CHALLENGING CASES

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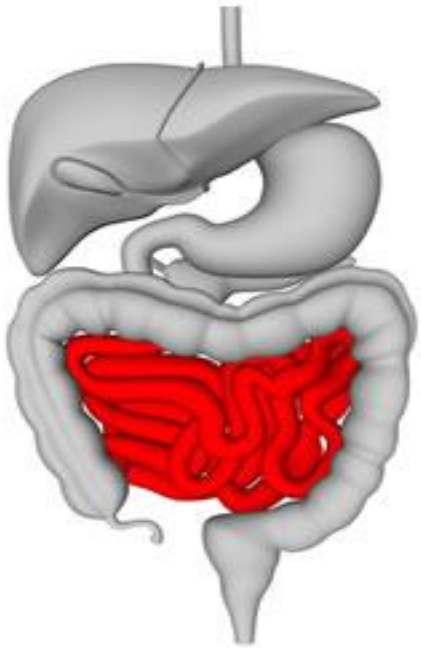
The Gastro Foundation of South Africa: Paediatric Gastroenterology and IBD Interest Group

27th August 2016



PATIENT ADC

10 year old girl

History

- Presented in Dec 2015 with severe persistent iron deficiency anemia

ORDERED: FBC-H, Iron Profile-B
SAMPLE: E01 EDTA Haematology, S01 SST

Test	ABN	Result	Reference	Unit
FULL BLOOD COUNT				
=> Haemoglobin	#*L Delta: 9.8 on 02/04/15-1340	5.2	11.5-15.5	g/dl
=> Red Cell Count	L	2.96	4.00-5.20	10^12/l
=> Haematocrit	#*L Delta: 34.4 on 02/04/15-1340	18.9	35.0-45.0	%
=> MCV	*L	63.9	77.0-95.0	fl
=> MCH	*L	17.6	25.0-33.0	pg
=> MCHC	L	27.5	31.0-36.0	g/dl
=> RDW	H	20.2	10.0-14.0	%
=> White Cell Count		5.11	5.00-13.00	10^9/l
=> Neutrophils		56.1		%
=> Neutrophils Abs		2.87	2.00-8.00	10^9/l
=> Lymphocytes		33.9		%
=> Lymphocytes Abs		1.73	1.00-5.00	10^9/l
=> Monocytes		7.4		%
=> Monocytes Abs		0.38	0.20-1.00	10^9/l
=> Eosinophils		0.8		%
=> Eosinophils Abs		0.04	0.00-1.00	10^9/l
=> Basophils		1.8		%
=> Basophils Abs		0.09	0.00-0.10	10^9/l
=> Platelet Count		248	178-400	10^9/l
HAEMATINICS				
=> Ferritin	# L Delta: 6 on 02/04/15-1340	2	7-140	ng/ml

- First seen by paediatrician in Jan 2015 with symptoms of tiredness
- Noted to have iron deficiency anemia based on results of FBC and iron studies
- Commenced on oral iron therapy x 3 months
- Hb increased from 4,3 to 9,8 in April 2015; Ferritin increased from 1 to 6 in April
- Iron therapy was discontinued

FBC progression Jan 2015 – Dec 2015

	01/2015	02/2015	04/2015	07/2015	10/2015	12/2015
Hb	4.3 (11.5-15.5)	7.7	9.8	6.2	7.4	5.2
Hct	15 (35.0-45.0)	28.2	34.4	22	25	18.9
MCV	64 (77.0-95.0)	71.8	77.1	62	71	63.9
MCH	18 (25.0-33.0)	19.6	22.0	17	22	17.6
Plts	277 (178-400)	370	349	284	292	248
S-Fe	<2 (5.7-18.6)	3.8	11	<2	<2	6.1
S-Transferrin	3.3 (1.6-3.3)	3.1	3	3.6	3.3	2.4
% Sat	2 (2-40)	5	15	2	2	10
Ferritin	1 (7-140)	6	6	1	2	2

Symptoms

- Only symptom reported was tiredness
- No pain, fever
- Stools: normal, no rectal bleeding/dark stools noted
- Premenarchal
- No loss of weight, night sweats or coughing
- No skin rashes, joint pains

Past medical history

- Sept 2014: presented with a 1w history of RIF pain after recent treatment for a UTI. Examination revealed RIF tenderness; infective markers were raised; ultrasound suspicious of appendicitis. At surgery a normal appendix was encountered; small bowel intussusception was found. Laparoscopic reduction failed requiring mid-line laparotomy and a small bowel resection of 15-20cm. Recovery uncomplicated.
- Histology: Intussusception with small bowel hyperplasia

Medications

- Dewormed + course of oral metronidasole (?paracytic infestation) in July 2015, No NSAIDs

Family history

- negative

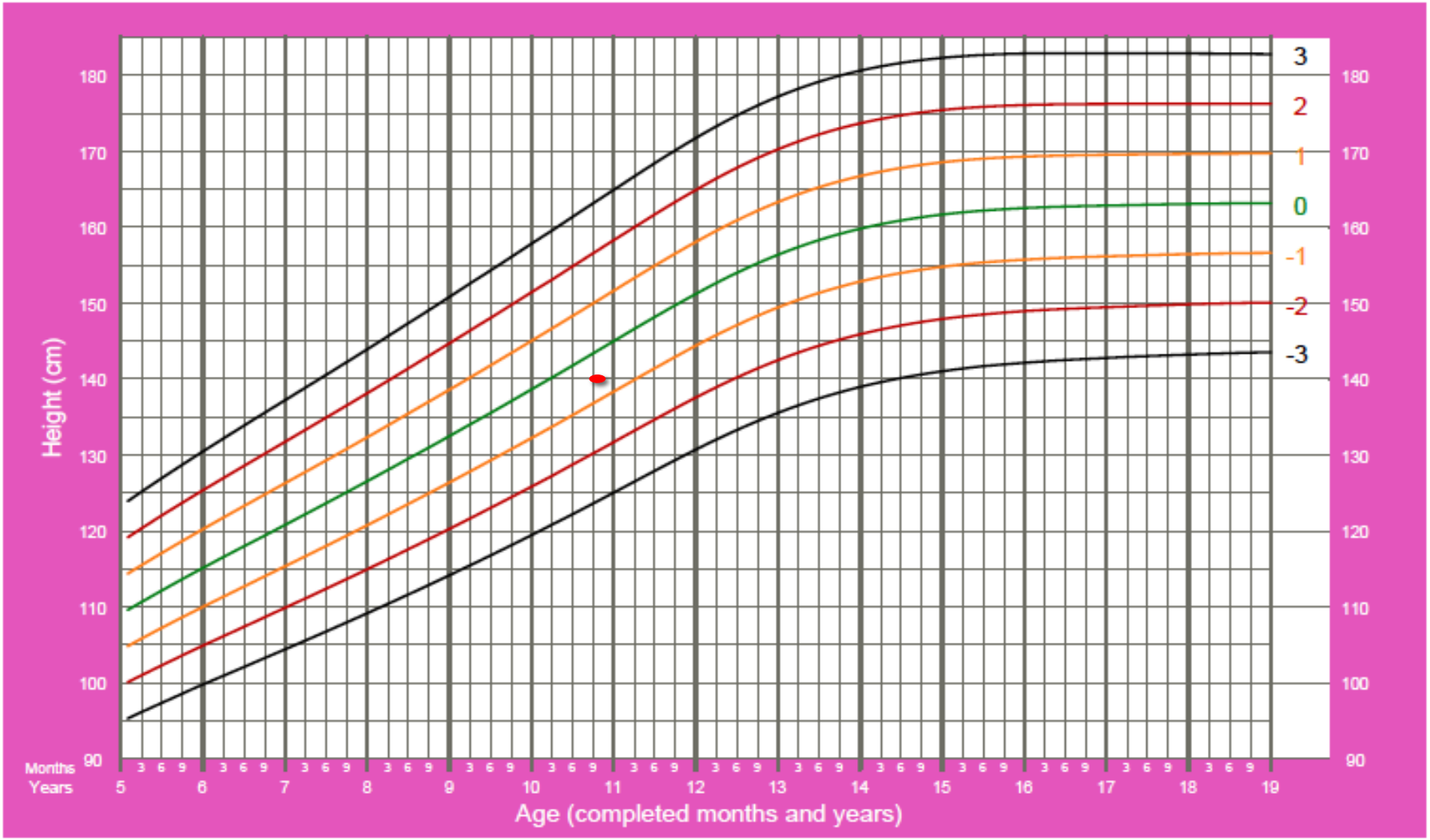
Examination

- Not acute / chronically ill
- Vitals normal incl, BP 100/50
- Pallor
- CVS: soft 2-3/6 PSM heard widely
- ABD: soft, non tender; no HSM or other masses felt
- No rashes / bruising
- Rest systemic exam NAD

Height-for-age GIRLS

5 to 19 years (z-scores)

World Health Organization

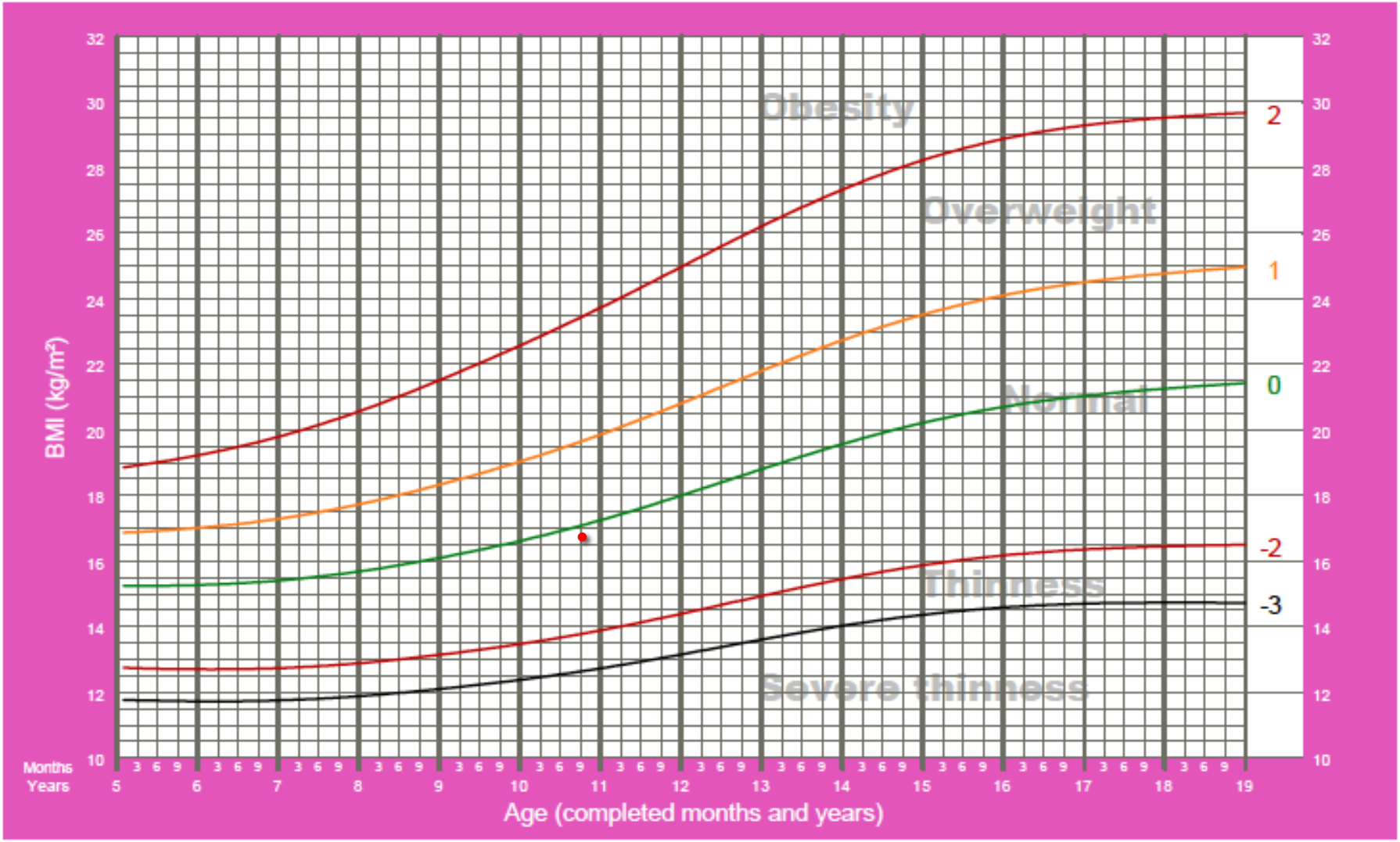


2007 WHO Reference

BMI-for-age GIRLS

5 to 19 years (z-scores)

World Health Organization



2007 WHO Reference

Investigations

- AXR: normal, incidental finding spina bifida oculta
- CXR: normal
- Abdominal sonar: normal

- Stool occult blood: negative in Jan 2015; positive on repeat in Dec 2015
- Stool MCS: negative
- Urine: NAD
-
- Retics 0,91% (abs 26,0)
- LDH: normal
- Smear: Hypochromic microcytic anemia; rouleaux formation; elliptocytes and target cells
- Renal function: normal
- LFTs: normal

- Celiac screen negative
- Faecal calprotectin: 307
- ESR: 10
- TB work-up: negative

Gastroscopy / Colonoscopy

- Blood transfusion prior endoscopy

- Gastroscopy:

Esophagus normal

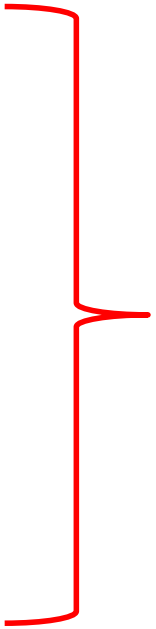
Stomach normal

Pylorus normal

D1-D4 normal

- Colonoscopy:

