Case #4

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07/23/2020

Children's Hospital Colorado

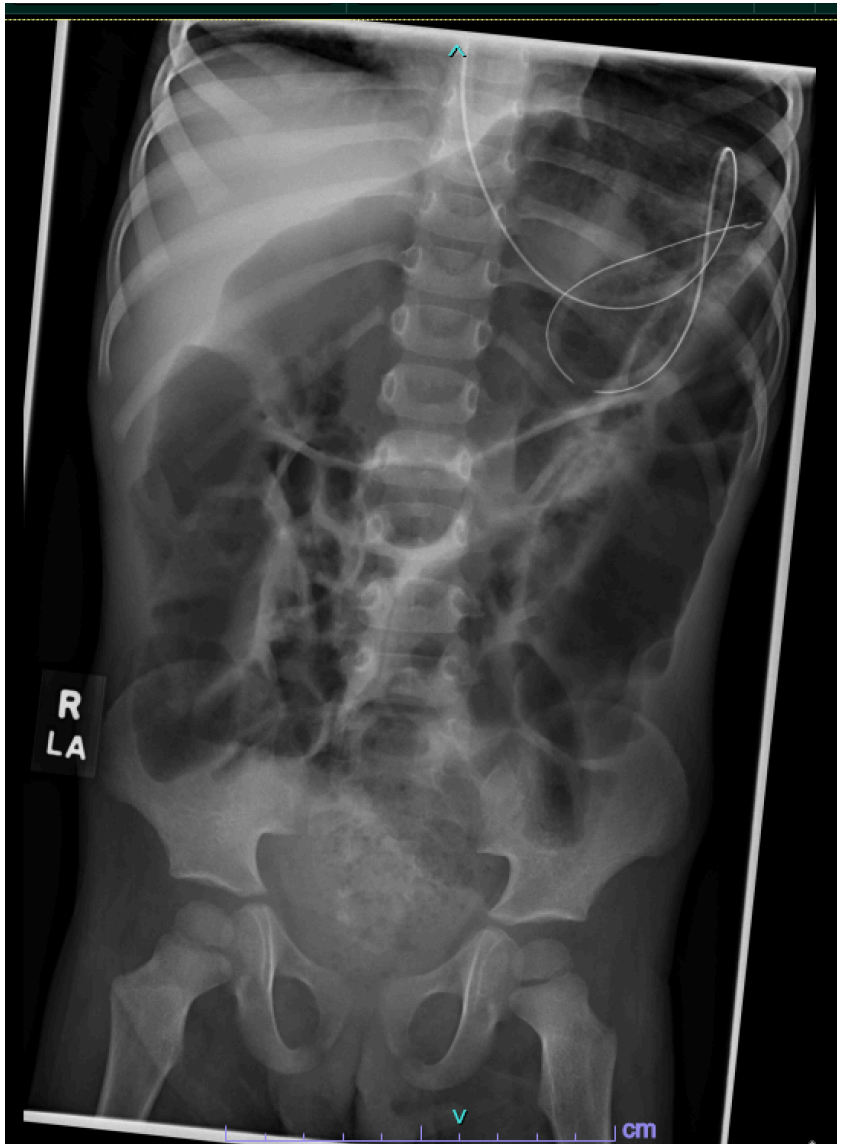
INTERNATIONAL CENTER FOR
COLORECTAL AND
UROGENITAL CARE

History from other Hospital

Male patient born in 2016
Hirschsprung
Enterostomy July 2016
Pull-through April 2017 (Per parents a Soave procedure)
Gastrostomy tube June 2017
Adoption May 2019 (from Europe)

