7 had malrotation on UGI.

Steven H. Borenstein and Jacob C. Langer Heterotaxia syndromes and their abdominal manifestations Current Opinion in Pediatrics 2006, 18:294–297

Borenstein & Langer

- 143 asymptomatic patients were followed closely
- 4 eventually developed gastrointestinal symptoms (bilious vomiting, abdominal pain or distention)
  - 1 child was found to have malrotation.
- The median follow-up for the remaining 139 patients was nine years
  - 60 (43%) died of cardiac disease and none developed symptoms of intestinal obstruction or ischemia.
- 3.4% of patients developed malrotation
  - all presented with symptoms that prompted urgent investigation, diagnosis and surgery.

Steven H. Borenstein and Jacob C. Langer Heterotaxia syndromes and their abdominal manifestations Current Opinion in Pediatrics 2006, 18:294–297



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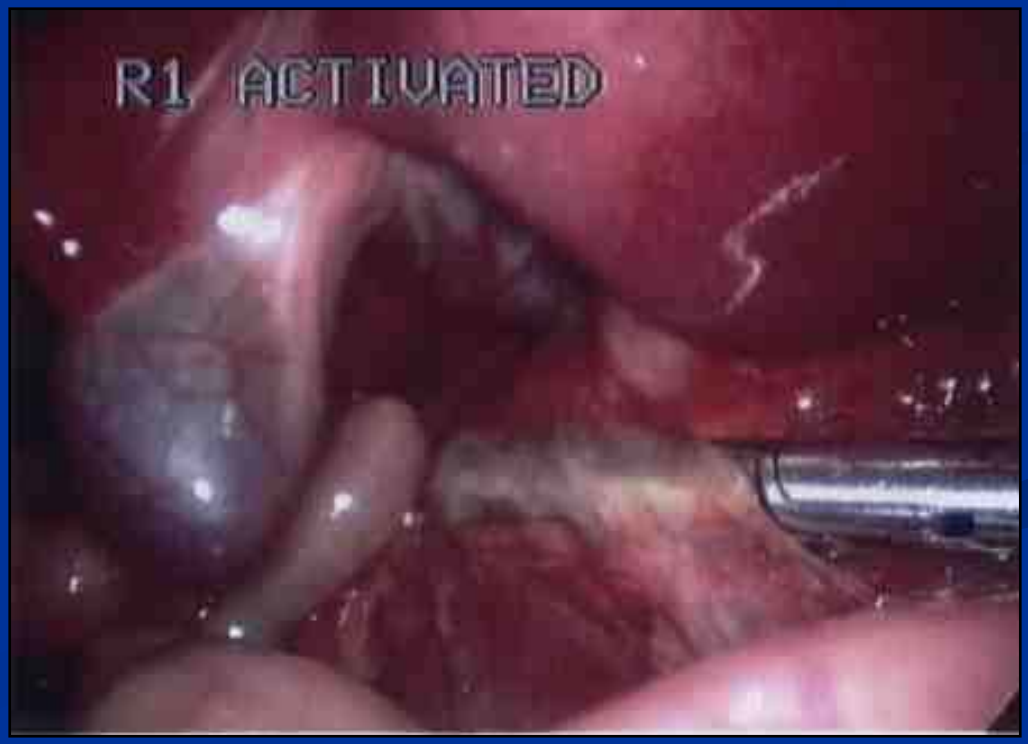
Langer (2006)

"When the risk of midgut volvulus in symptomatic patients is unclear from the radiological studies, laparoscopy or laparotomy should be done, with subsequent Ladd procedure for those in whom the mesenteric base is felt to be narrow."