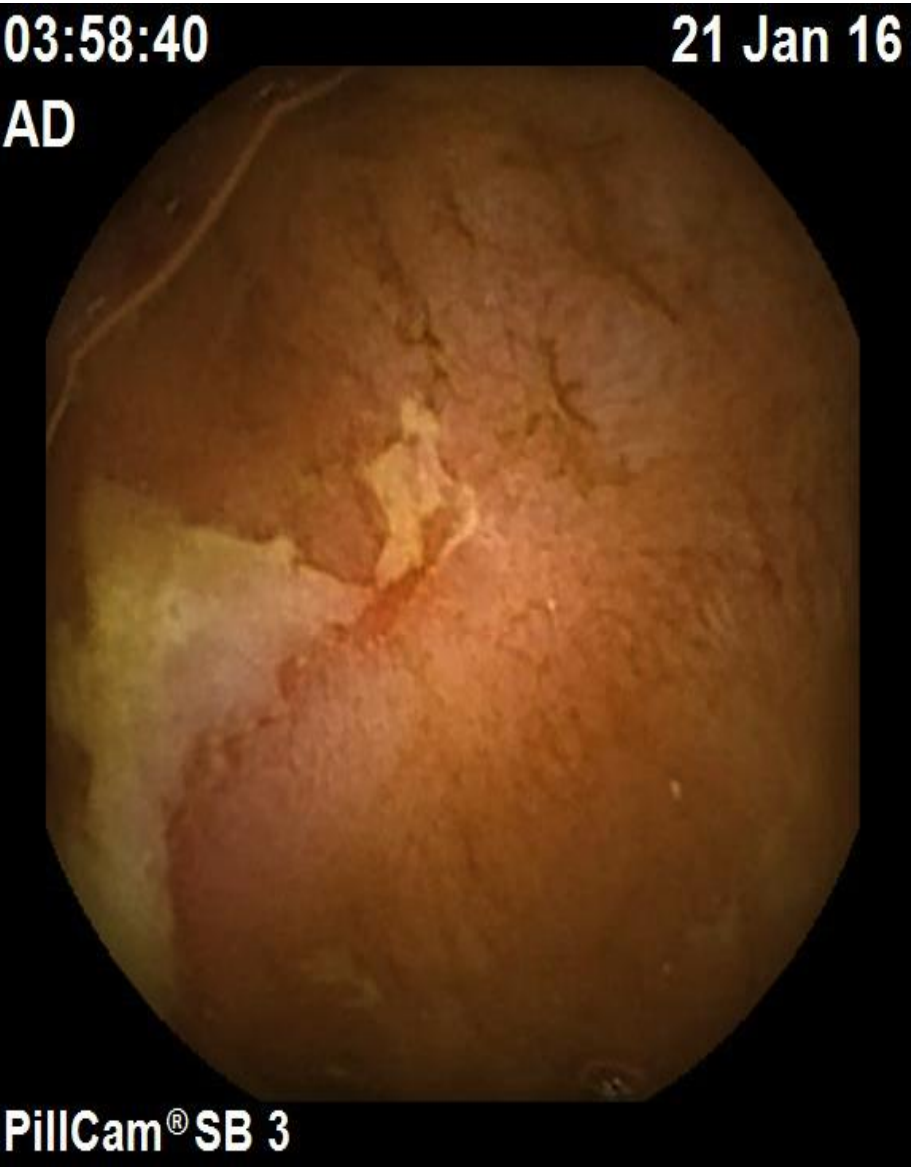
rectum to TI normal

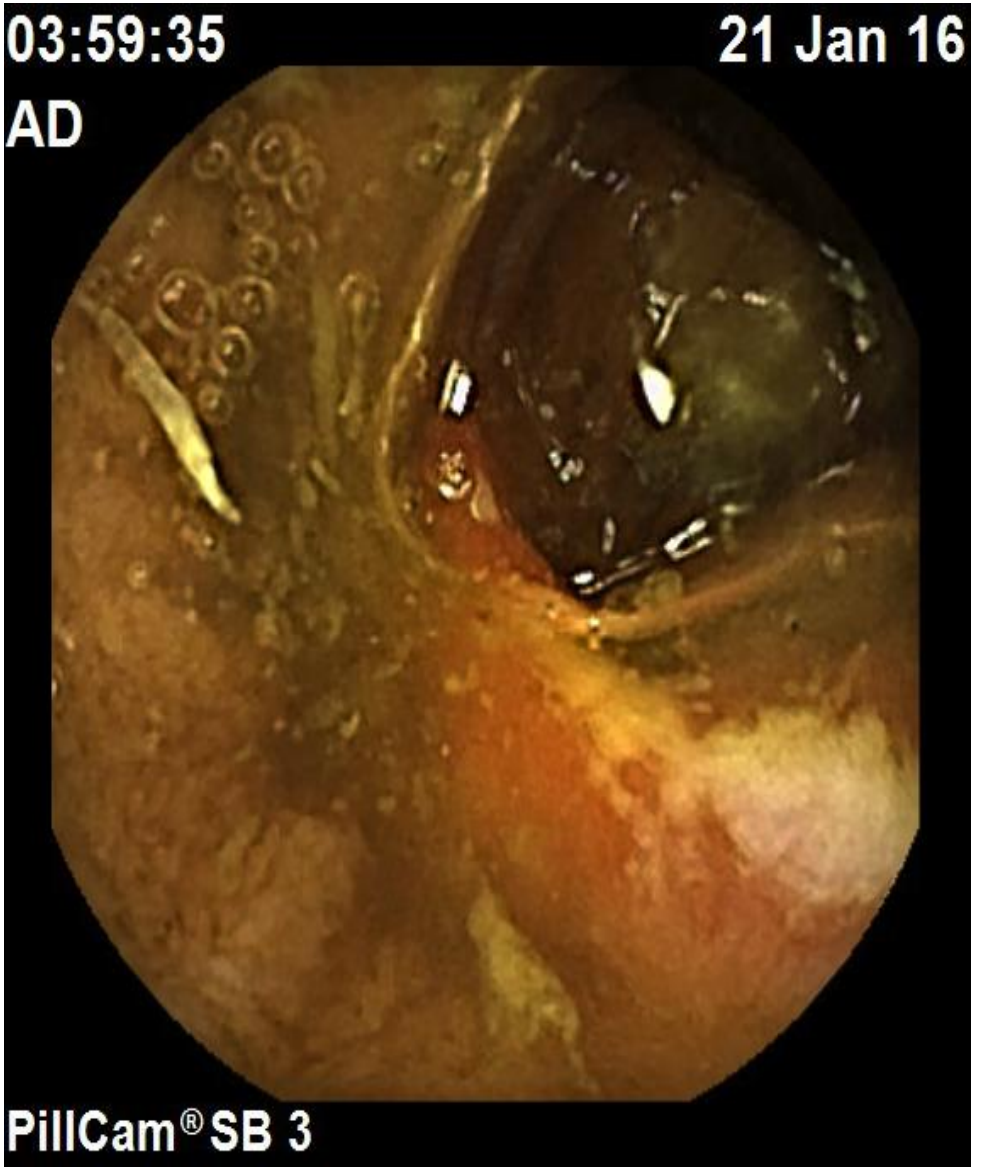


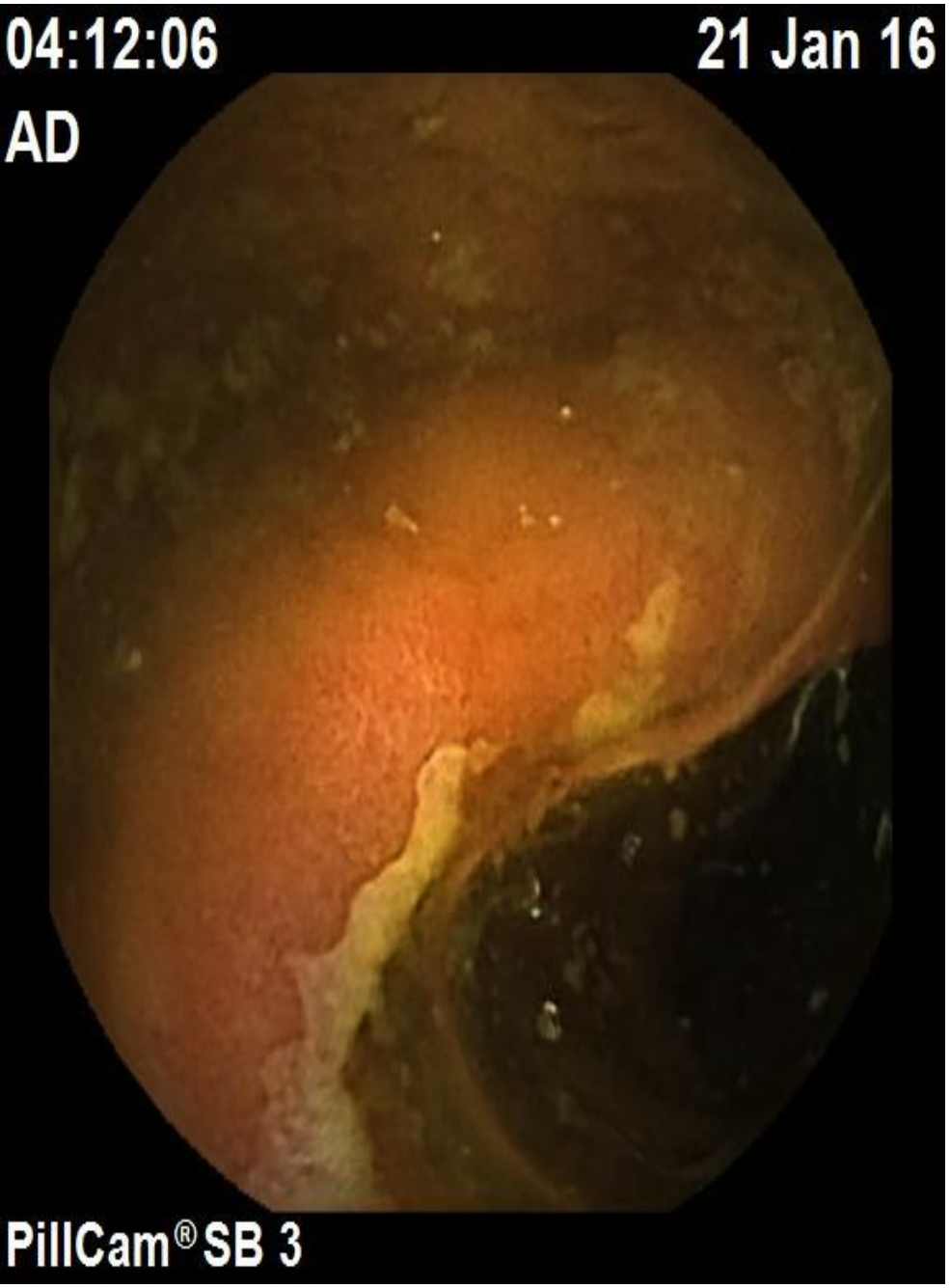
Normal Histology

Next step for obscure bleeding?

Video capsule endoscopy



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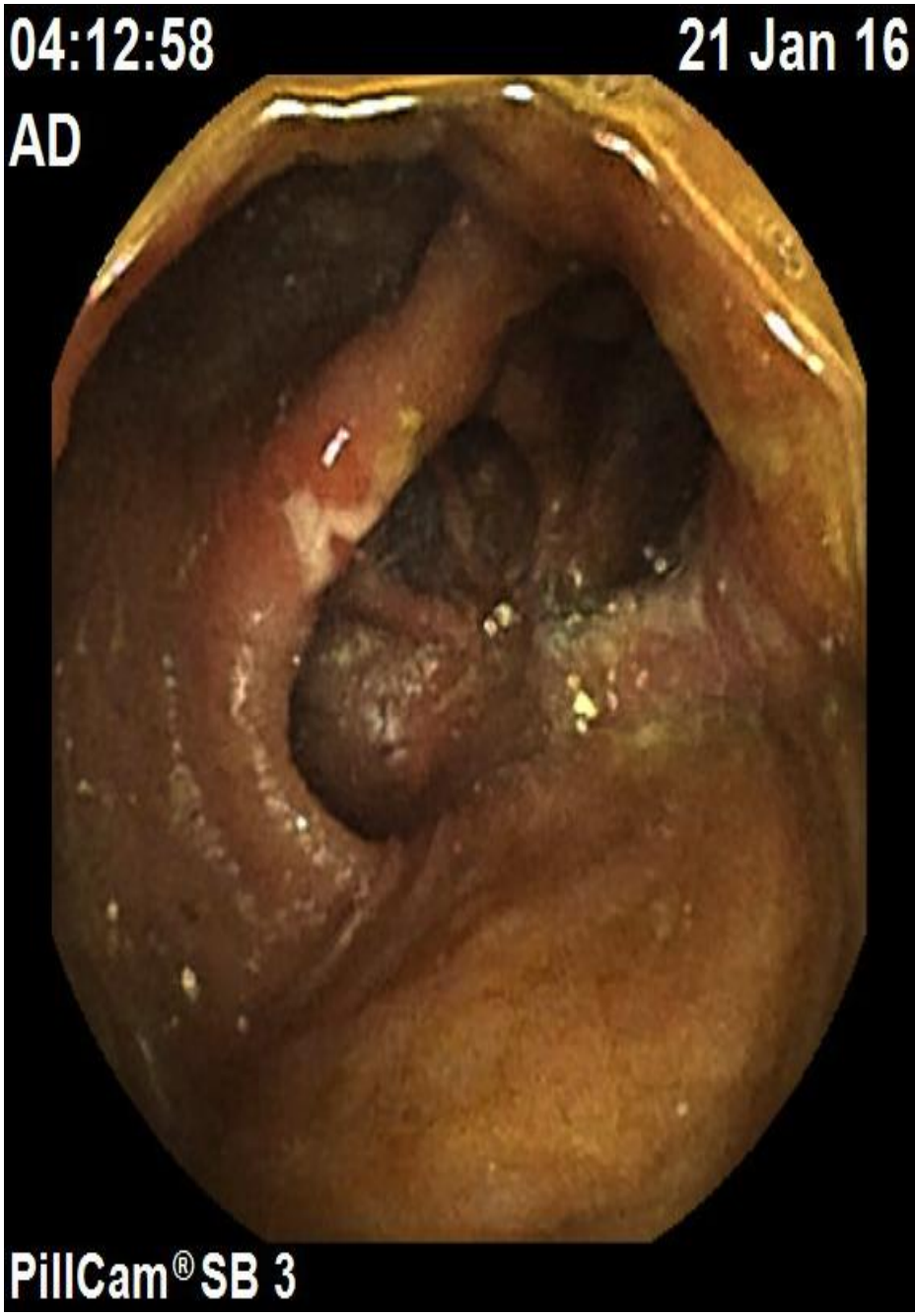
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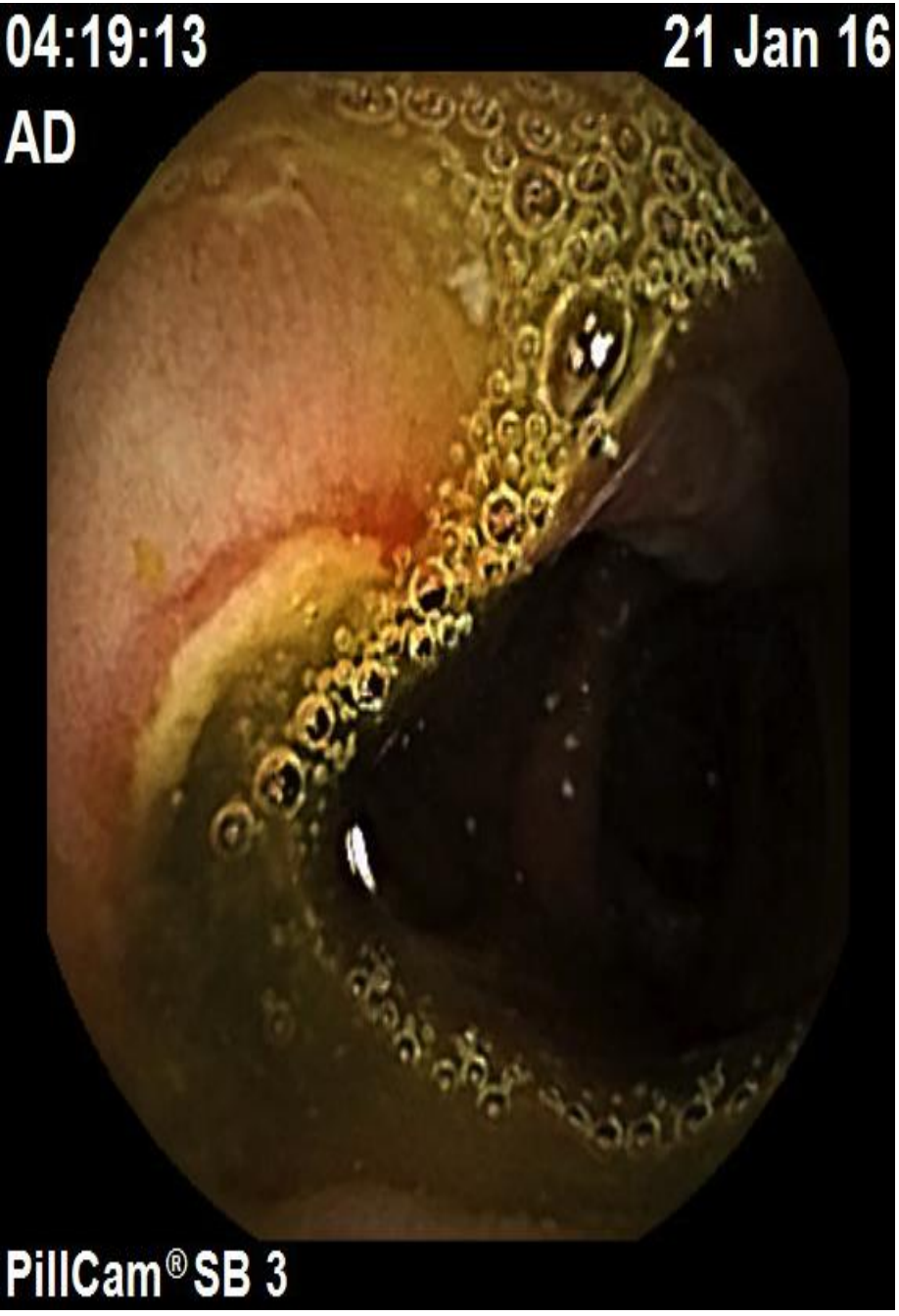
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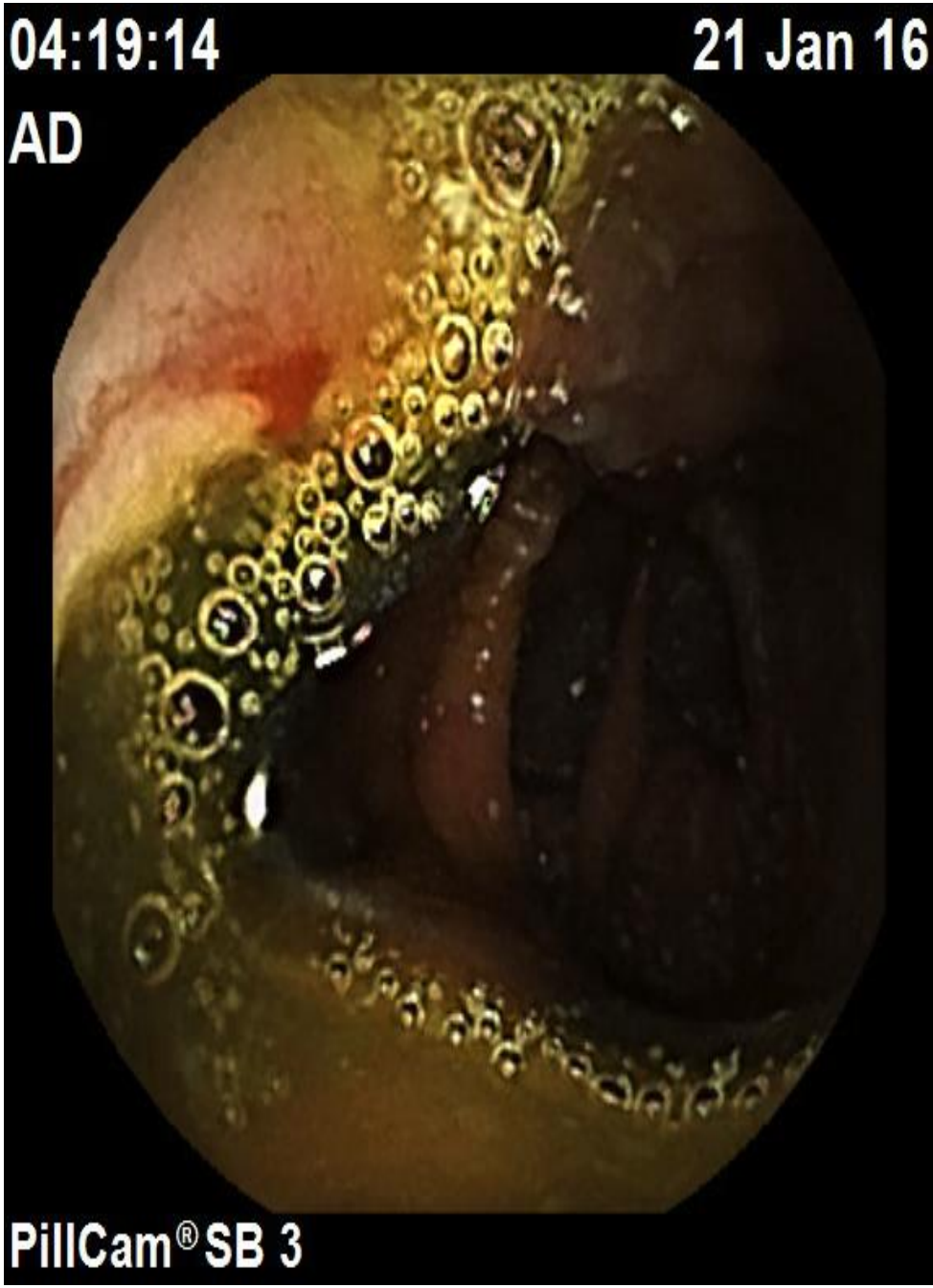
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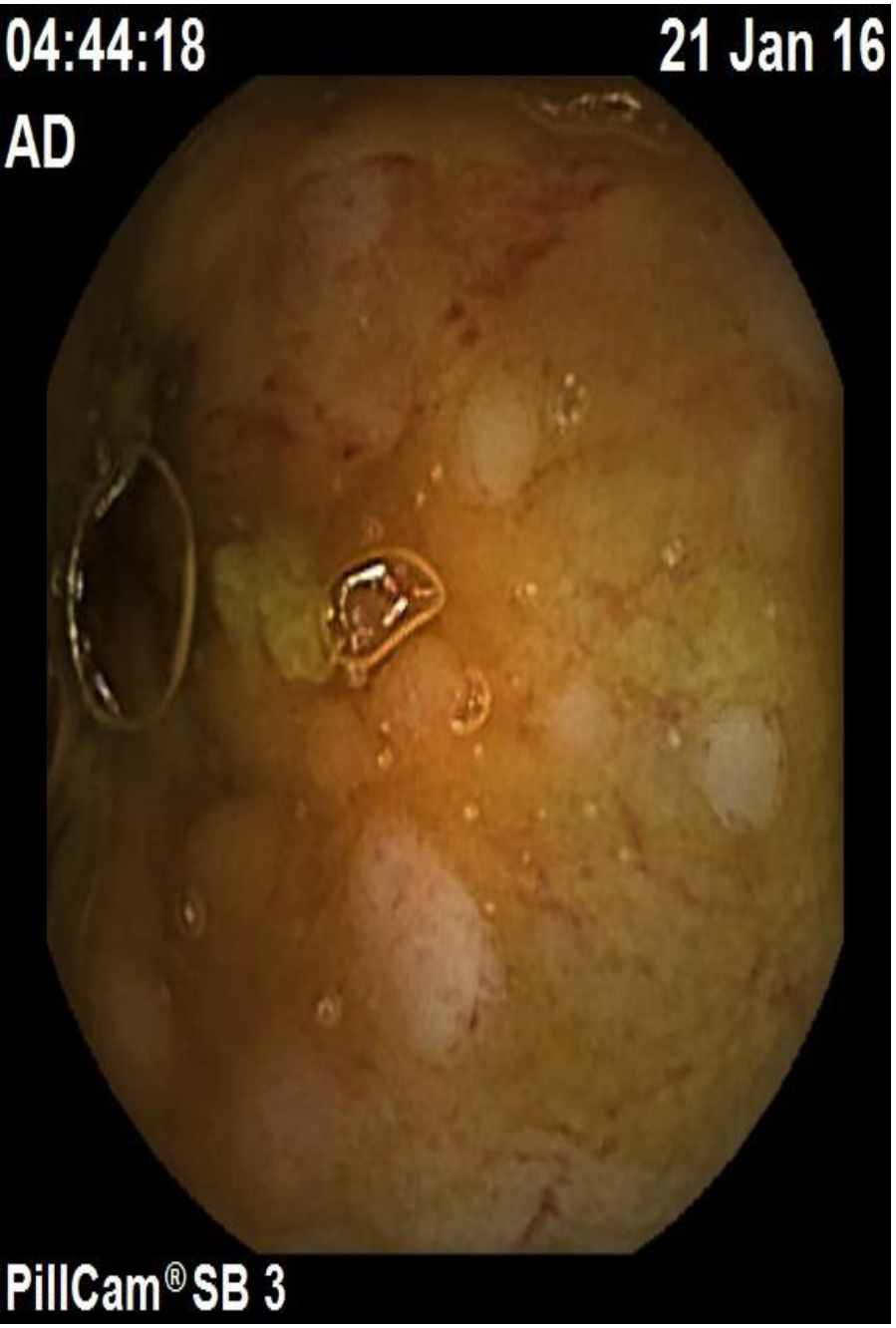
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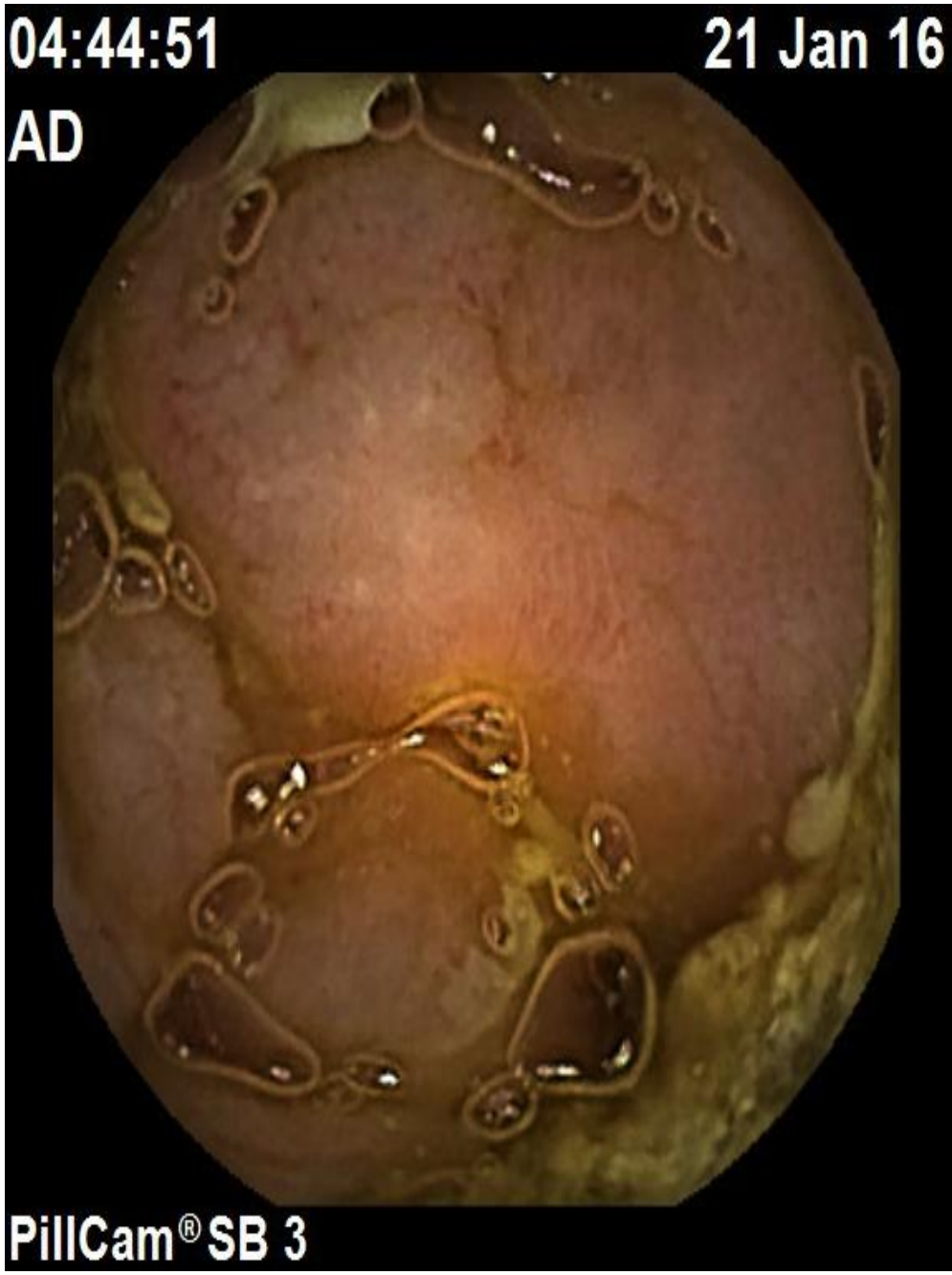
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Planned retrograde double balloon enteroscopy