Emergency Department
2019

Abdominal distention
Nausea
Fever



1. Based on this story, what would be the most likely diagnosis?

A. Colitis

B. Residual aganglionosis

C. Transitional pull-through

D. Torsion of the pull-through

E. Stenosis of the anastomosis

2. What will be your first therapeutic approach?

A. Contrast enema

B. Rectal biopsy

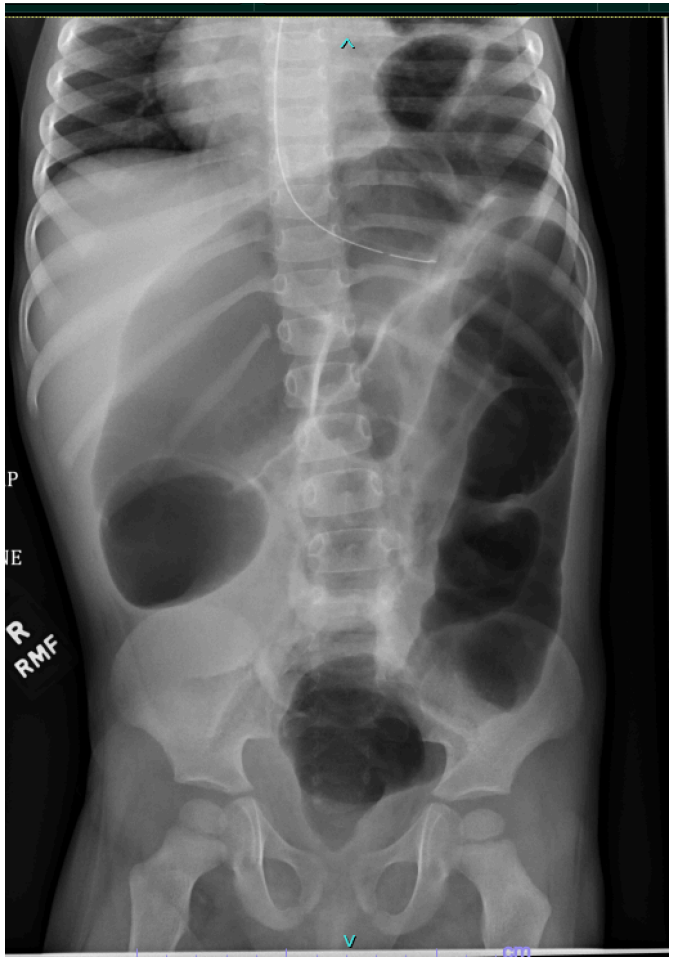
C. Metronidazol and rectal irrigations

D. Nasogastric tube

E. Colostomy

October 2019

- Patient improved partially
- Daily irrigations every 8 hours
- Flagyl 10 mg every 8 hours
- Abdomen less distended but not a big difference



3. What will be your next diagnostic evaluation?

A. Contrast enema

B. Rectal biopsy

C. Anorectal manometry

D. Exam of the anal canal under anesthesia

E. Biopsy from the pull-through

August 2019

1. Normal anal canal
2. Line of the colorectal anastomosis at 1 cm from the anal canal
3. Redundant bowel on the pelvis, more on the left side.
4. Possible partial obstruction 15 cm proximal from the anus
5. Rectal biopsy of the neo-rectum

DIAGNOSIS:
RECTUM, FULL THICKNESS BIOPSY:

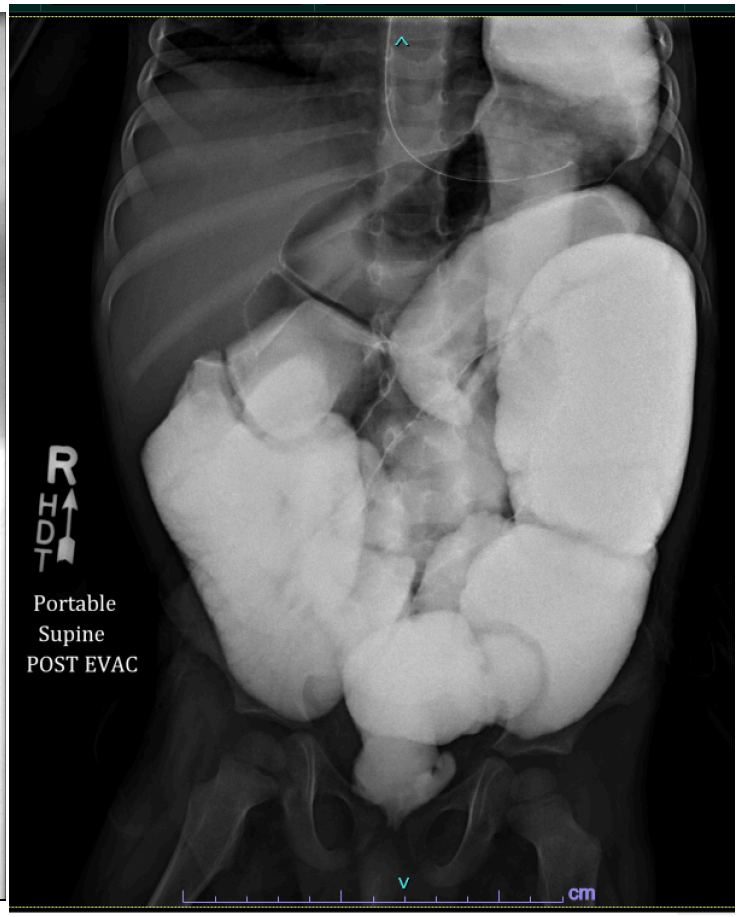
- COLITIS WITH ABSCESS FORMATION.

- ABSENT GANGLION CELLS CONFIRMED BY CALRETININ.

- SUBMUCOSAL FIBROSIS WITH PROLIFERATION AND DISORGANIZATION OF THE SMOOTH MUSCLE LAYERS.

- HYPERTROPHIC NERVE FIBERS.


