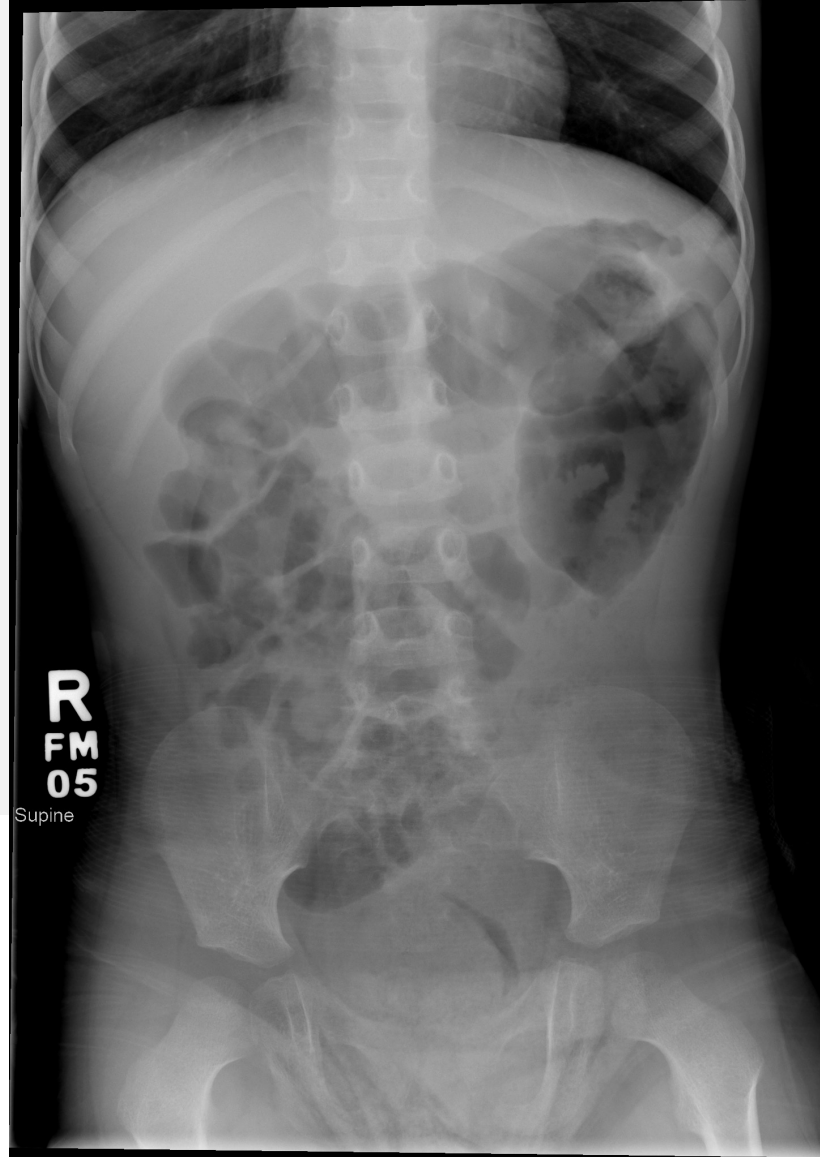


Contrast enema and biopsies post Hirschsprung pull through

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Male, age 3 years:
intermittent
abdominal
distension, stooling
problems, Hx of
sigmoid HD

Patient H,
age 3 years
Mowat
Wilson
Syndrome

Born January 2019 37 weeks, 3.4kg

Prenatal: cystic hygroma

Failure to pass meconium, HD on suction rectal biopsy DOL 4

Other: hypotonia, penile hypospadias, left hydronephrosis

Sent home on rectal irrigations

- *Mowat-Wilson syndrome – This is caused by haploinsufficiency of the [ZEB2](#) gene. Approximately 50 percent of individuals with Mowat-Wilson syndrome have HD; other features are distinctive facial characteristics, moderate to severe intellectual disability, genitourinary anomalies, and heart defects.*

- **Picture from Web: not our patient**



Surgical procedures

March 2019: Lap pull thru age 9 weeks, transition in sigmoid

Pathology: good ganglion cells no nerve hypertrophy, anastomosis 3.3-4.7 cm above the aganglionosis

Ileostomy and washout for anastomotic leak on Day 6

June 2019: Closure of ileostomy

Revision / repair of leaking ileal anastomosis on day 5

July 2022

- "H was doing fine but for past month or two he seems to just pass small amounts gas and stool all the time. His colon is distended and she can see it sticking out. Irrigations every other night. He was not like this before."
- Booked for contrast enema, botox injection, rebiopsy, also trial of oral Metronidazole.