- Ulcers seen +-15cm into distal ileum
- Area of surgical anastomosis
- **Histology:** Acute ulcerative terminal ileitis with changes of mild chronicity

Management

- Entocort EC (budesonide)

9mg x1mo; 6mg x 1mo; 3mg x1mo; 3mg alt days x 1mo

- + Iron therapy

- **During month 3 of treatment:** Hb 11,1 (MCV and MCH normal); Iron 4,3 (transferrin / % saturation / ferritin normal). Child was well with only mild cushingoid appearance

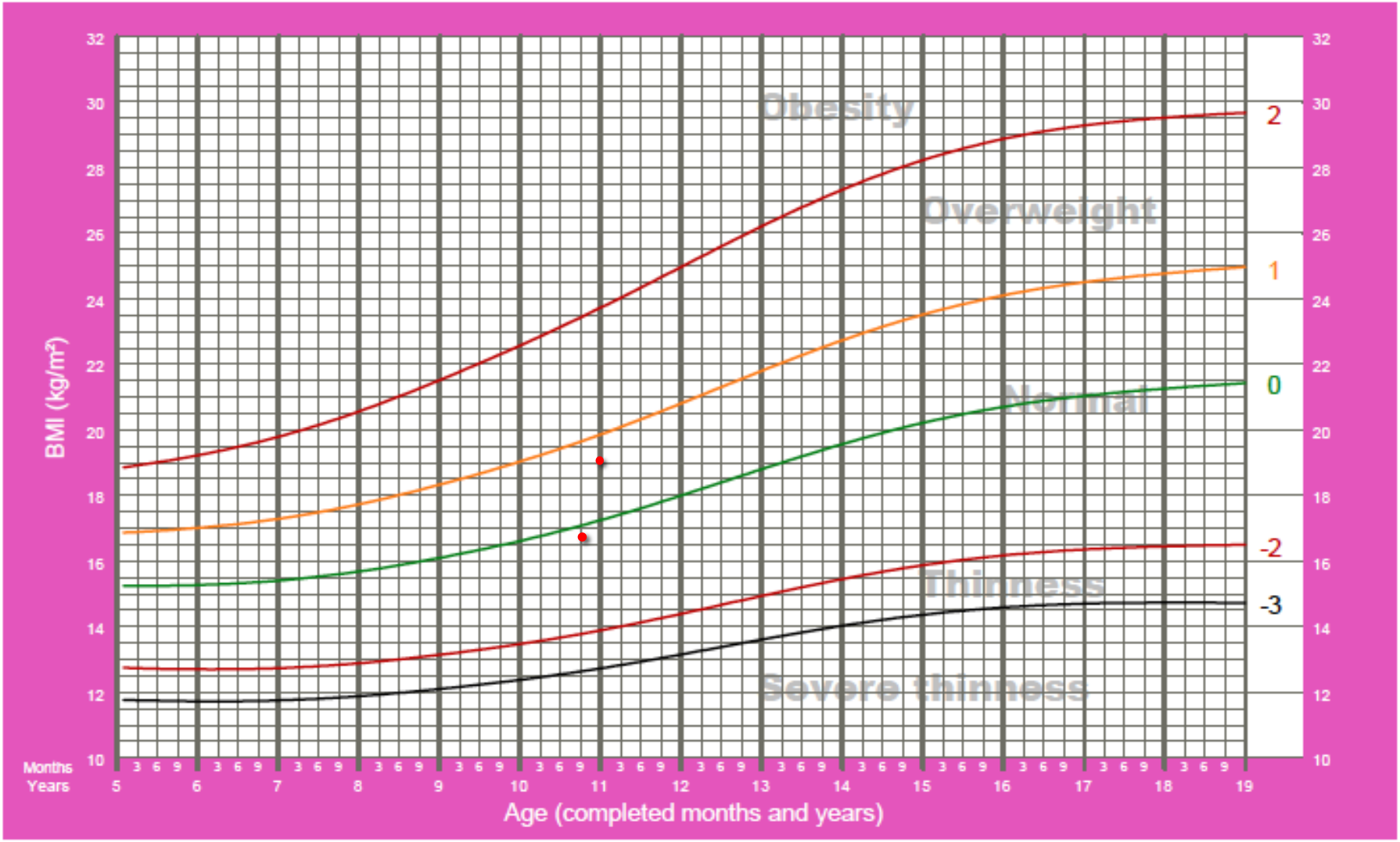
- Due for follow up and repeat FBC, iron studies + FC

- What should long-term management of these isolated TI ulcers be? Stop entocort and follow up clinically and biochemically?

BMI-for-age GIRLS

5 to 19 years (z-scores)

World Health Organization



2007 WHO Reference

PATIENT KP

2 year 2 month old boy

History

- Diarrhea, for almost 2 years (from around age 6 months)
- Stools: watery, no PR bleeding, up to 6 a day and also during the night
- Crying and irritability
- No clear abdominal pain, no fever, no vomiting, no rashes

Birth history

- Unremarkable

Growth and Development

- Within normal limits

Family history

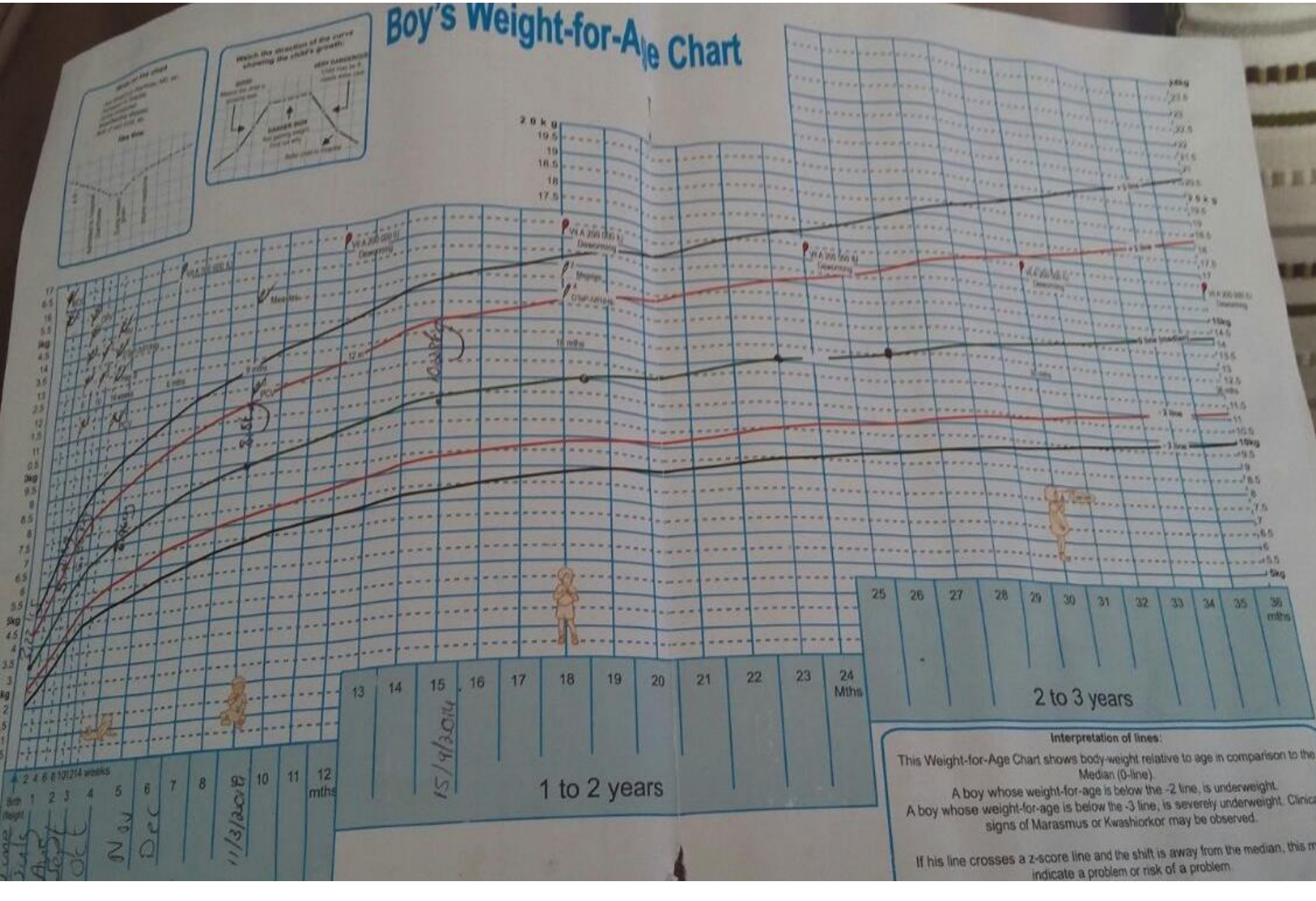
Brother with eczema, improved on dairy restricted diet