* cm


R
HDT
Portable
Supine
POST EVAC
cm


Upright
jls
* cm

4. Based on these studies, what is your diagnosis?

A. Stenosis

B. Obstruction due to the rectal cuff

C. Transition zone PT

D. Aganglionic PT

E. Dysmotility of the remaining colon

5. What will be your treatment plan?

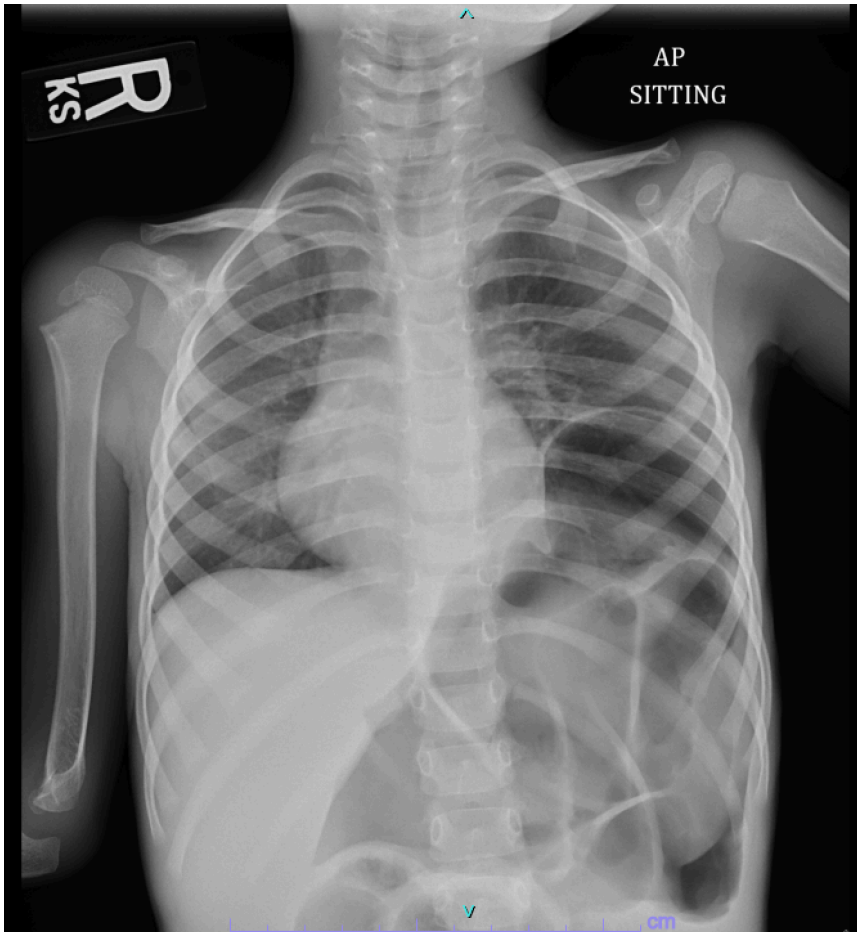
A. Colostomy

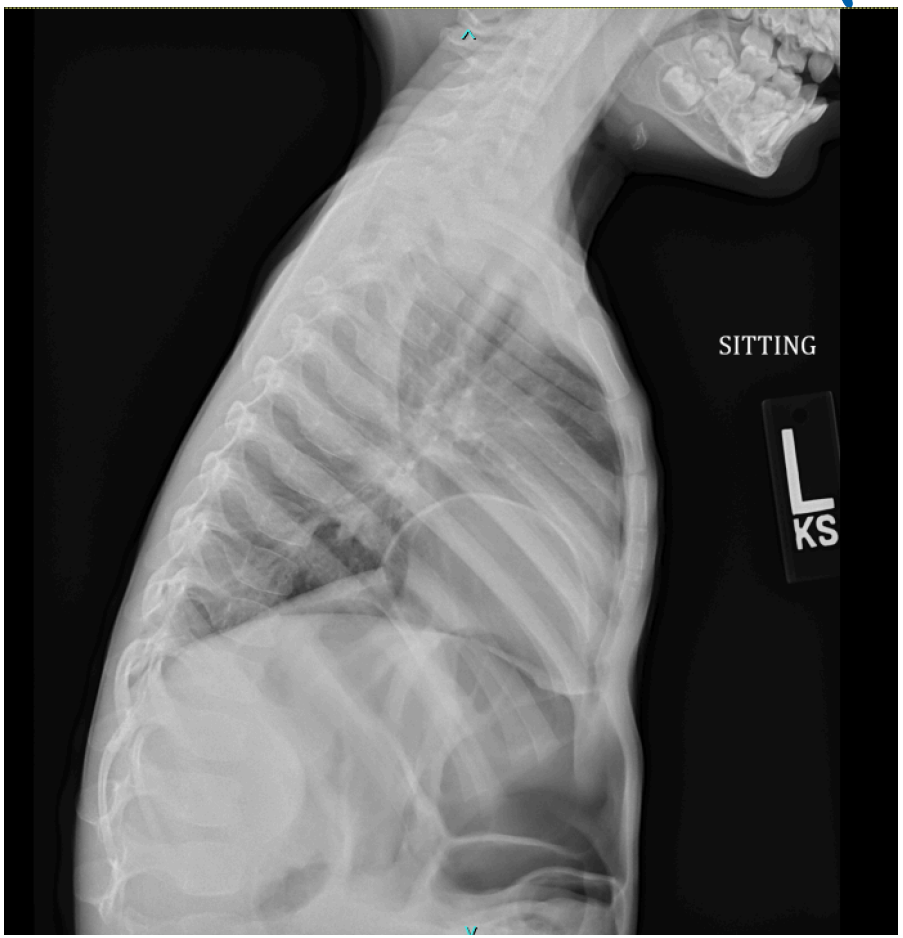
B. Resection of the muscular cuff

C. Laparotomy with new pull-through and repair of the diaphragm hernia

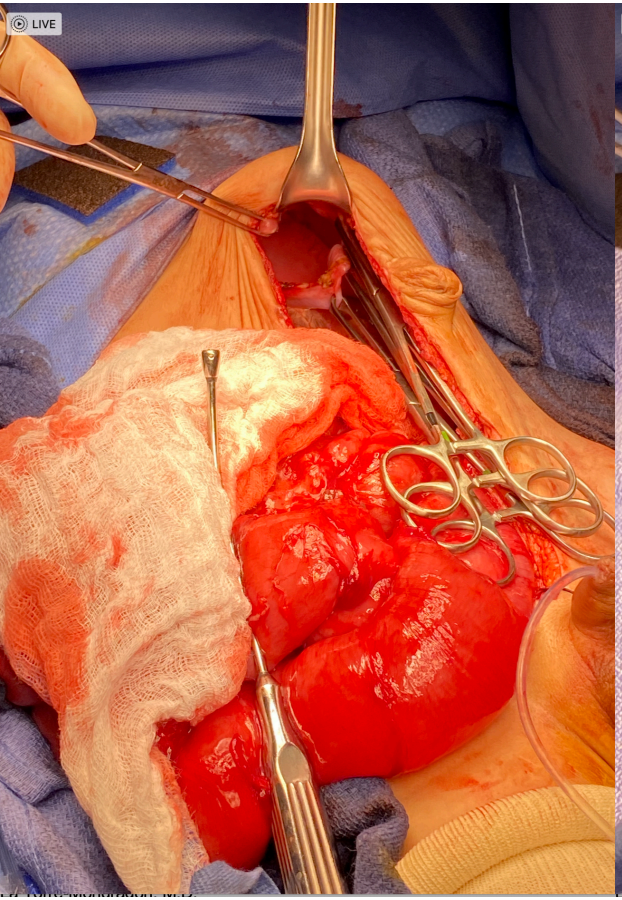
D. Ileostomy and biopsies of the remaining colon