Exam
under
anesthesia
Fall 2022

Abdomen softly distended, anus
normal appearance

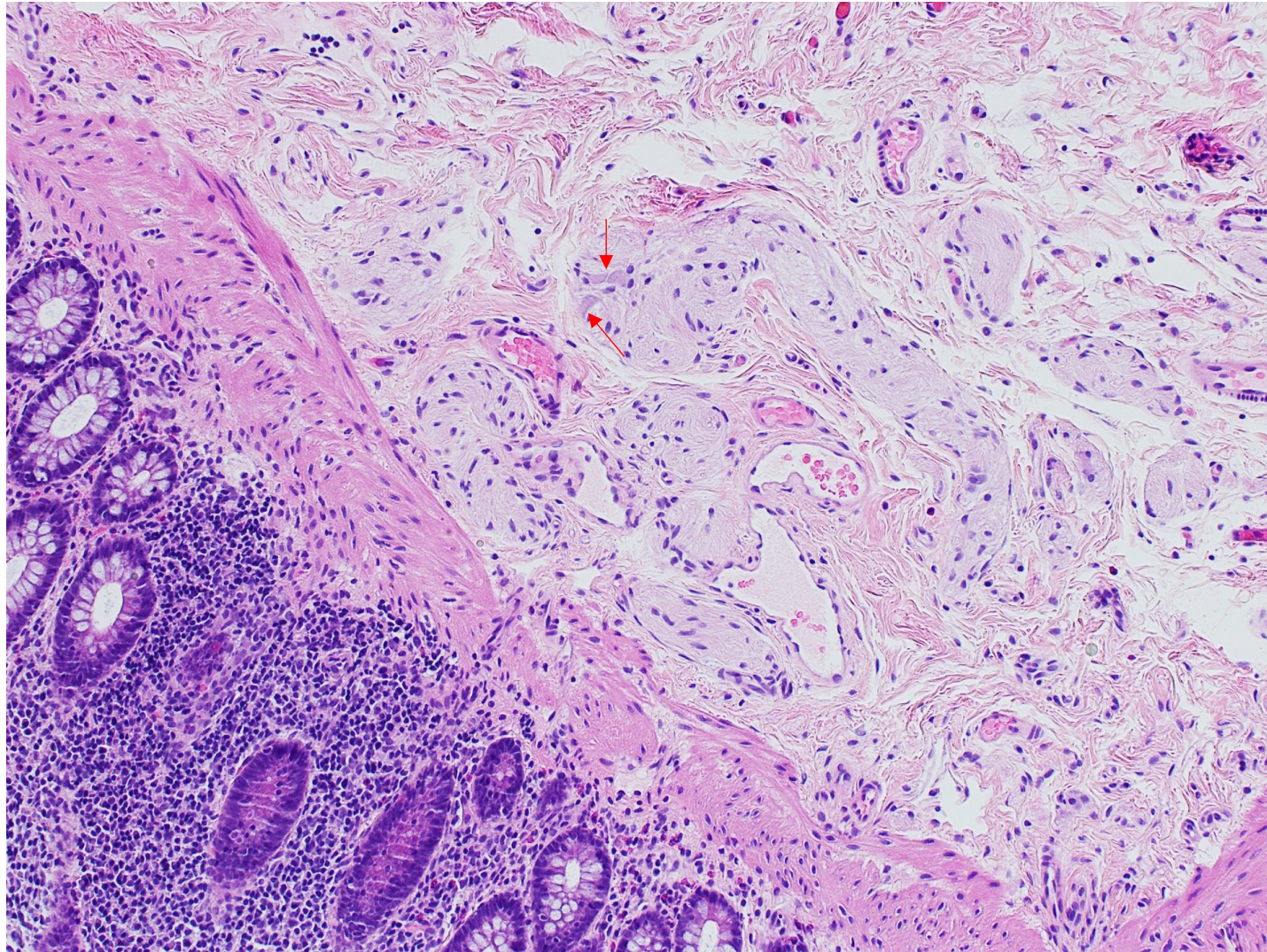
Rectal exam anal tone OK, liquid stool
in “rectum”. Sigmoidoscopy to 20 cm
no obstruction noted, no colitis seen.