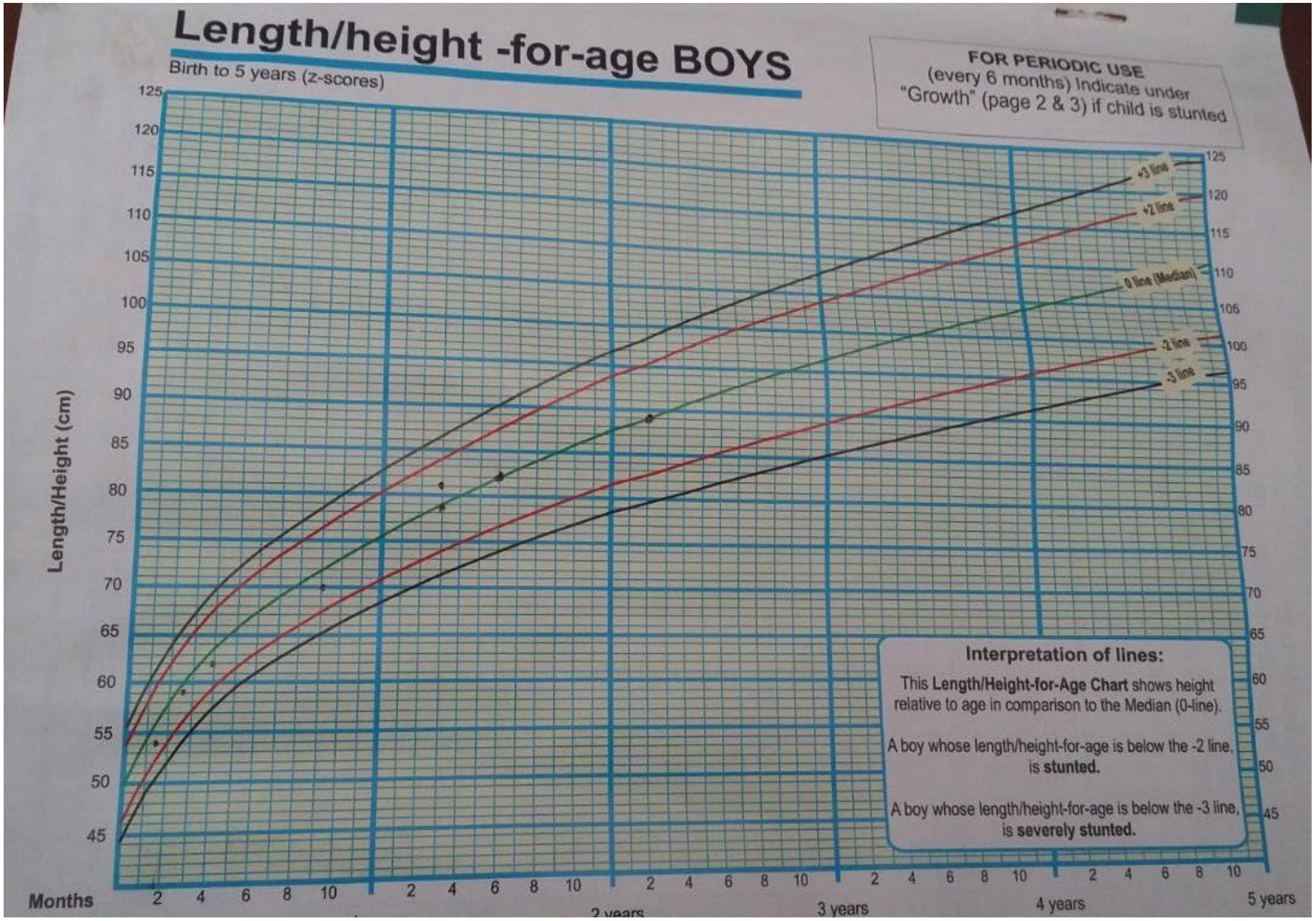
Allergies

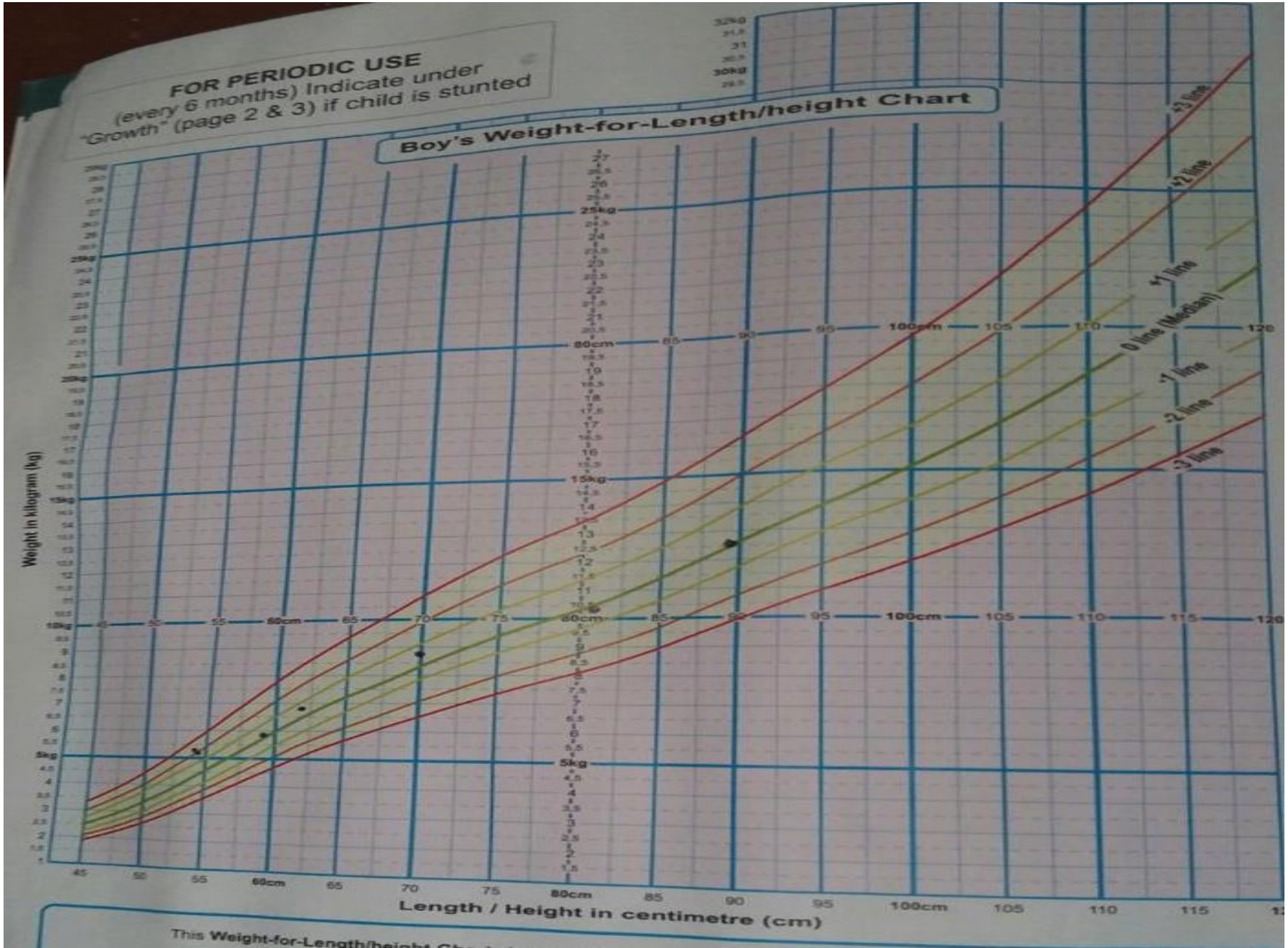
- None known

Past medical / surgical history

- 2 URTIs with fever, nil else


Boy's Weight-for-Age Chart
Watch the direction of the curve showing the child's growth.
1 to 2 years
2 to 3 years
Interpretation of lines:
This Weight-for-Age Chart shows body-weight relative to age in comparison to the
Median (0-line).
A boy whose weight-for-age is below the -2 line, is underweight.
A boy whose weight-for-age is below the -3 line, is severely underweight. Clinica
signs of Marasmus or Kwashiorkor may be observed.
If his line crosses a z-score line and the shift is away from the median, this m
indicate a problem or risk of a problem


Length/height -for-age BOYS
Birth to 5 years (z-scores)
FOR PERIODIC USE
(every 6 months) Indicate under
"Growth" (page 2 & 3) if child is stunted
Length/Height (cm)
125
120
115
110
105
100
95
90
85
80
75
70
65
60
55
50
45
+3 line
+2 line
0 line (Median)
-2 line
-3 line
Interpretation of lines:
This Length/Height-for-Age Chart shows height relative to age in comparison to the Median (0-line).
A boy whose length/height-for-age is below the -2 line, is stunted.
A boy whose length/height-for-age is below the -3 line, is severely stunted.
Months
2 4 6 8 10
2 years
3 years
4 years
5 years


FOR PERIODIC USE
(every 6 months) Indicate under "Growth" (page 2 & 3) if child is stunted
Boy's Weight-for-Length/height Chart
+3 line
+2 line
+1 line
0 line (Median)
-1 line
-2 line
-3 line
Weight in kilogram (kg)
Length / Height in centimetre (cm)
This Weight-for-Length/height Ch

Examination

- Not acute / chronically ill
- Normal vitals
- No jaundice, pallor, clubbing,
- No skin rashes
- No features of nutritional deficiencies
- Systemic examination unremarkable

Investigations

- **Faecal elastase:** normal
- **Faecal calprotectin:** > 600
- **Stool mcs:** negative
- **Stool C-diff:** negative
- **FBC:** Hb 9,9; HCT 0,32; MCV 67; MCH 21
  WCC 16,6 (↑) – normal neutr,lymph,eosin; Plts 467 (↑)
- **Iron studies:** S-Fe 3,4 (9-21.5); S-transferrin normal; %-saturation 4; Ferritin 16 (20-300)
- **CRP:** 10
- **ESR:** 5
- **CEU:** Na 135 (↓), S-bicarb 18 (↓)
- **LFTs:** normal
- TP 71 / Alb 41
- IGG, IGM, IGA normal
- **TTG IGA:** normal
- RAST paed mix FX: normal

Gastroscopy / Colonoscopy

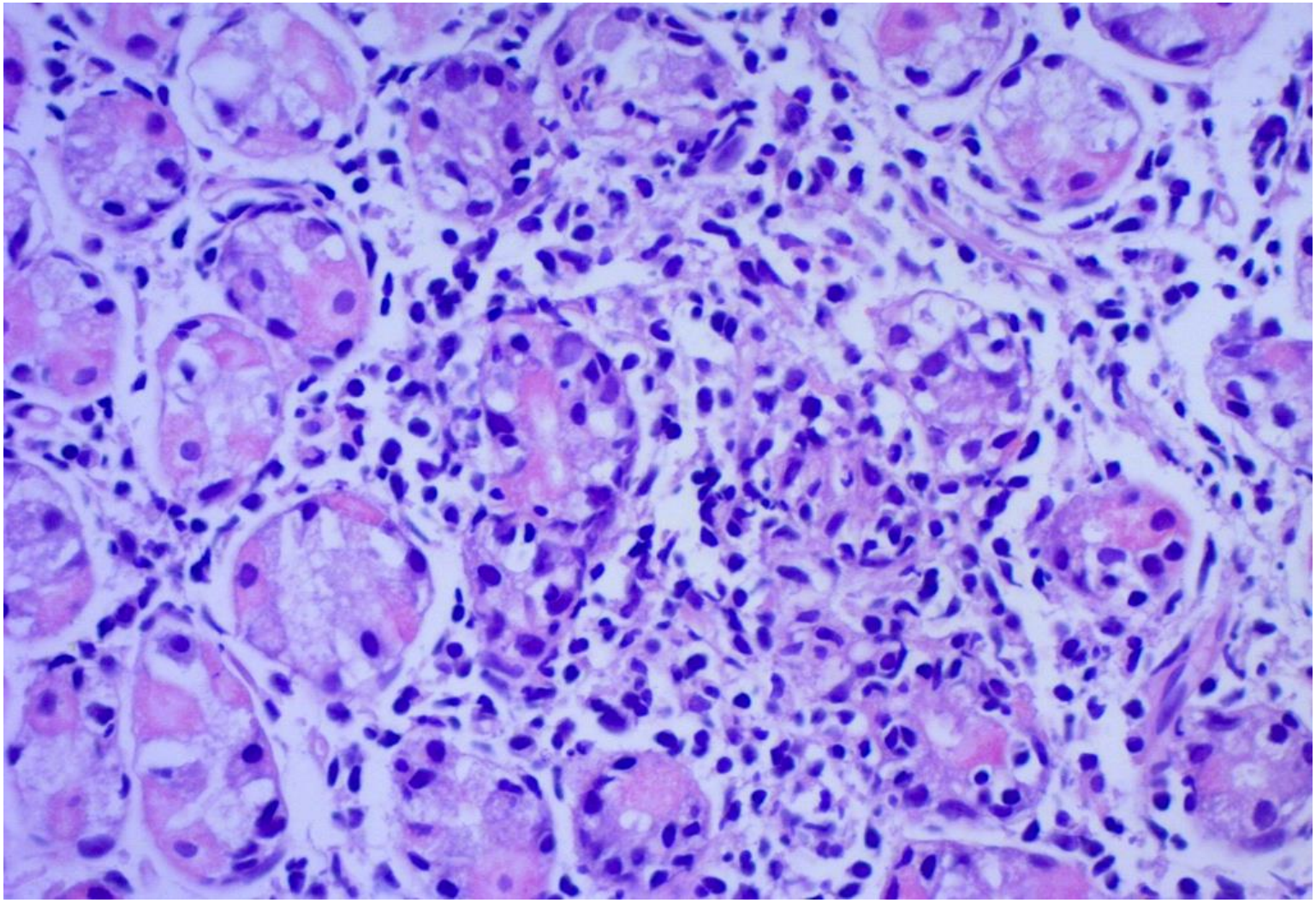
Macroscopic:

- G-scope: normal
- C-scope: pancolitis - anorectal junction to caecum - loss of vascular markings, mucosal edema and contact bleeding

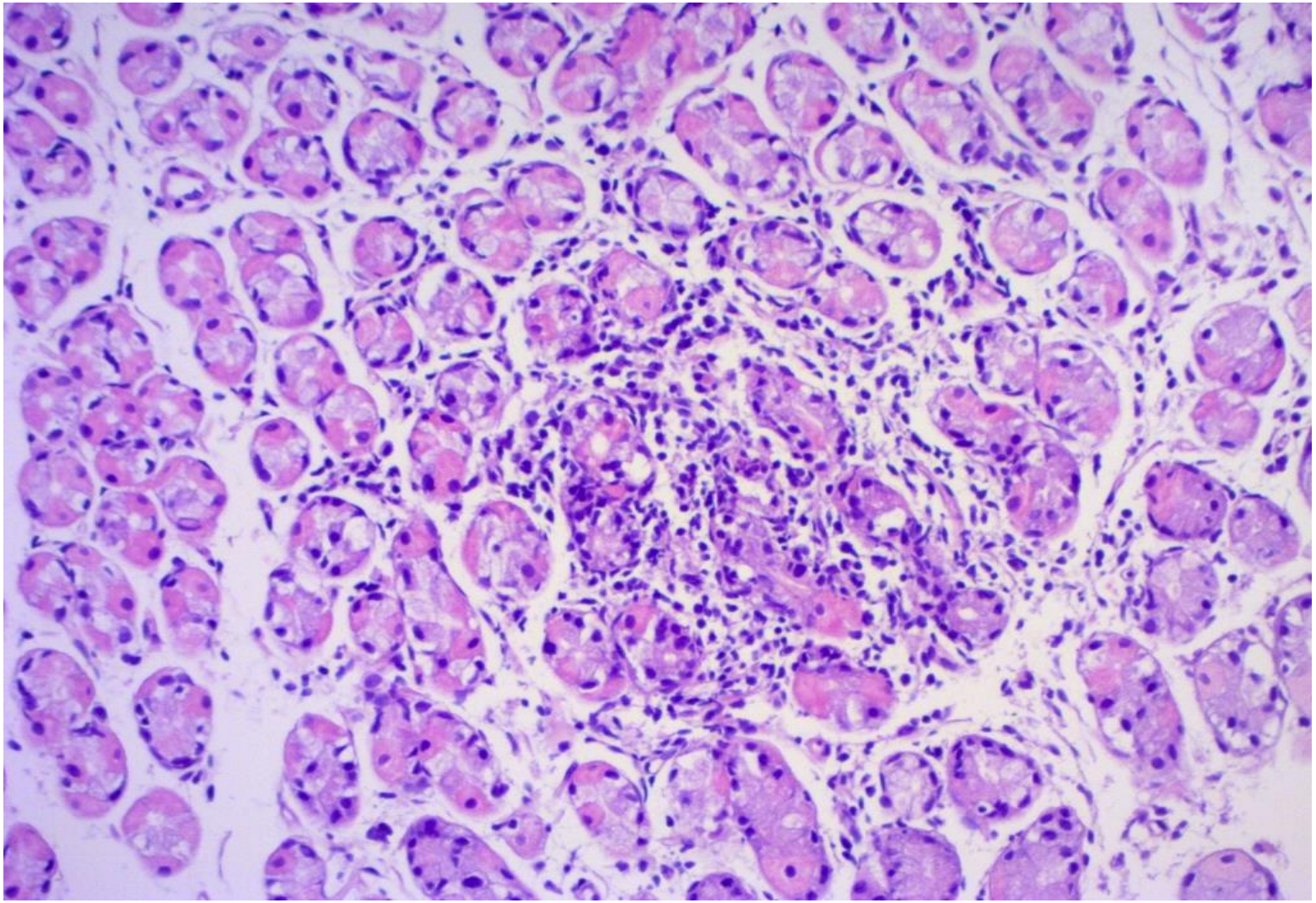
Histology:

- Esophagus normal
- Duodenum normal

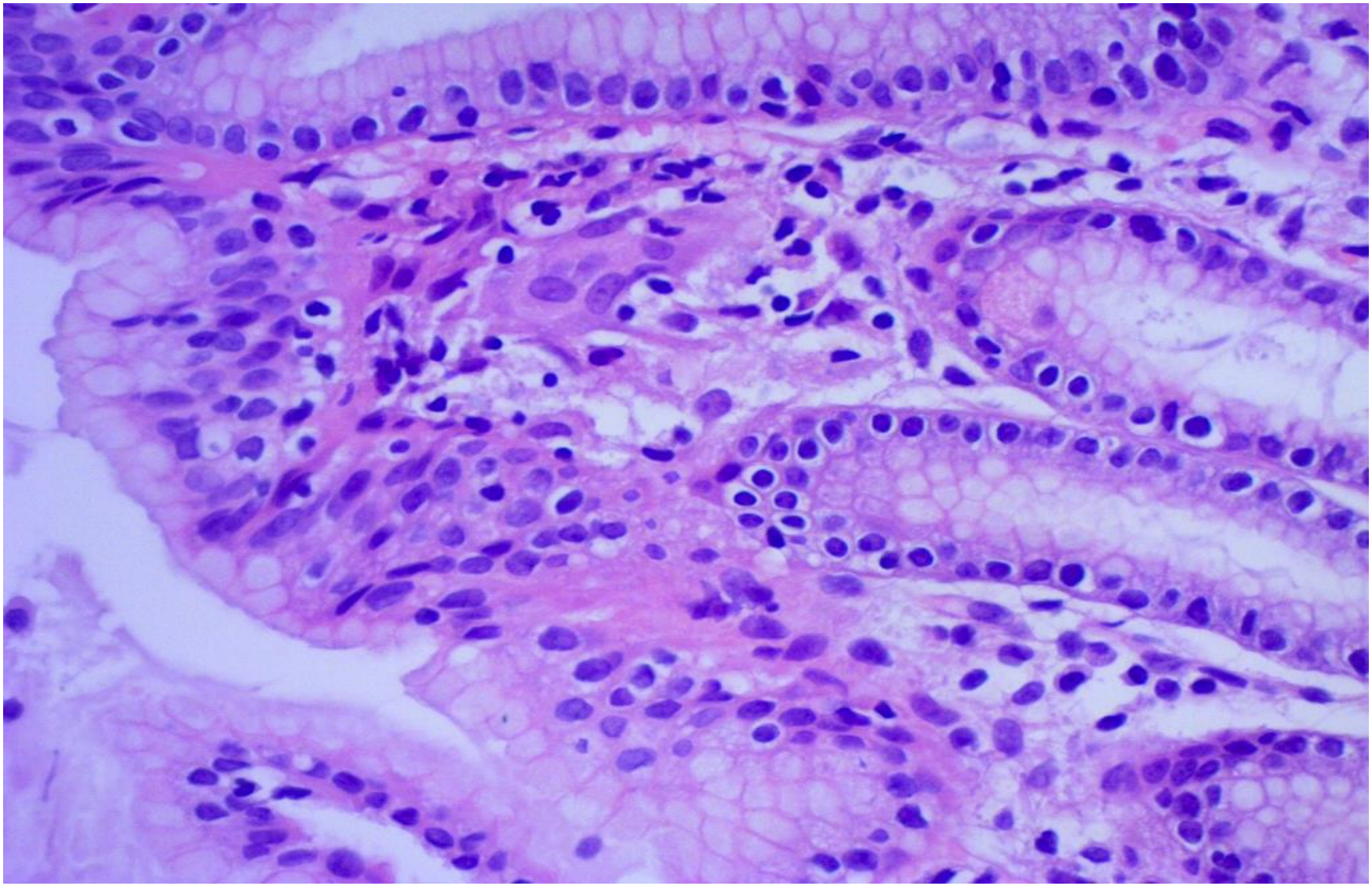
Gastroscopy / Colonoscopy



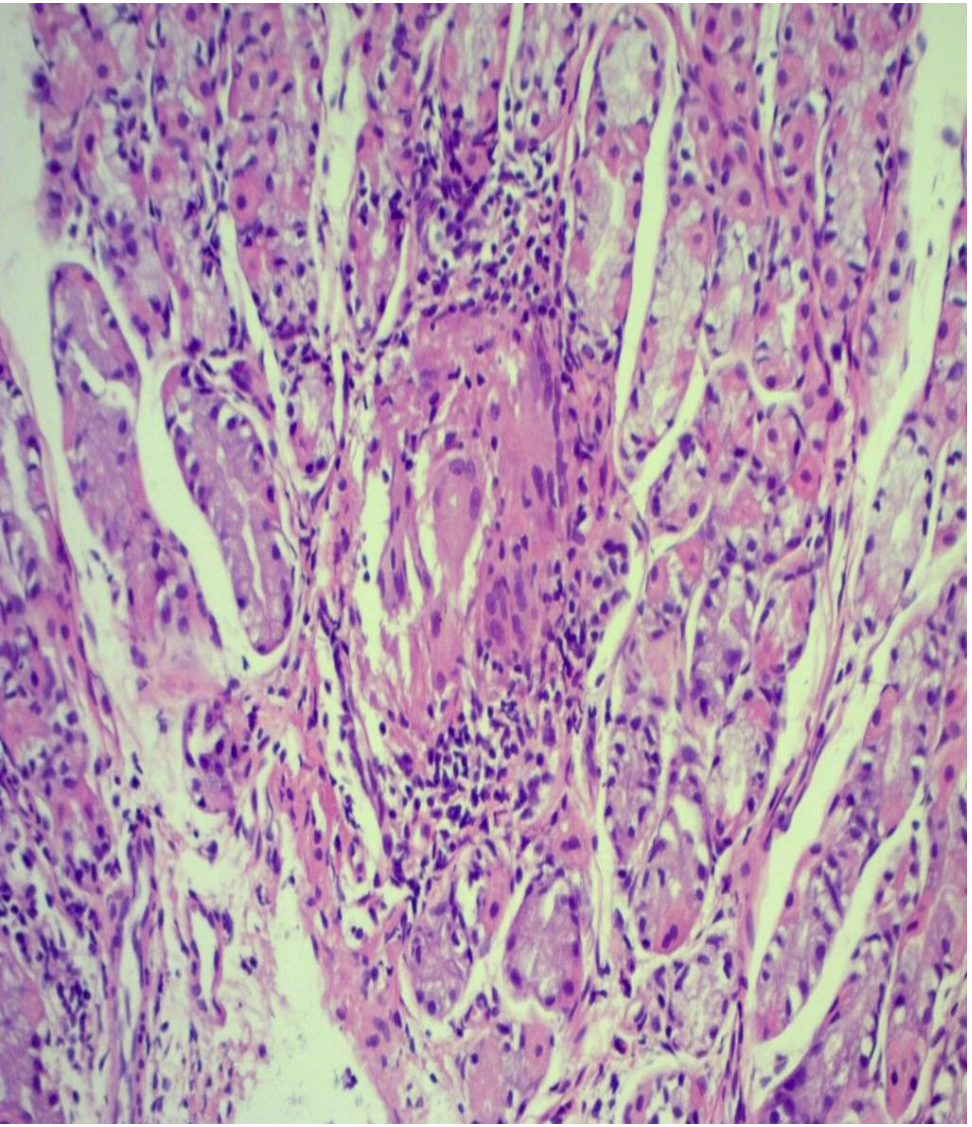
Stomach biopsy showing crypt destruction

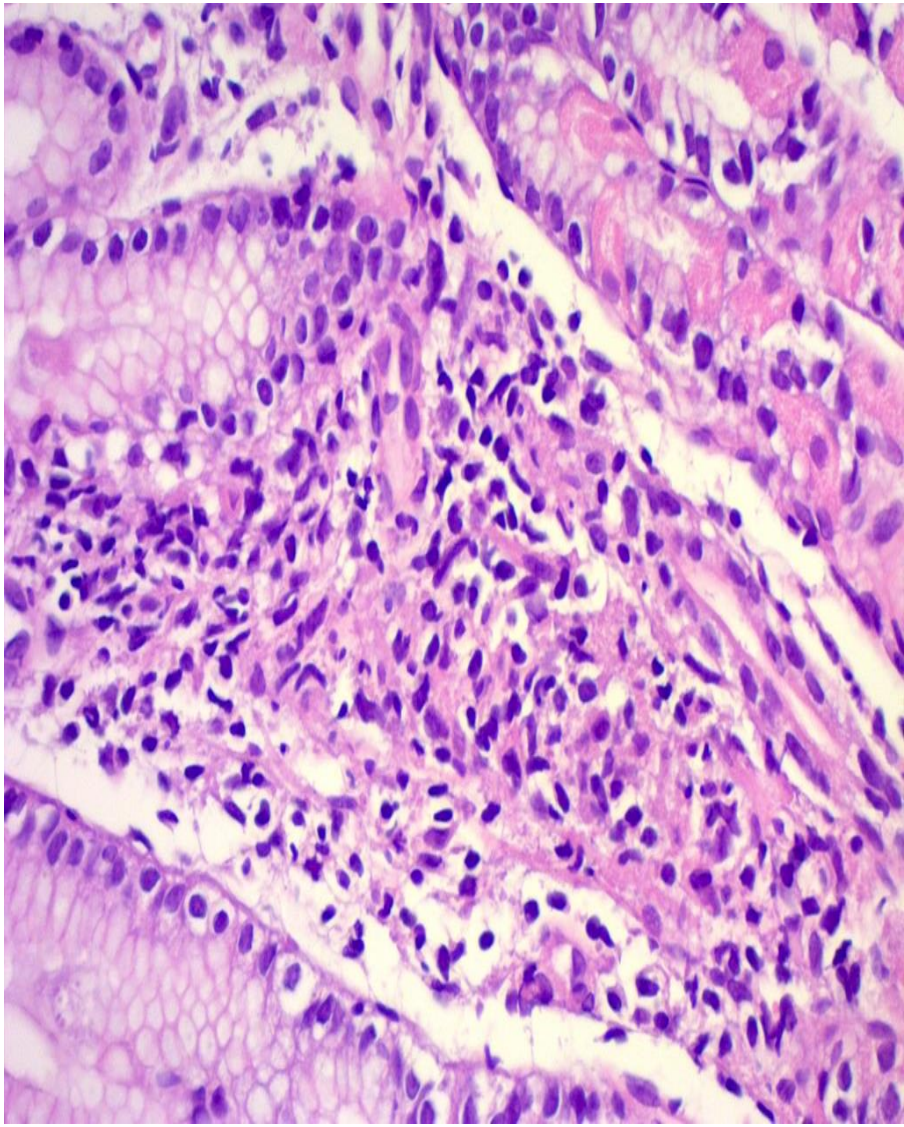


Crypt granuloma H&E 200x

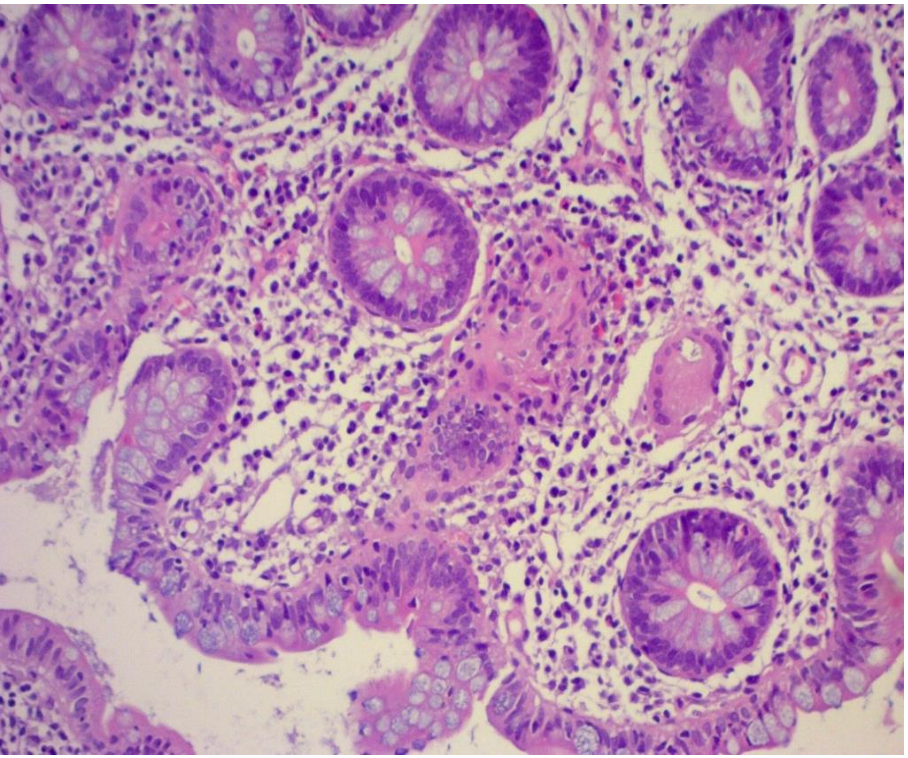


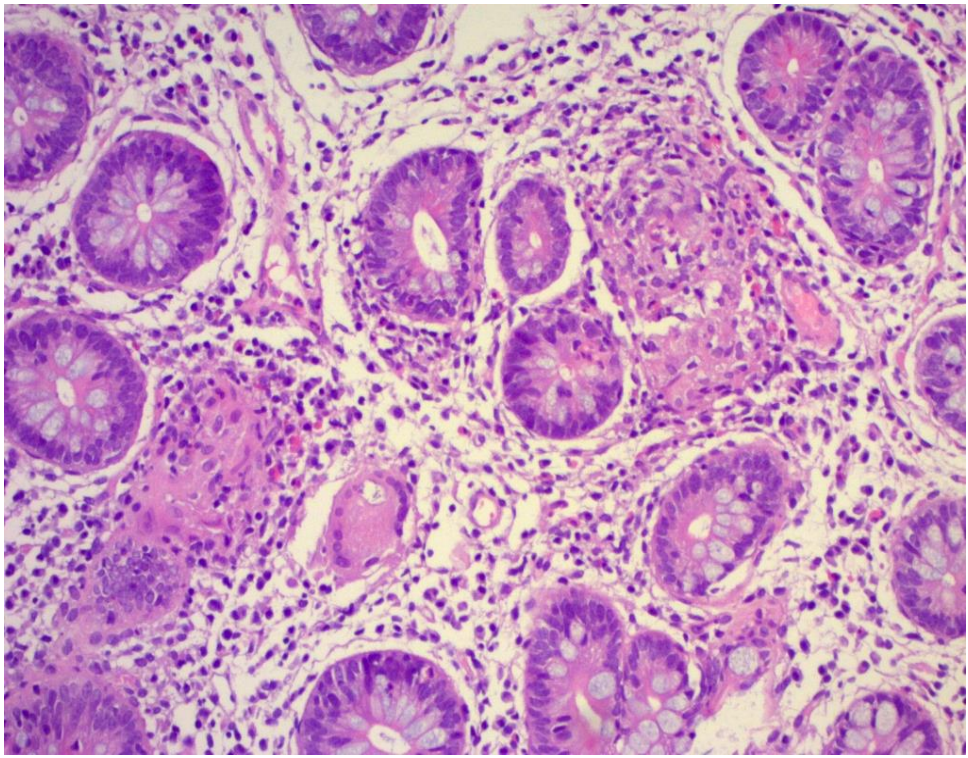
Crypt microgranuloma H&E 400x



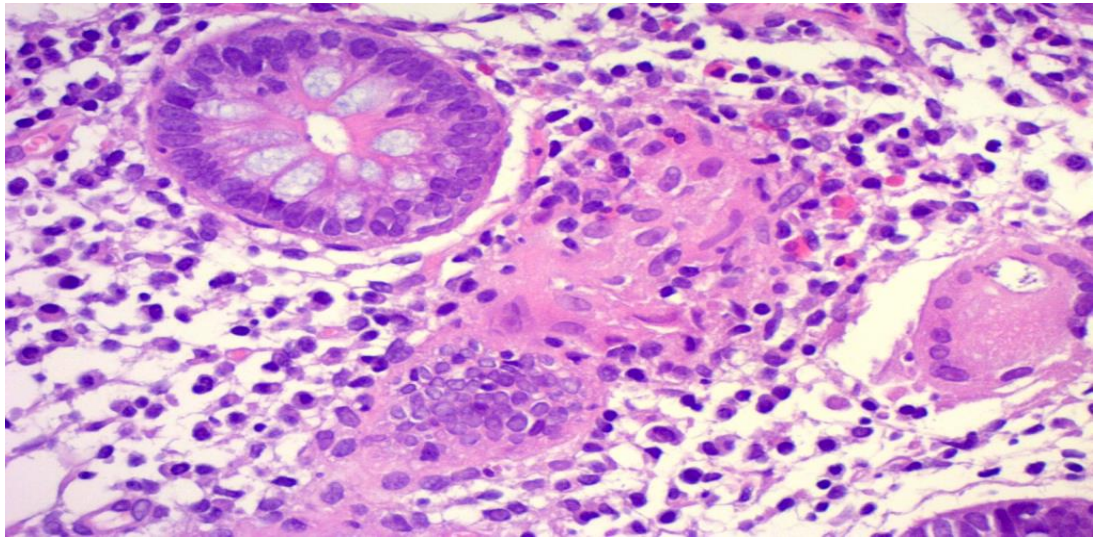


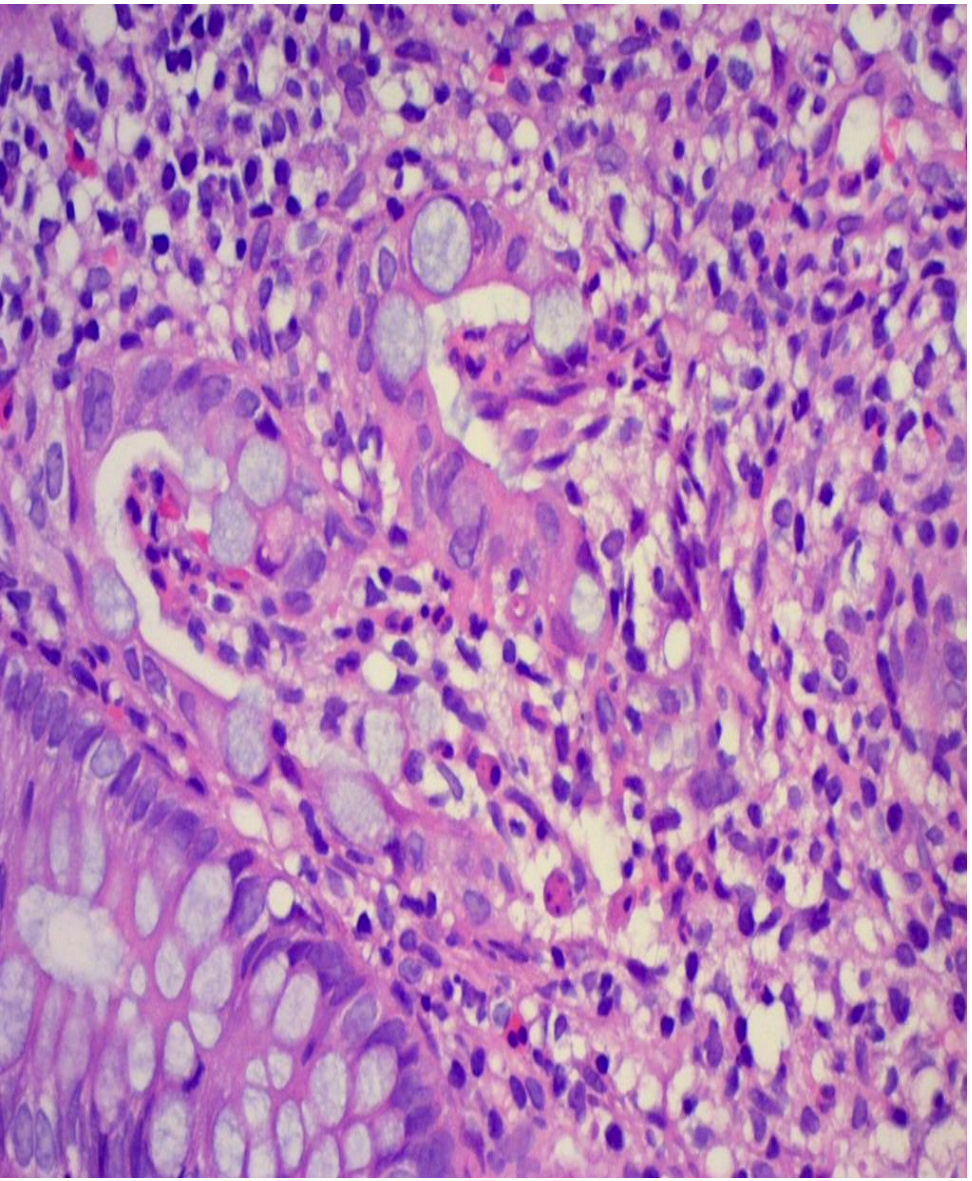
Stomach biopsy: chronic gastritis and crypt destruction