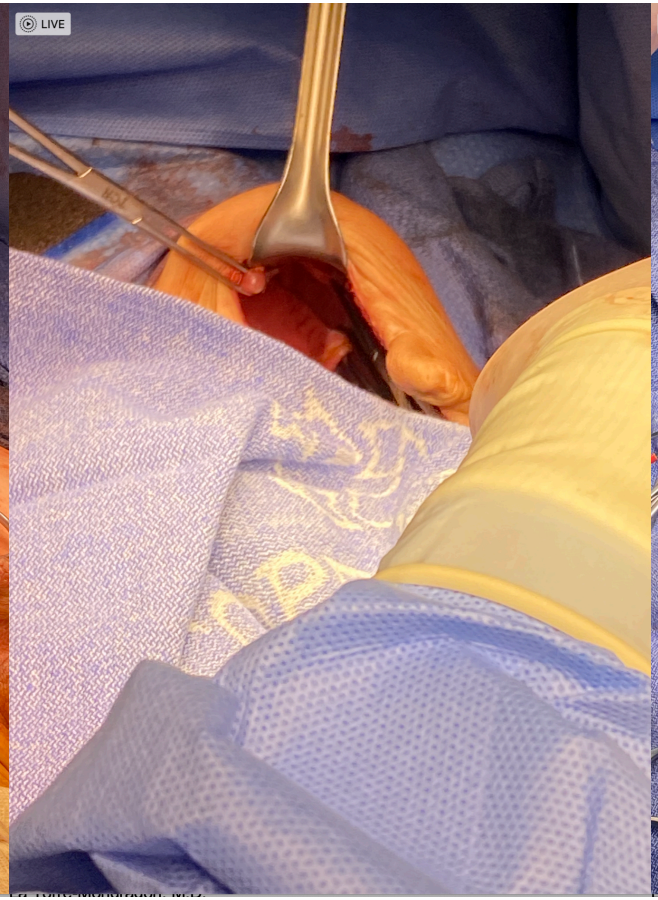
E. Transanal Swenson pull-through

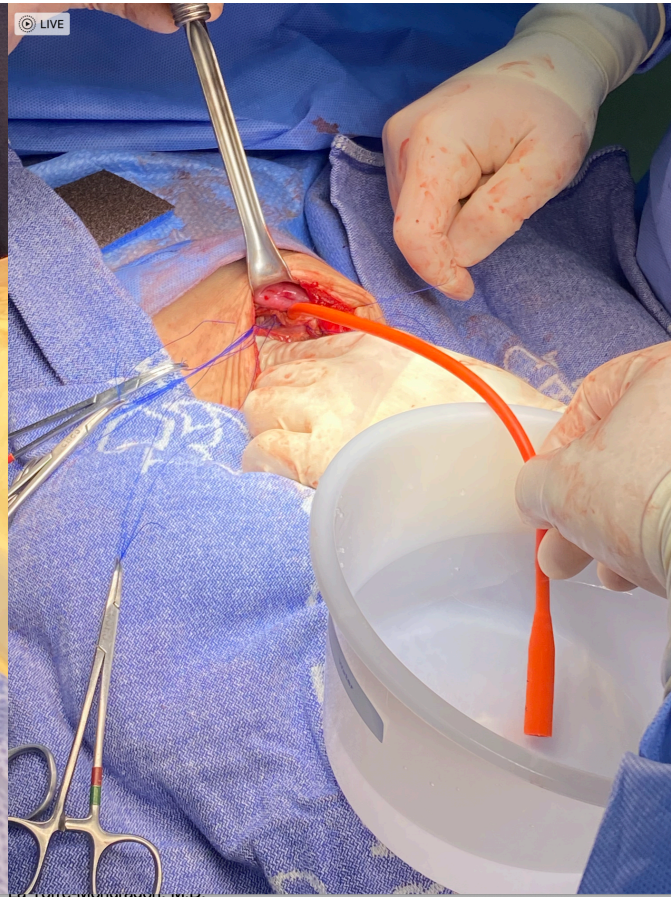

R
KS
AP
SITTING


SITTING
L
KS