50 units Botulinum toxin

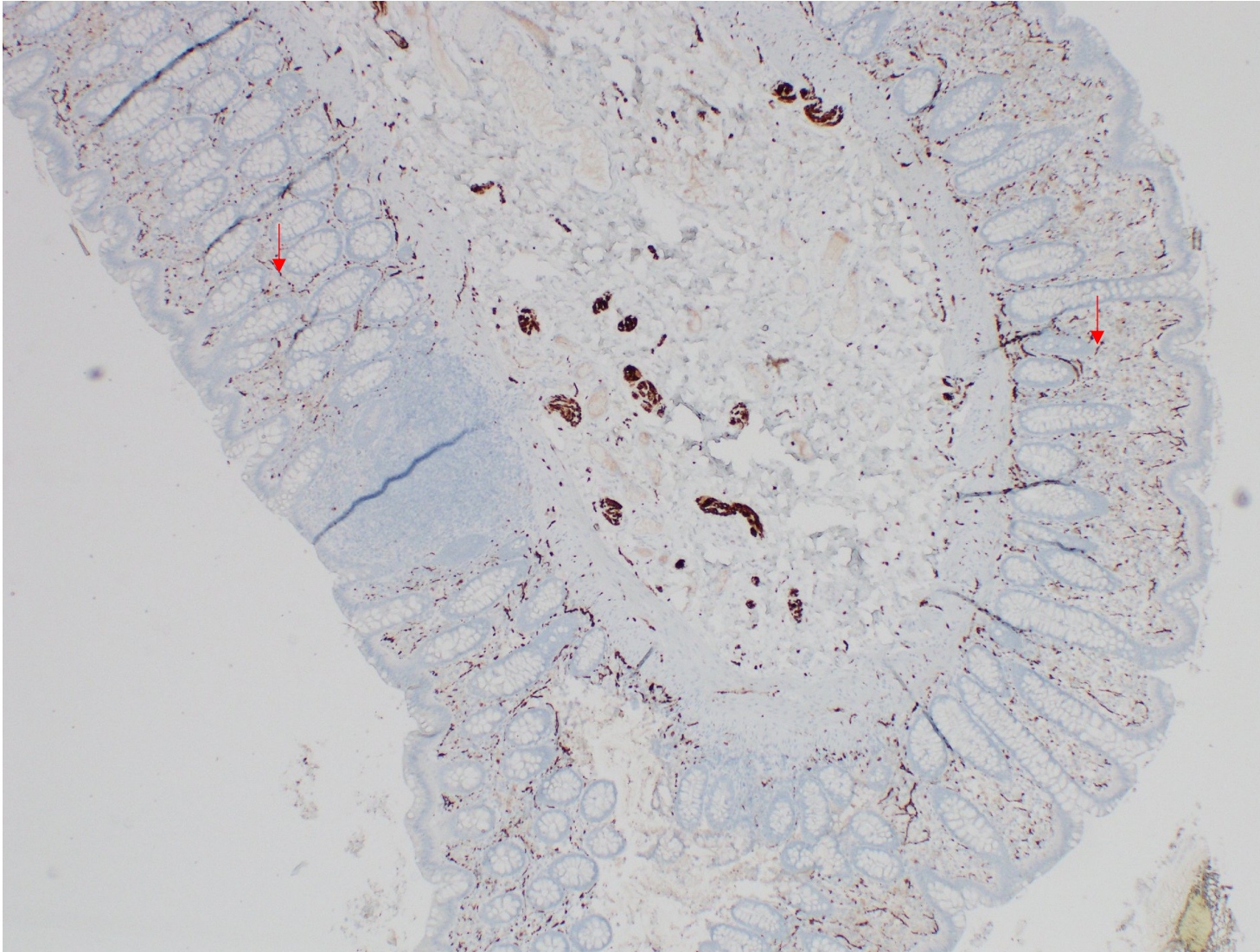


Suction
Biopsy Aug
2022 at 4cm
from anal
verge

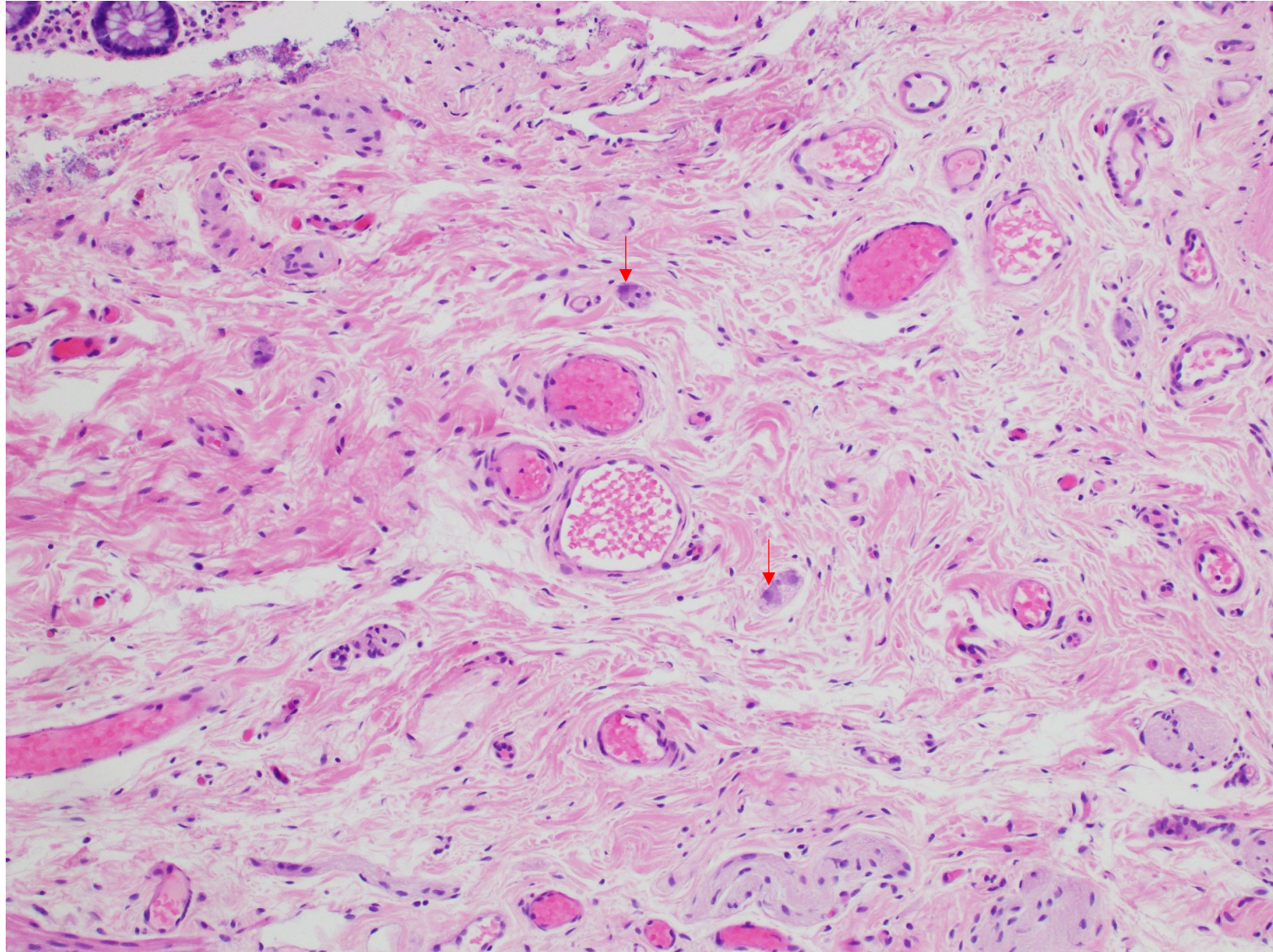
- Abundant ganglion cells
- Calretinin positive
- Occasional nerve trunks 50 microns
- Original pathology was good at anastomosis (reviewed)
- Original anastomosis was 3.3-4.7 cm above aganglionic