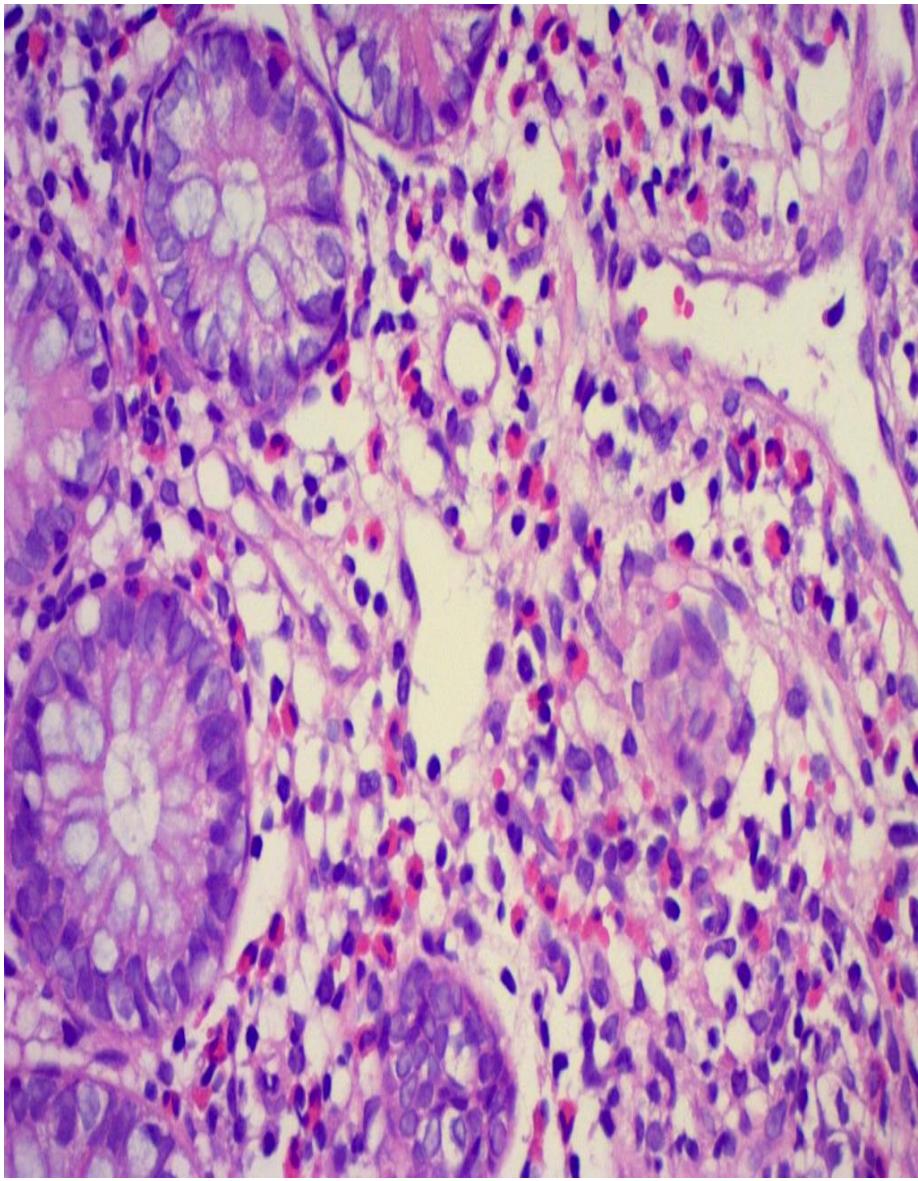




Terminal ileum portraying microgranulomas and increased eosinophils







Colon with active colitis and as shown, markedly increased eosinophils and a microgranuloma

Eosinophilic gastroenteritis (vs. Inflammatory bowel disease +- immune deficiency in a young child)

Management

- 6-food elimination diet
- Addition of 5-ASA (Pentasa 750mg od)

- Within a week of starting elimination diet, stools normalised
- Reported irritability and crying resolved
- Appetite good
- Gained weight

Follow up

- After about 3mo of normal stools and being asymptomatic, re-introduction of food was attempted

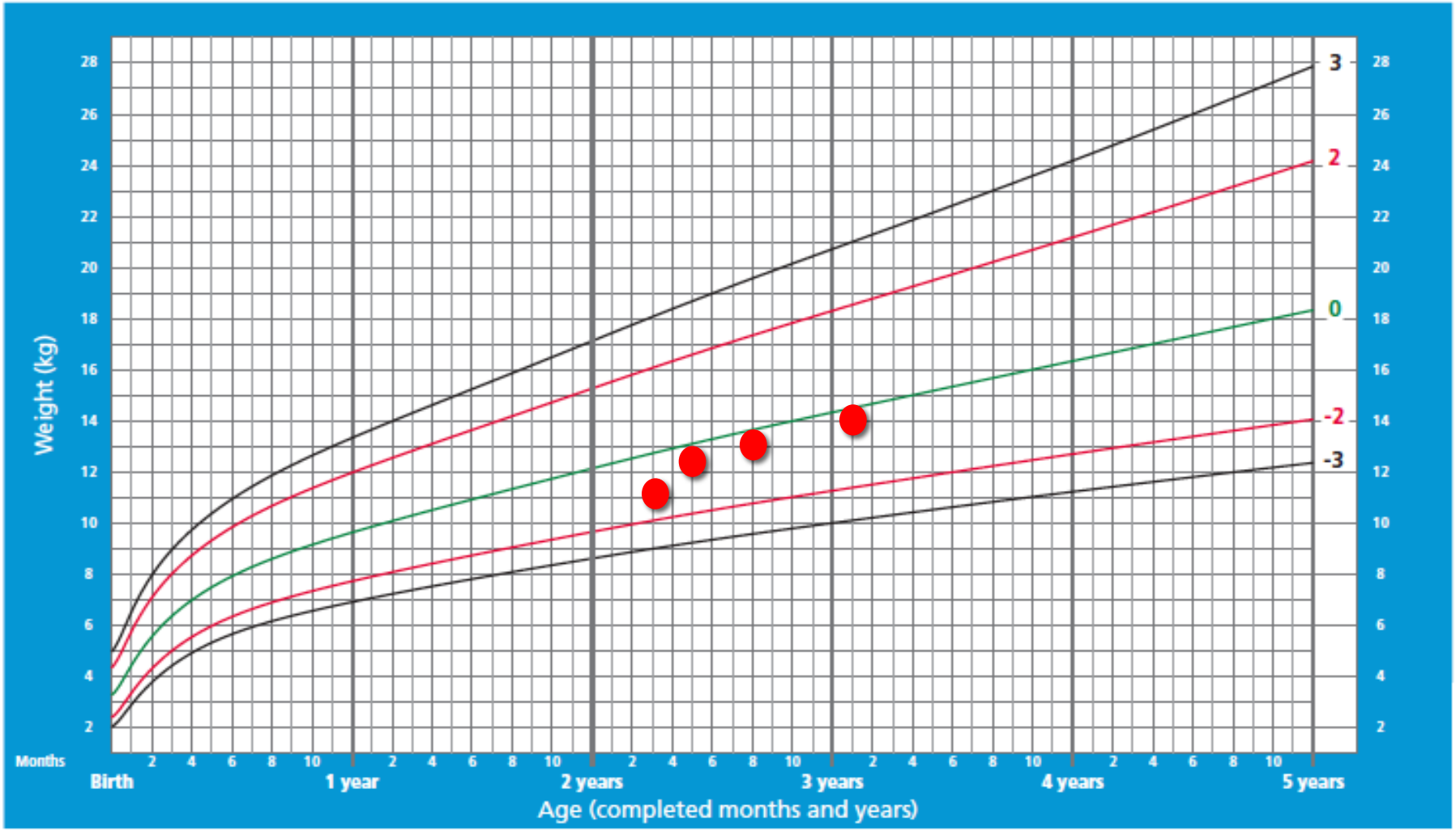
- Failed…
- Elimination diet re-instated (while continuing 5-ASA)
- Faecal calprotectin > 600

About 1 year later, Faecal calprotectin 977, asymptomatic with normal stools unless cheats on diet. Gaining weight and has a healthy appetite!

Weight-for-age BOYS

Birth to 5 years (z-scores)

World Health Organization



WHO Child Growth Standards

- Will have colonoscopy at the end of the month
- **?? Treatment options**

PATIENT RVM

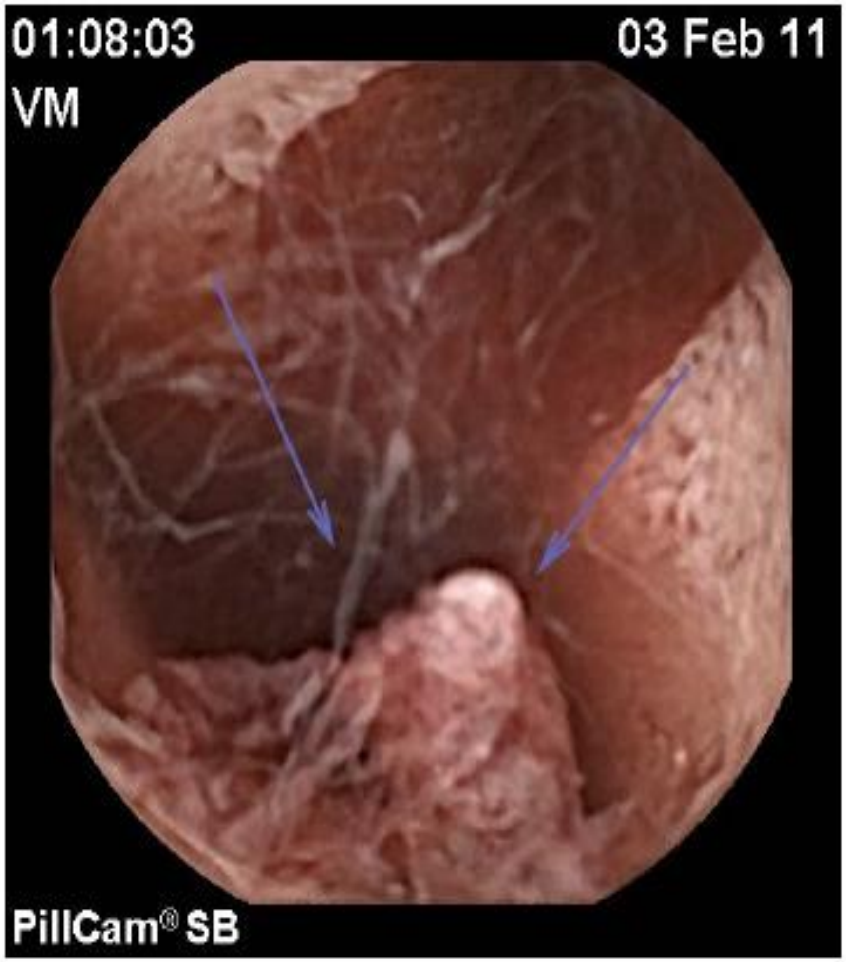
13 year old girl

History

- Noonan syndrome phenotype
- Well known since birth with pulmonary valve stenosis and ASD
- No genetic testing done due to parental hesitation and strong phenotypic features making another diagnosis unlikely
- As infant underwent balloon valvuloplasty followed by patch valvuloplasty and ASD closure at age 2 years
- At age 8 she presented with tiredness, diarrhea and bilateral pitting edema due to protein-losing enteropathy (Albumin 10, low globulins and lymphocytes)