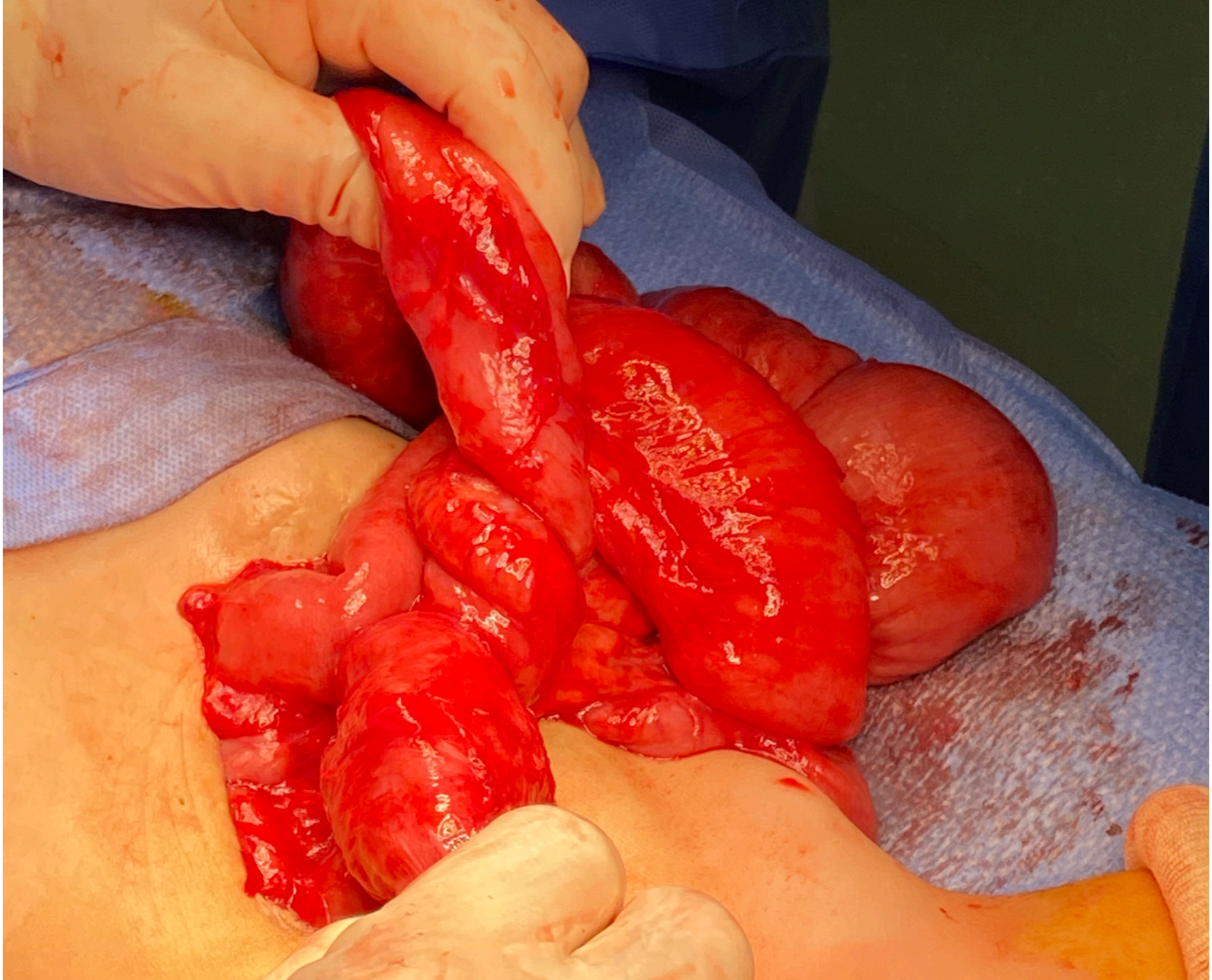
November -2019

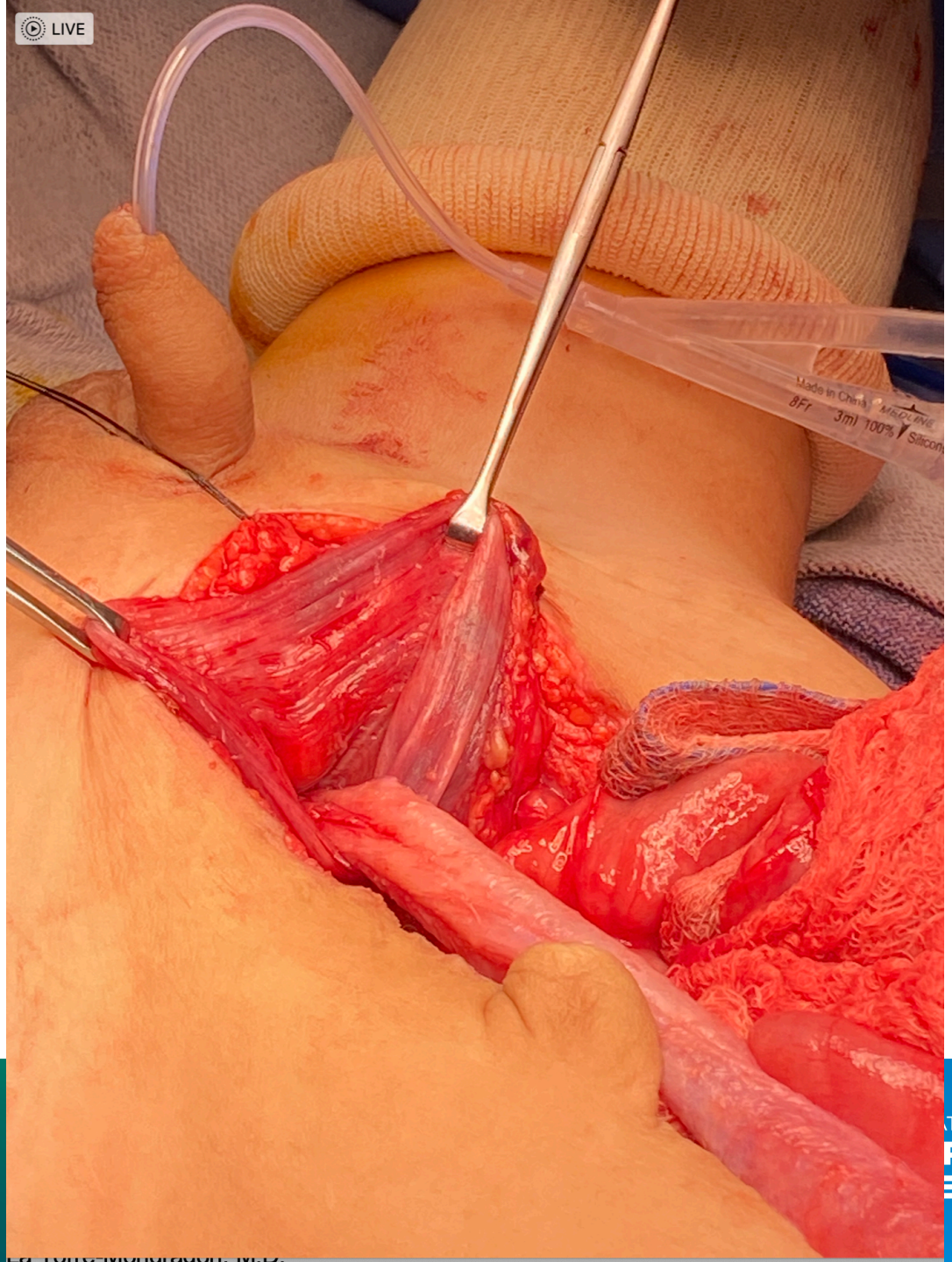






The terminal ileum was entering in the pelvis.
The patient did not have a colon.