Submucosal nerve hypertrophy, some with ganglion cells (arrows)

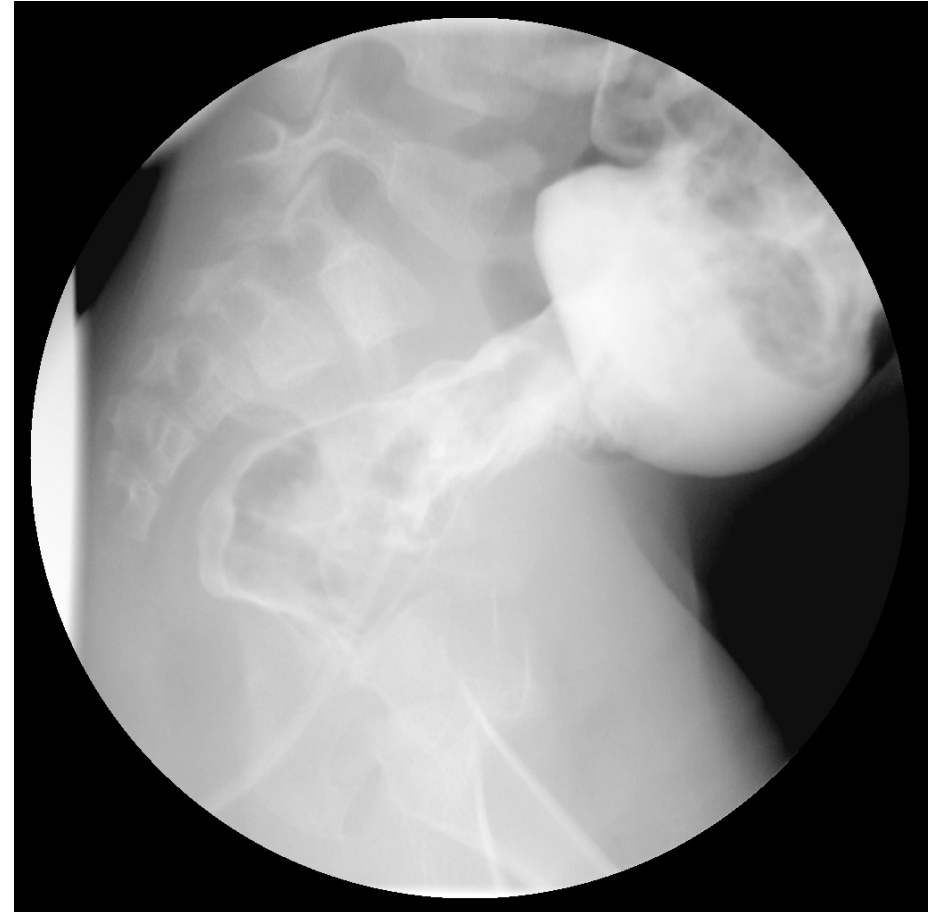


Intact mucosal innervation on calretinin immunostain (arrows)