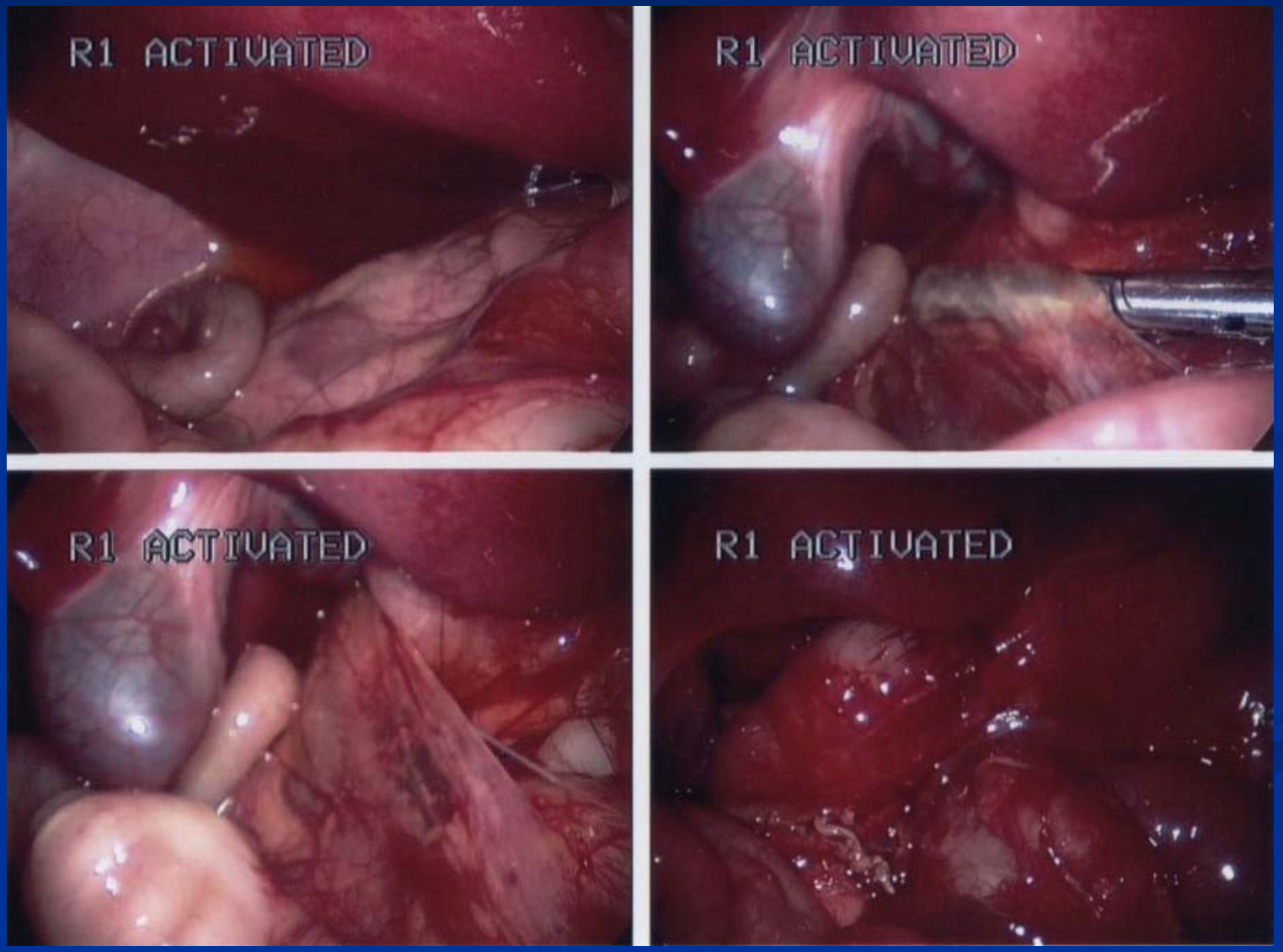
Laparoscopy

- Recent studies discuss the advantages of laparoscopic inspection for malrotation

- No long term data on laparoscopy in malrotation patients with CHD are available, yet.



Laparoscopic Malrotation



"Given the potential for a catastrophic abdominal event resulting from uncorrected intestinal malrotation in the setting of complex CHD and the overall improved survival of patients with complex CHD … an elective Ladd procedure for patients with heterotaxia and asymptomatic malrotation has an acceptably low morbidity."

Outcomes after a Ladd procedure for intestinal malrotation with heterotaxia. David B. Tashjiana, Bevin Weeksb, Martina Bruecknerb, Robert J. Touloukiana Journal of Pediatric Surgery (2007) 42, 528– 531

Recommendations

"Ladd procedure should be considered when symptoms of patients' heart disease are well controlled rather than delaying interventions until an emergent operation is required at a time when patients' hemodynamic status may be less favorable."

Outcomes after a Ladd procedure for intestinal malrotation with heterotaxia. David B. Tashjiana, Bevin Weeksb, Martina Brueckner b, Robert J. Touloukiana
Journal of Pediatric Surgery (2007) 42, 528– 531

“Atypical Malrotation”

- The authors reviewed retrospectively 176 patients undergoing operative correction of malrotation

- The ligament of Treitz (LOT) was classified as
  - Typical Malrotation if LOT was absent or right of midline.
  - High if left of midline and above the 12th thoracic vertebra,
  - Low if left of midline and below the 12th thoracic vertebra, and

Management of typical and atypical intestinal malrotation .
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Atypical malrotation

- Results: A total of 201 patients underwent operation for malrotation, in 176 there were adequate radiologic studies to allow classification of the LOT
- Conclusion: Atypical malrotation patients are at significantly lower risk of volvulus and internal hernia compared with typical malrotation patients, and operation appears to come with increased morbidity

Comment about "Atypical Malrotation"

- Many surgeons to do not consider patients with a duodenum to the left of the midline to have malrotation.

Conclusions: Heterotaxia

- "It is our opinion that providing there is close clinical follow-up, children with heterotaxia do not need routine gastrointestinal imaging to screen for intestinal rotational abnormalities."

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