Gigantic dilation of the small bowel.
Mesenteric defect
Pull-through done with a small bowel.
Pull-through was twisted 360 grades.





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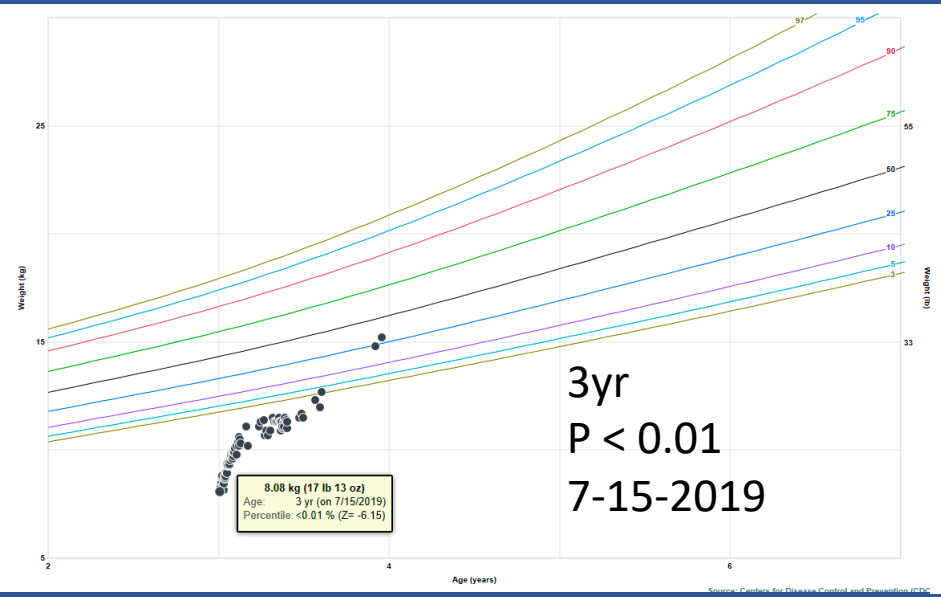
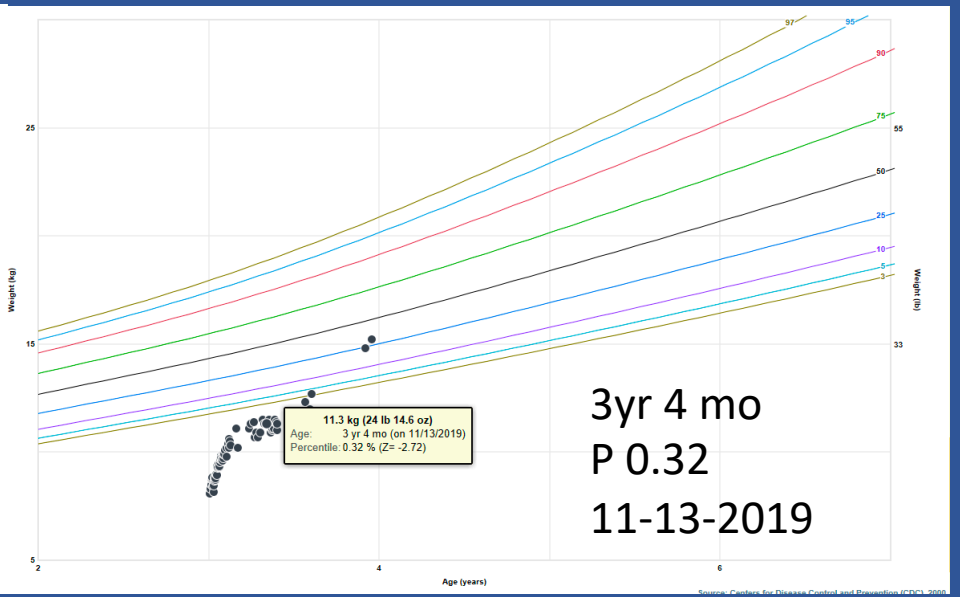
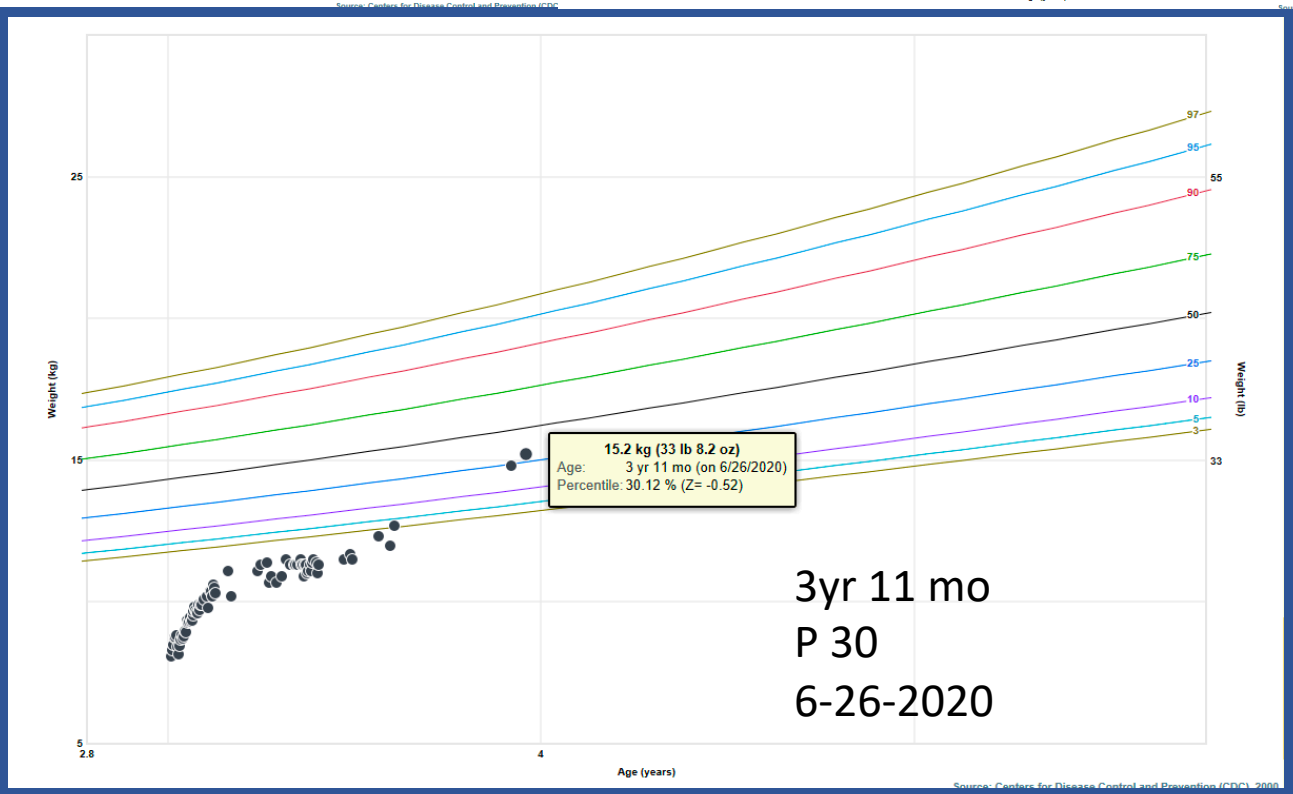