Capsule endoscopy at age 8 years

- Villous atrophy in most of small bowel, with edema and lymphatic stasis
- Duodenal varices (cardiac related) with bleeding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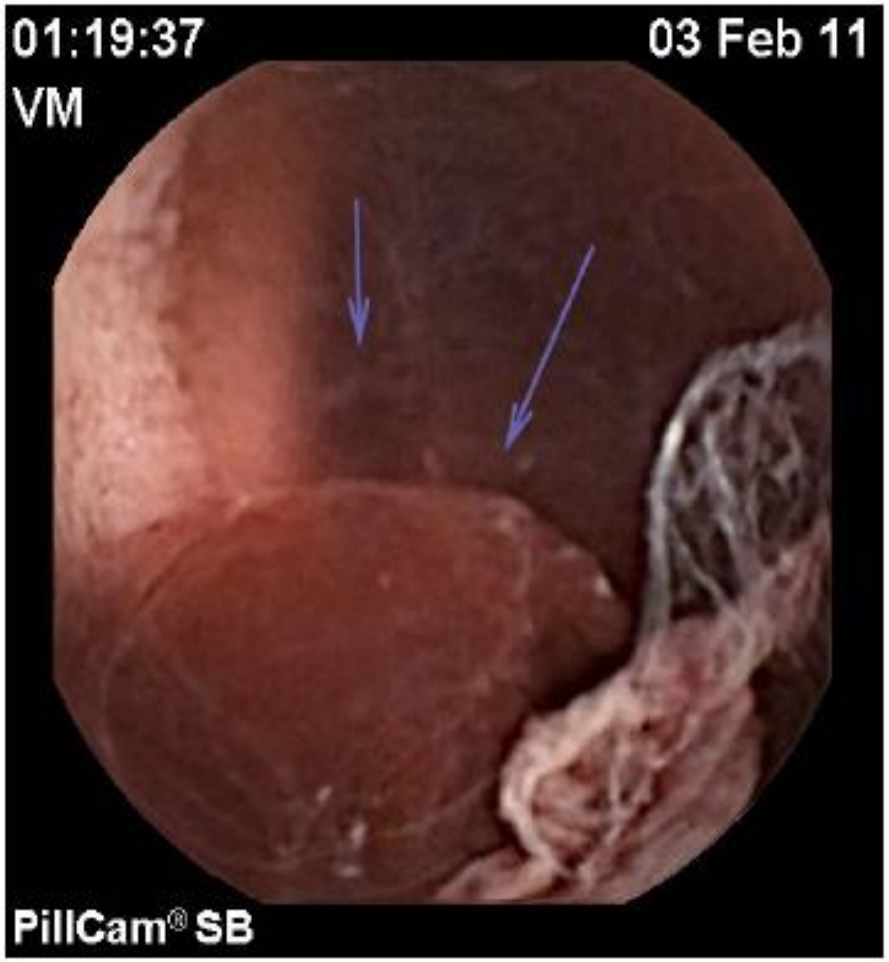
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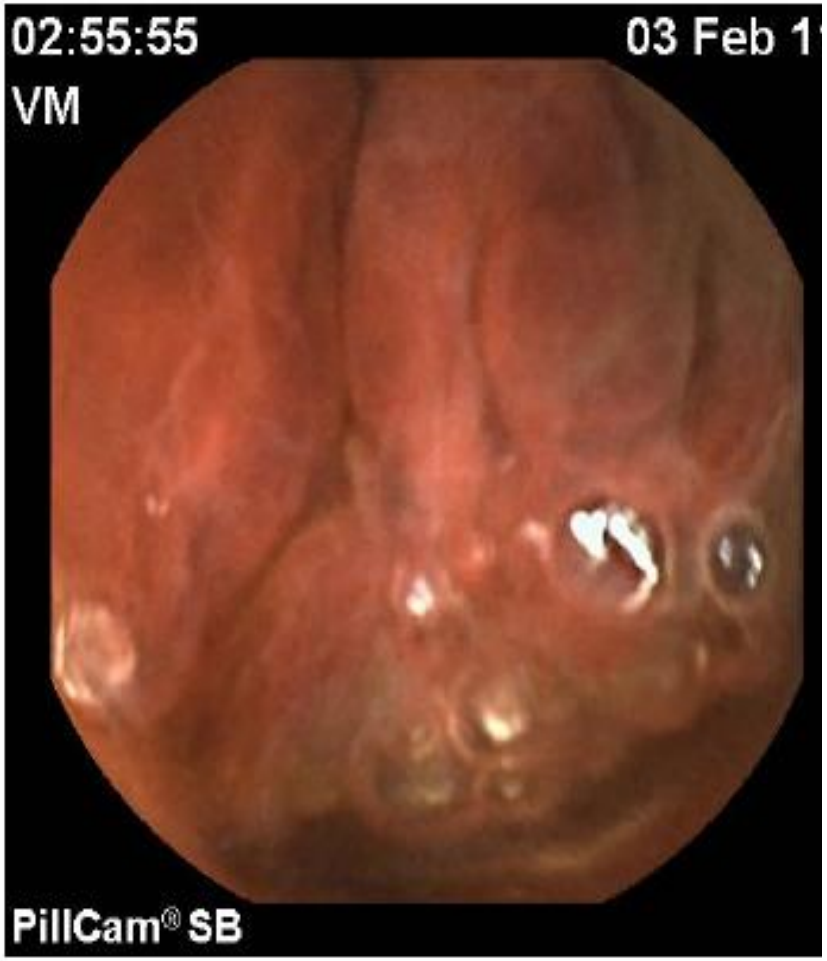
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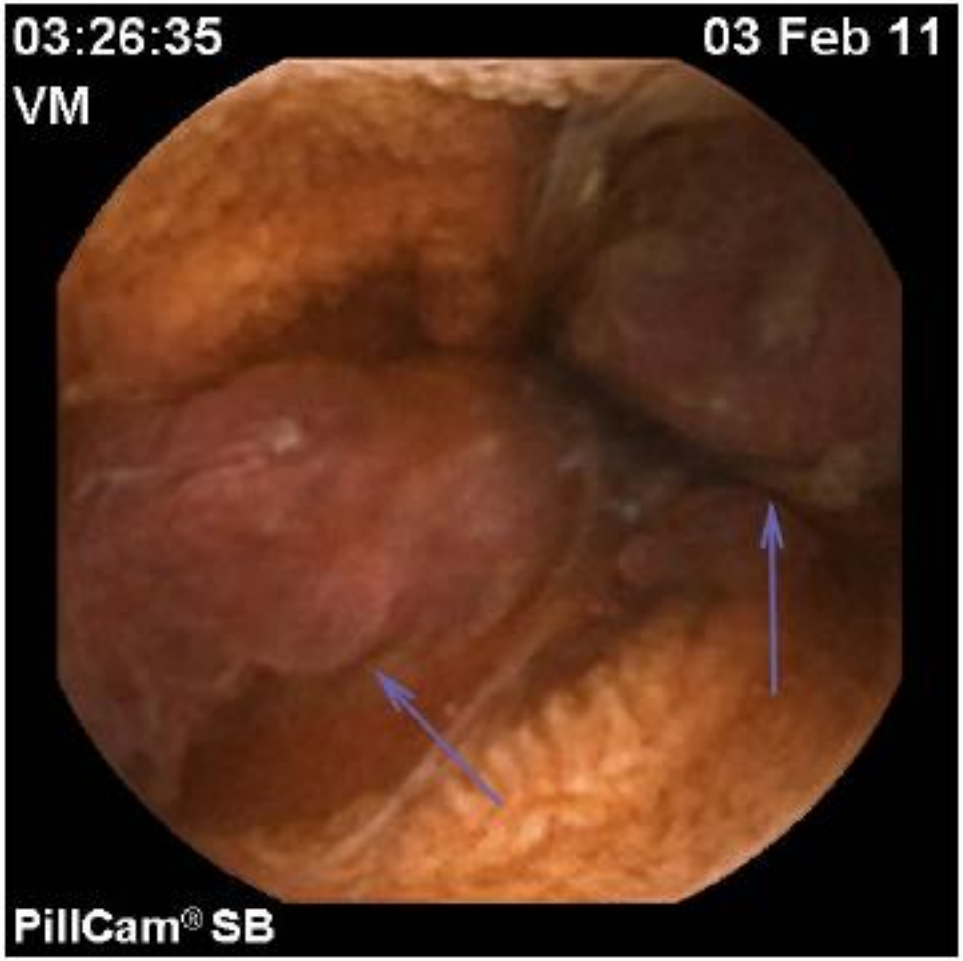
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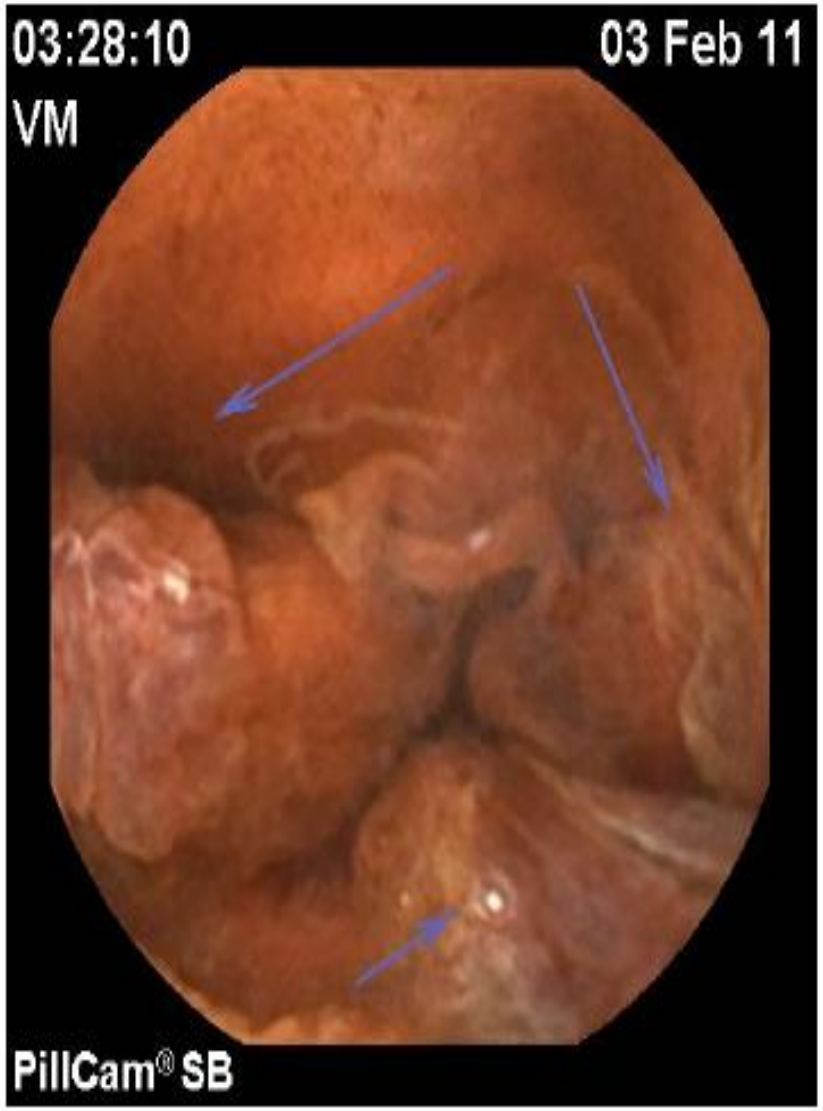
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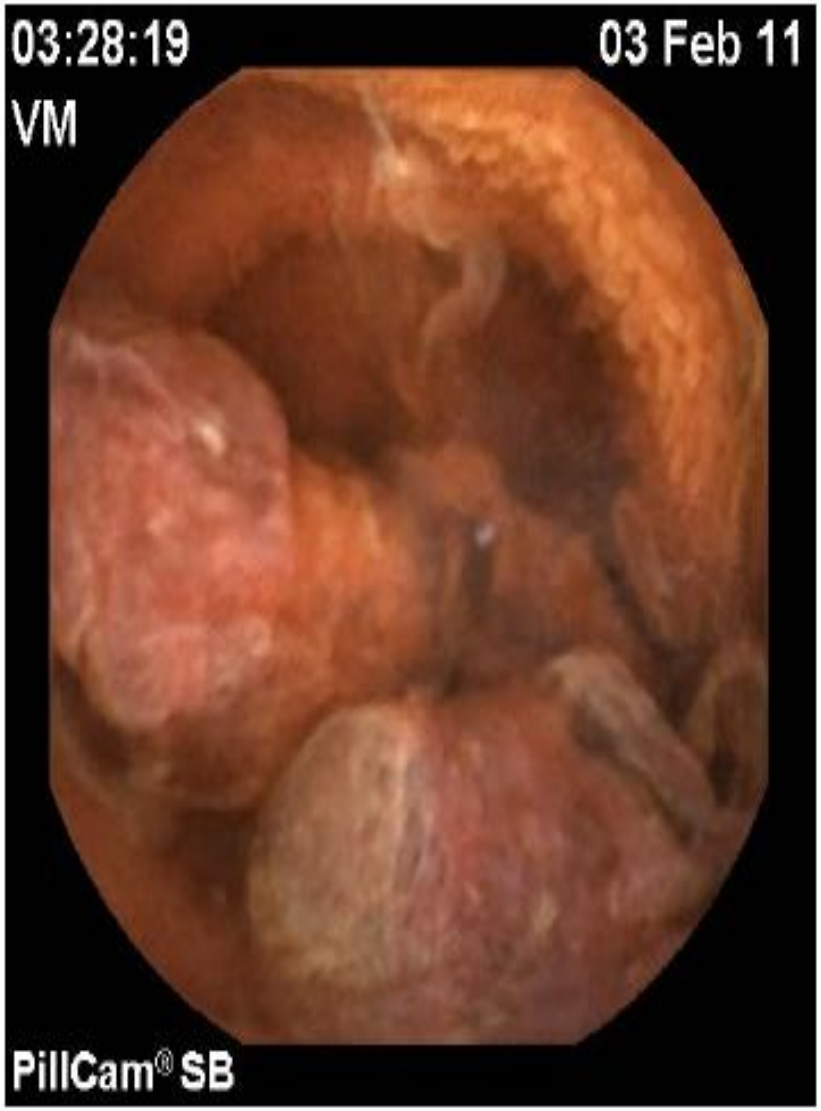
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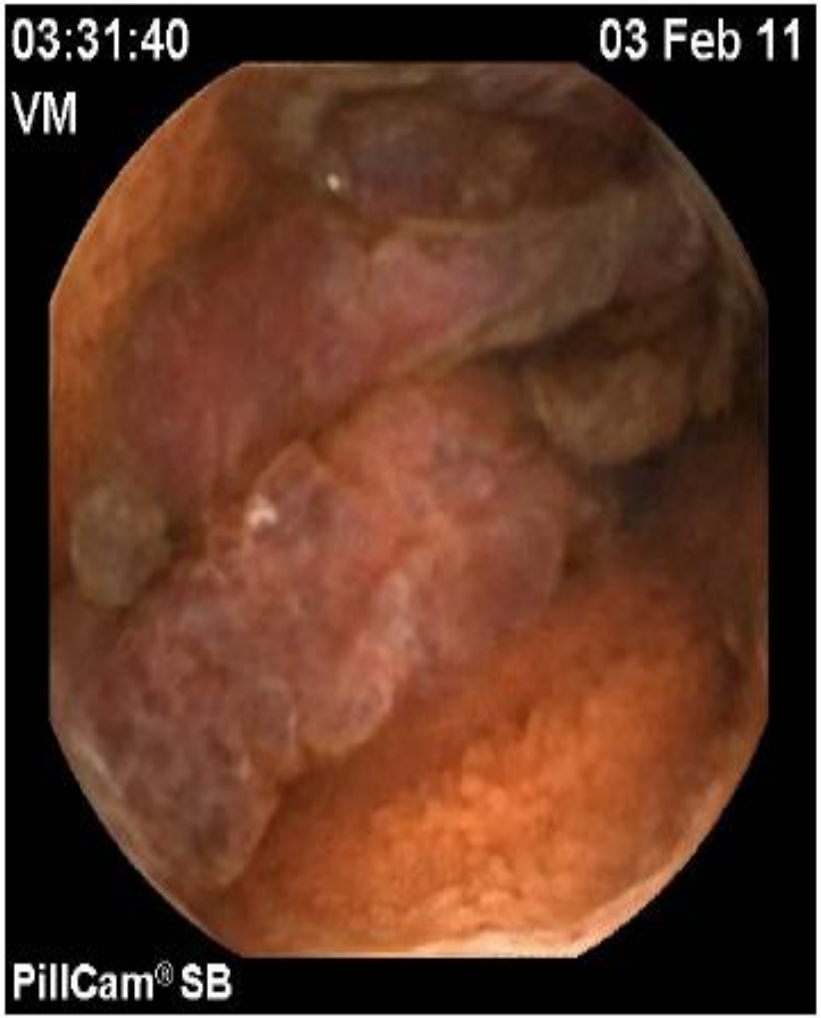
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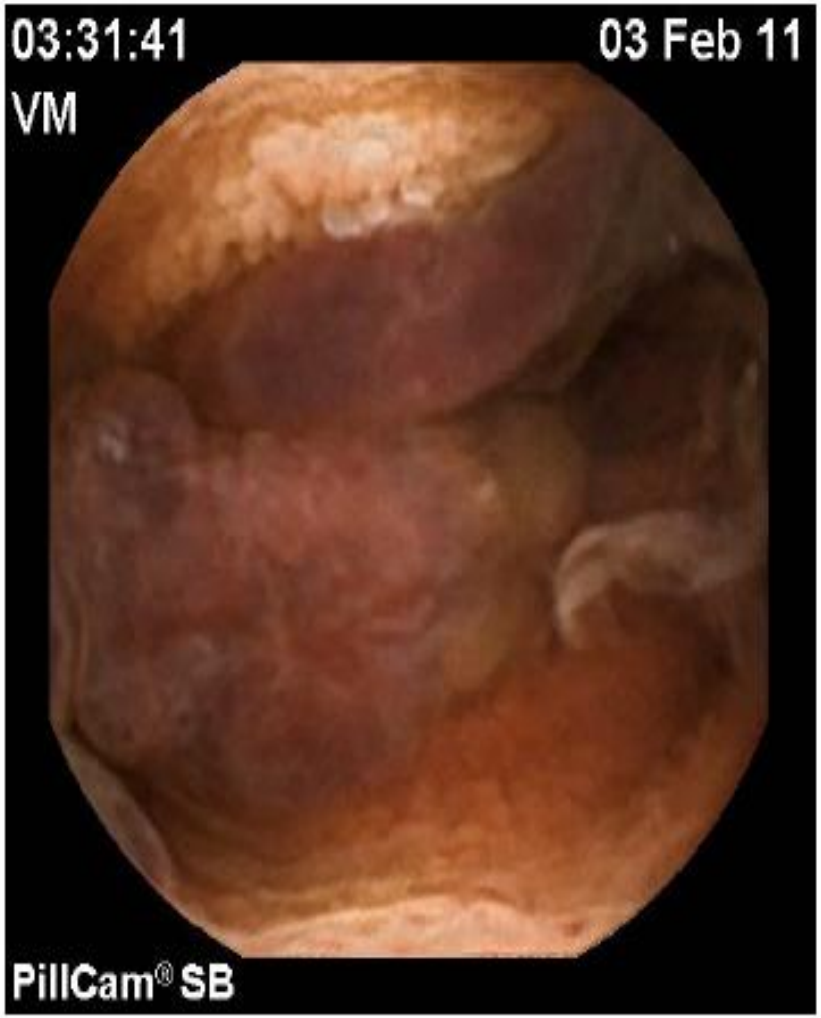
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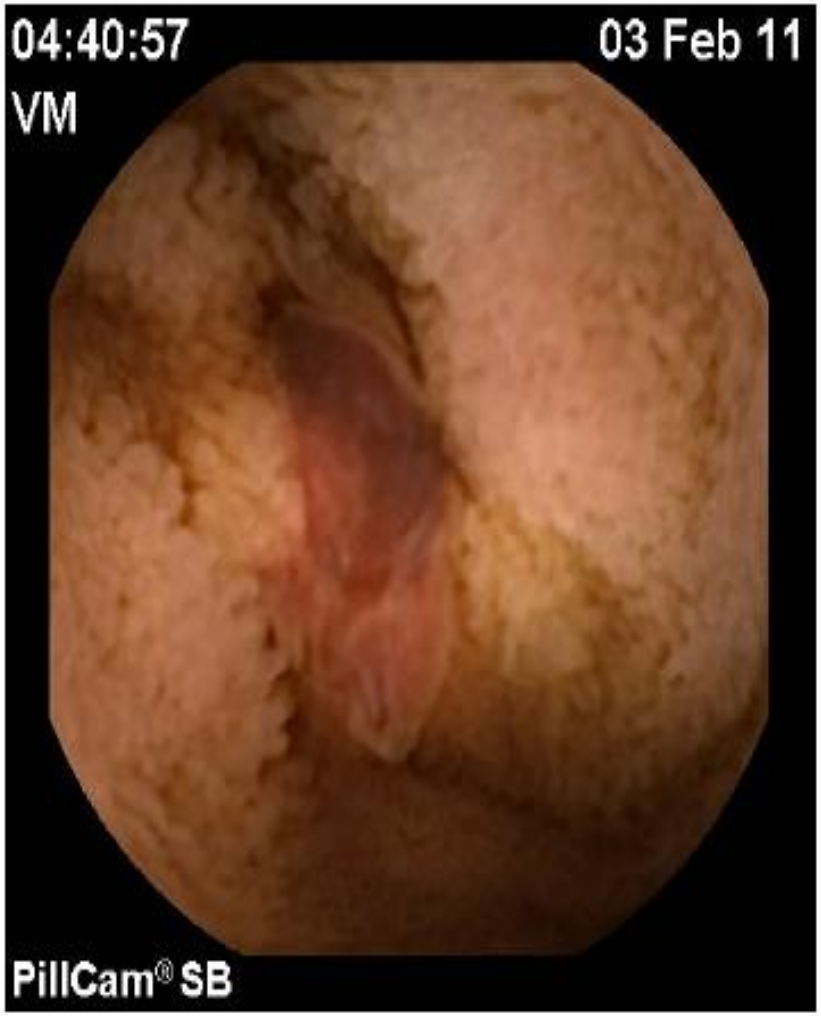
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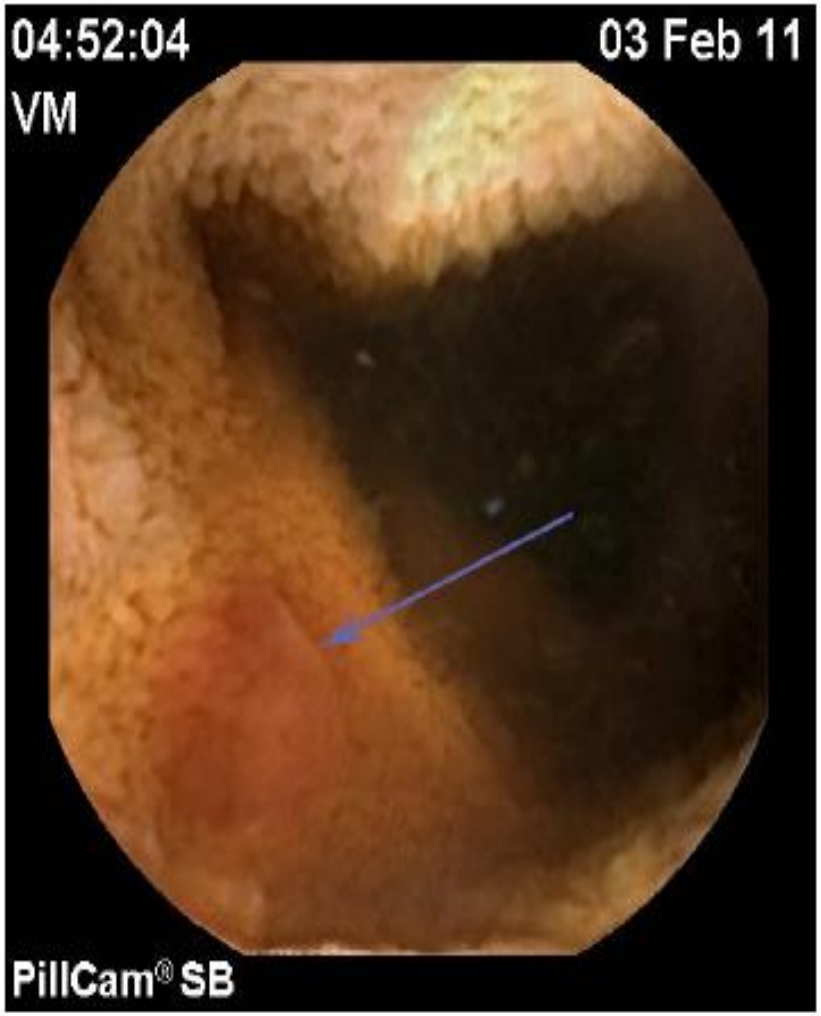
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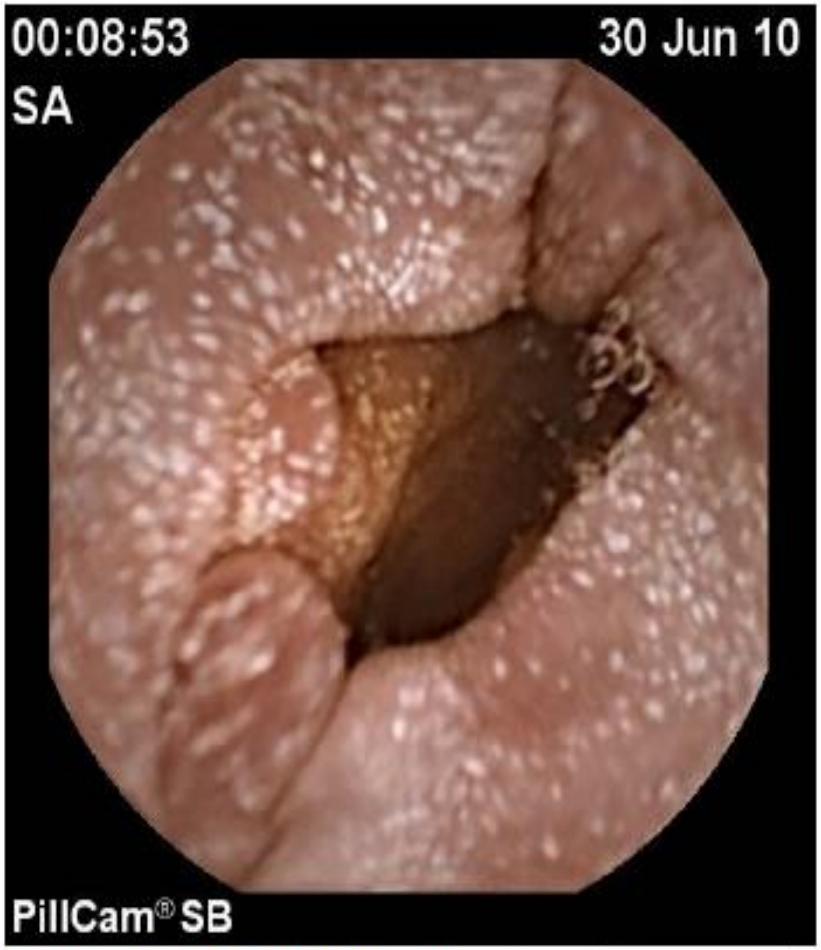
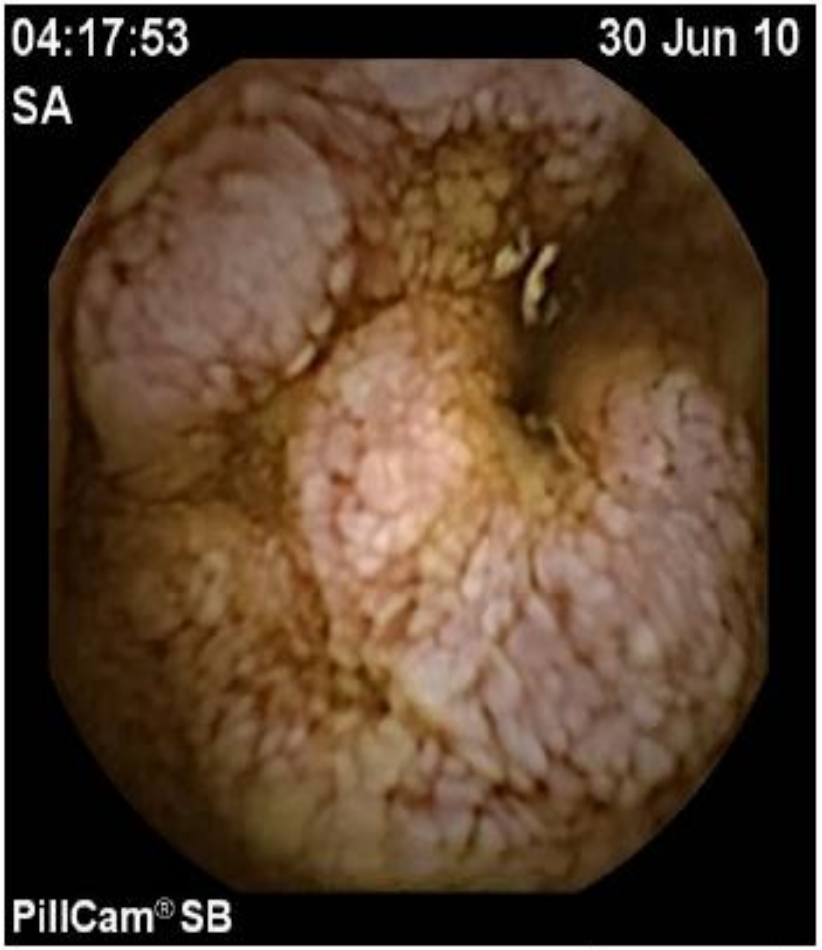
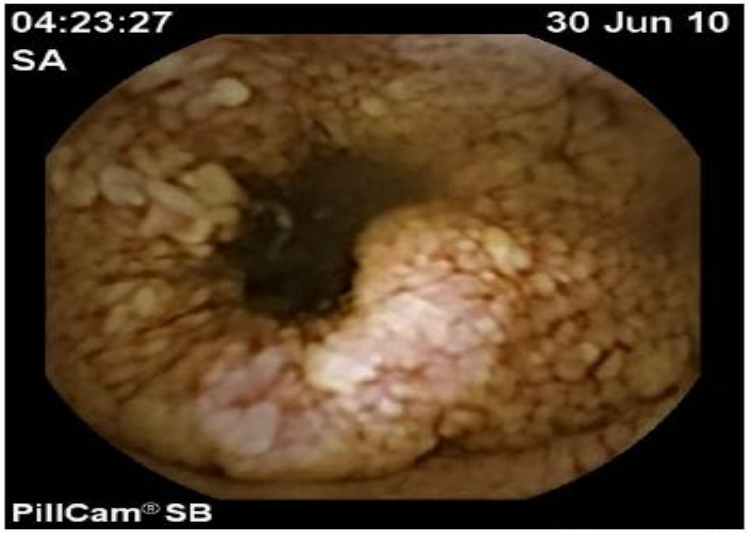