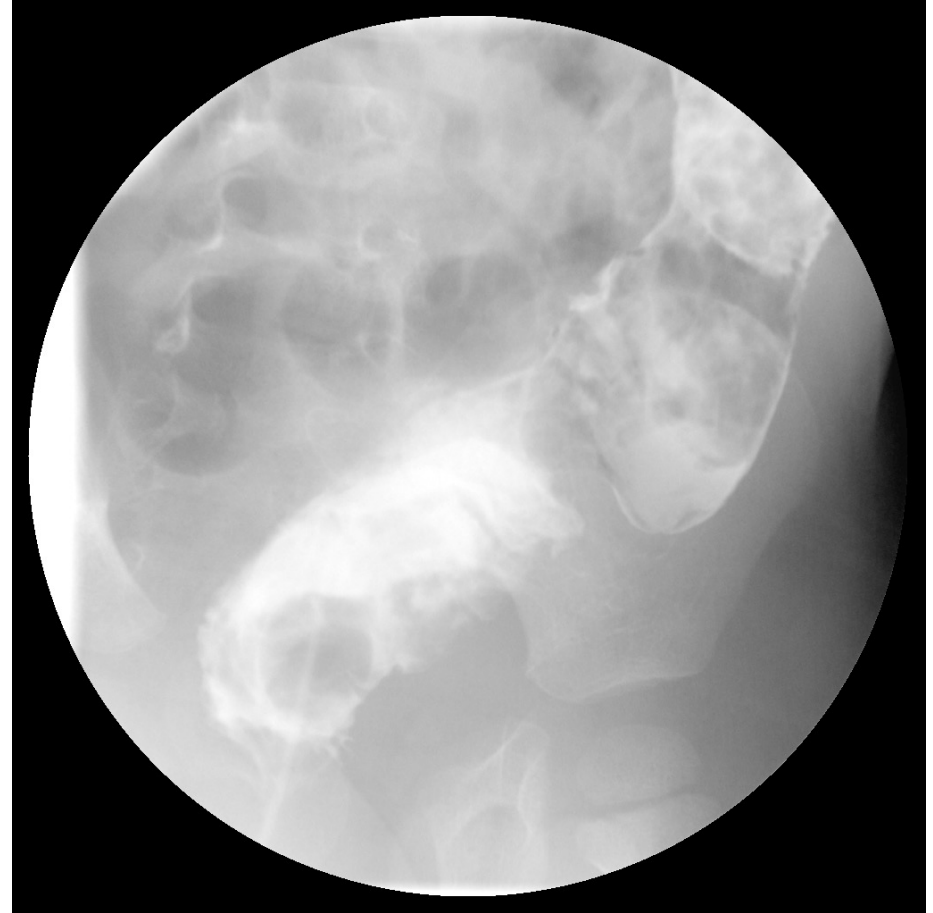


Submucosal ganglion cells that are not associated with nerve hypertrophy (arrows)