3yr
P < 0.01
7-15-2019



3yr 4 mo
P 0.32
11-13-2019



3yr 11 mo
P 30
6-26-2020

6. Is there any association between Hirschsprung and diaphragmatic hernia?

A. Yes

B. No

C. Never hear about it

D. I do not know

Is there any association between Hirschsprung and diaphragmatic hernia?

Fryns syndrome: a surviving case with associated Hirschsprung's disease and hemidiaphragmatic agenesis. J Paediatr Child Health. 2002 Jun;38(3):318-20.

Fryns syndrome survivors and neurologic outcome. m J Med Genet. 1995 Nov 20;59(3):334-40.

PUBMED 161 papers

Abnormality of connective tissue.

Diaphragmatic Hernia

Abnormality of head or neck
Abnormality of limbs
Abnormality of metabolism/homeostasis
Abnormality of prenatal development or birth
Abnormality of the cardiovascular system
Abnormality of the digestive system

Hirschsprung disease

Abnormality of the eye
Abnormality of the genitourinary system
Abnormality of the immune system
Abnormality of the integument
Abnormality of the musculature
Abnormality of the nervous system
Abnormality of the respiratory system
Abnormality of the skeletal system
Ear malformation
Growth abnormality

Fryns syndrome
Autosomal recessive inheritance
https://omim.org/entry/229850

7. With this evolution after the re-operation.... What would be the next therapeutic plan?

A. Ileostomy closure when urinary control is achieved

B. Permanent ileostomy

C. Try bowel management via ileostomy for fecal incontinence

D. I do not know