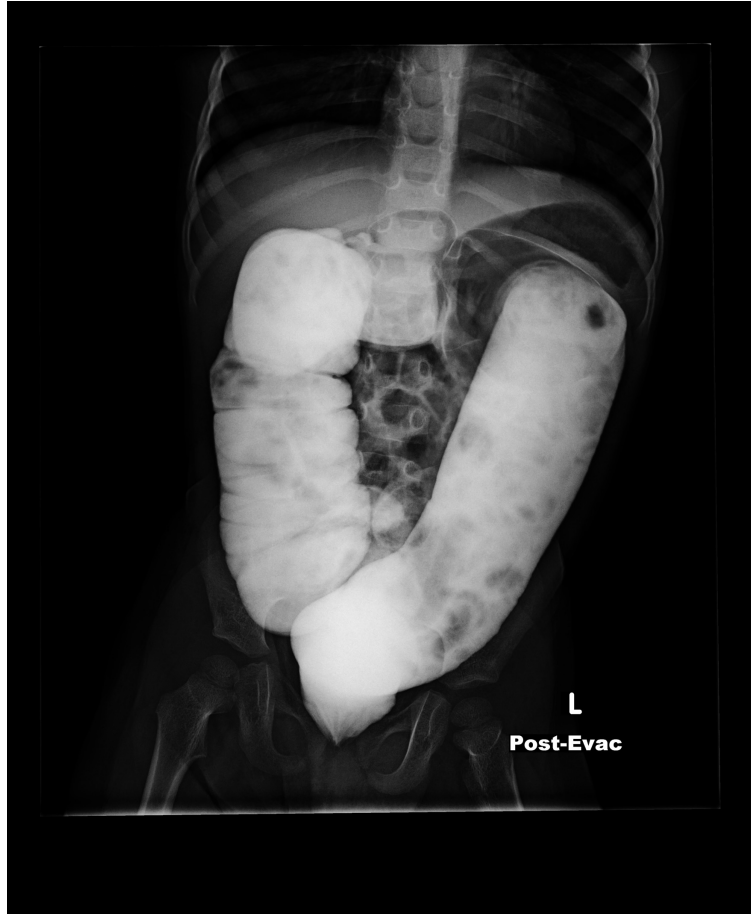
Contrast enema Oct 2022



Contrast enema Oct 2022



Post evacuation images



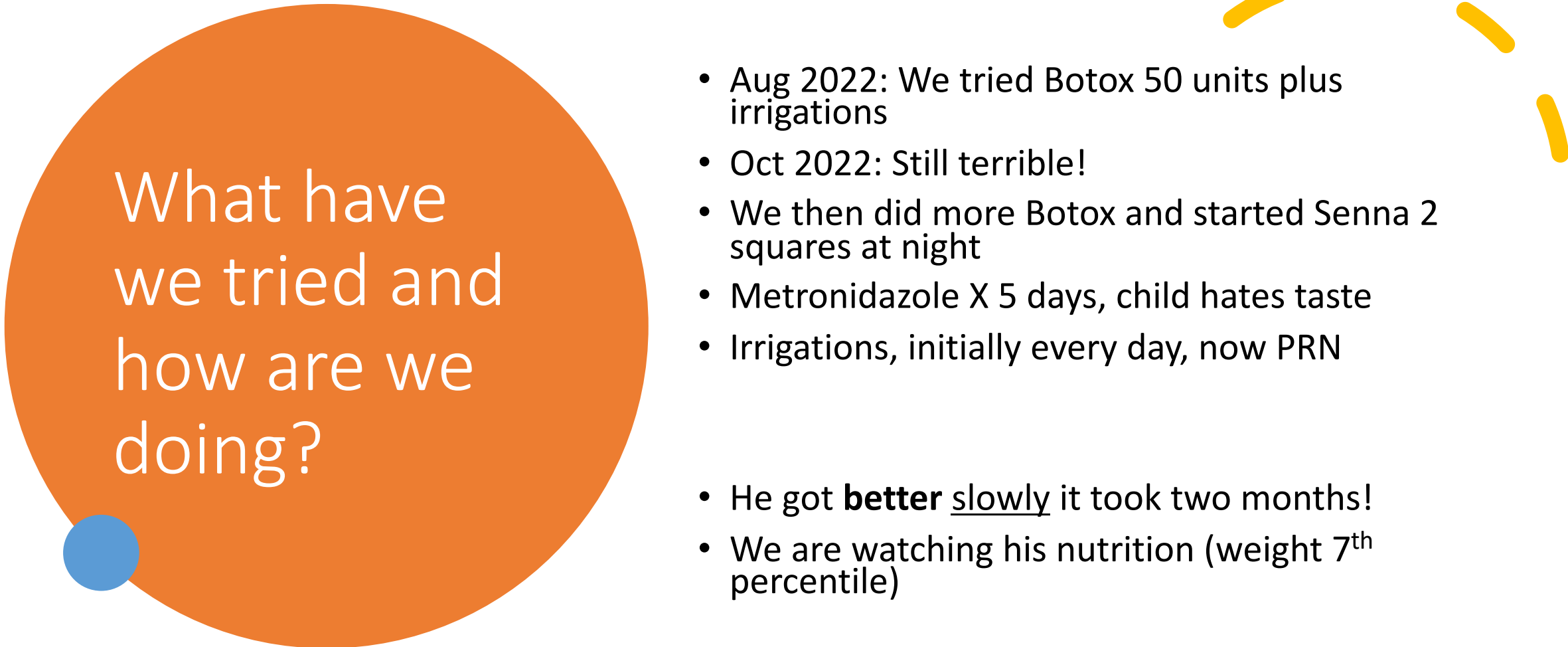
Summary

Age 3 years, Mowat Wilson

Obstructive symptoms post pull thru

Biopsy: ganglion cells but a few hypertrophied nerves

Contrast possible narrowing at upper rectum, but scope goes through.



What have
we tried and
how are we
doing?

- Aug 2022: We tried Botox 50 units plus irrigations
- Oct 2022: Still terrible!
- We then did more Botox and started Senna 2 squares at night
- Metronidazole X 5 days, child hates taste
- Irrigations, initially every day, now PRN

- He got **better** slowly it took two months!
- We are watching his nutrition (weight 7th percentile)



Questions for team

- What do you think of the contrast enema?
- Do you think the biopsy result is significant?
- Do you think we will win?
- When would you give up and do ileostomy?
- What about his syndrome?