Proximal: Note villous atrophy (or severe edema)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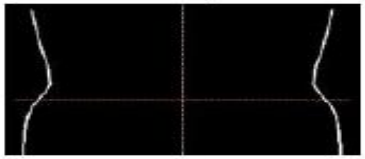

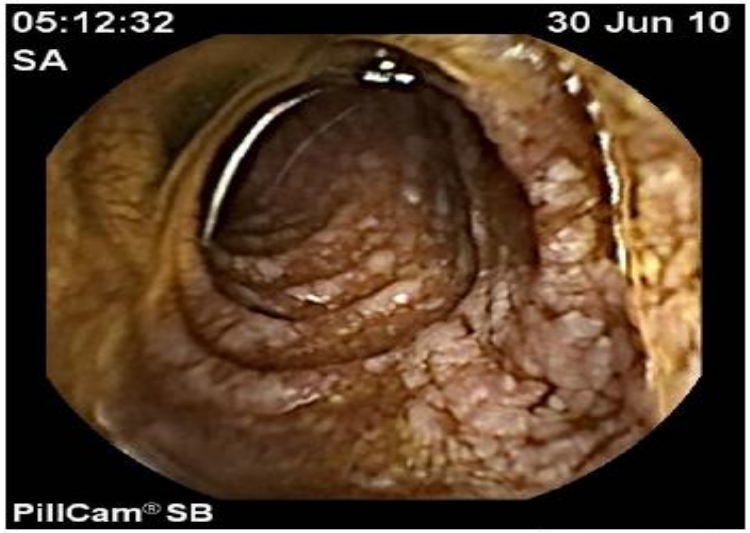
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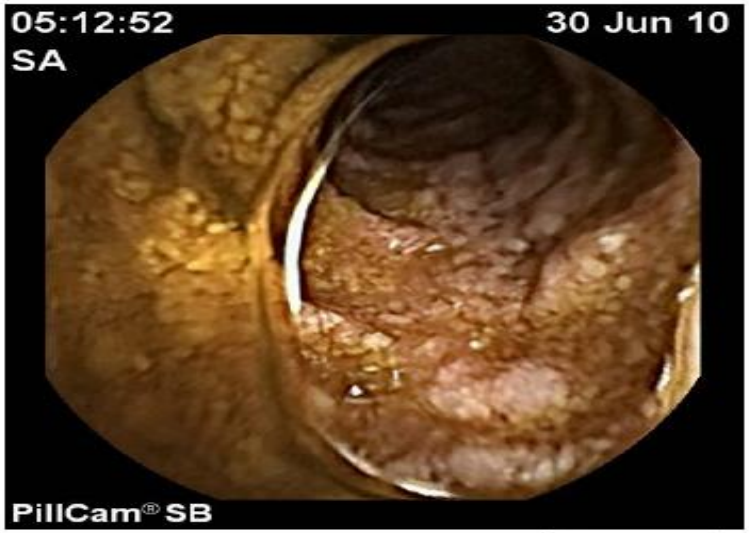
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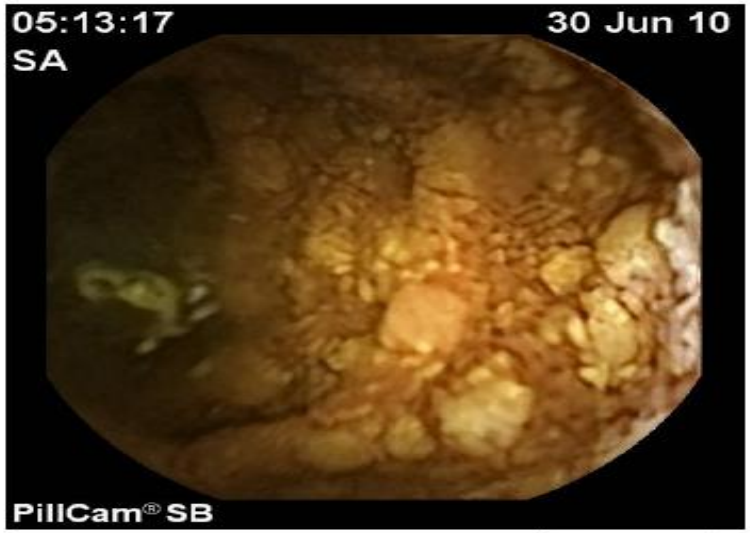
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PillCam® SB

Nuclear medicine: protein loss study at age 8 years

- Demonstrated protein loss in the colon at the level of the hepatic flexure as well as more inferiorly in the ascending colon

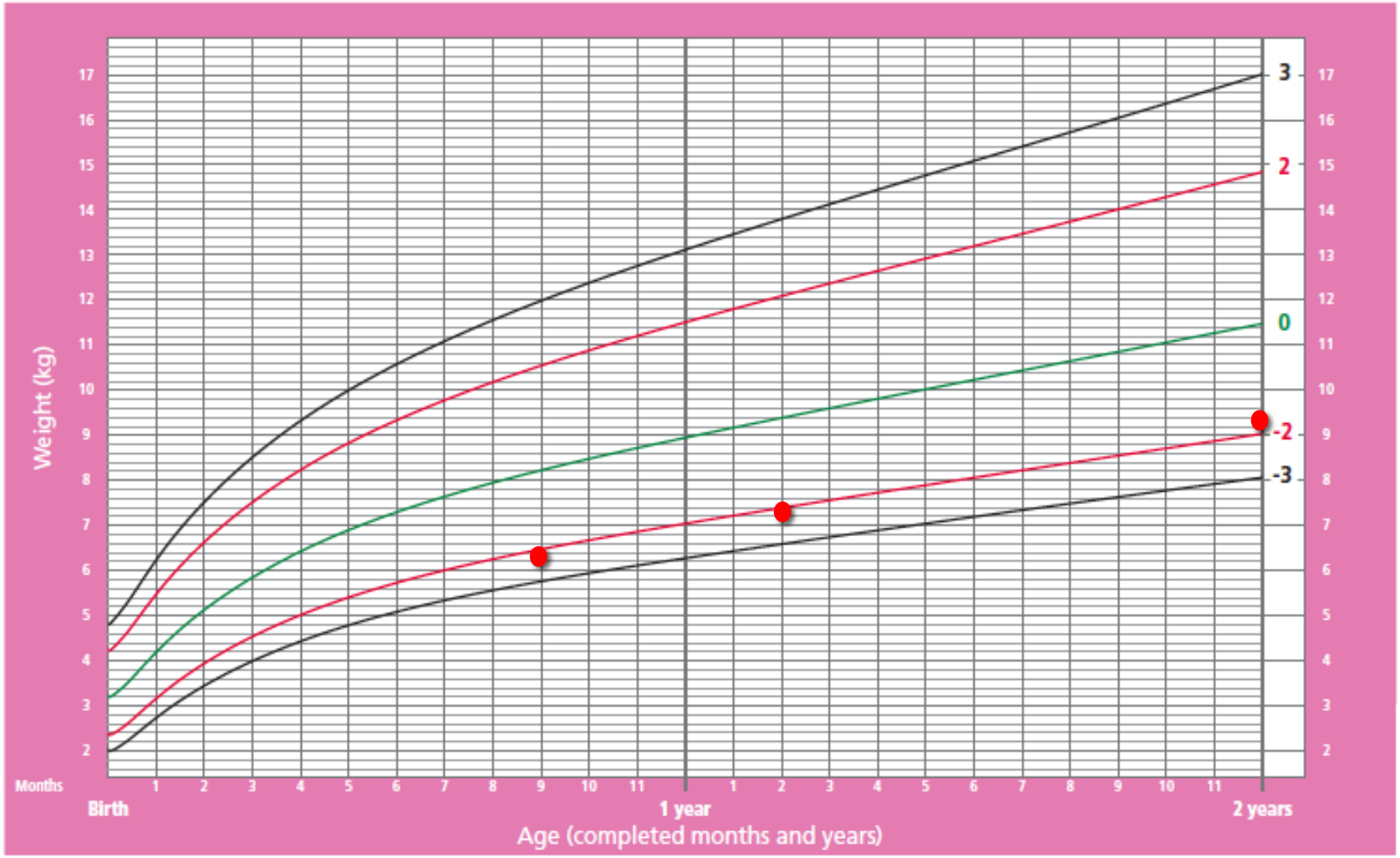
- At this stage she was found to have right ventricular outflow obstruction
- Underwent surgical excision of obstruction and placement of an aortic homograft in a pulmonary valve position
- Complicated by haematemesis and melena post-op secondary to variceal bleed which settled with conservative management
- Marked improvement in protein-losing enteropathy resulted
- Things went well for approximately 1 year after which **diarrhea and edema recurred (age 10 years)**
- Cardiology review demonstrated normal right sided pressures despite some pulmonary valve regurgitation

- Capsule endoscopic study repeated and showed lymphatic stasis as previously seen (varices however had resolved)
- In the following 2 - 3 years Rianke failed to thrive

Weight-for-age GIRLS

Birth to 2 years (z-scores)

World Health Organization

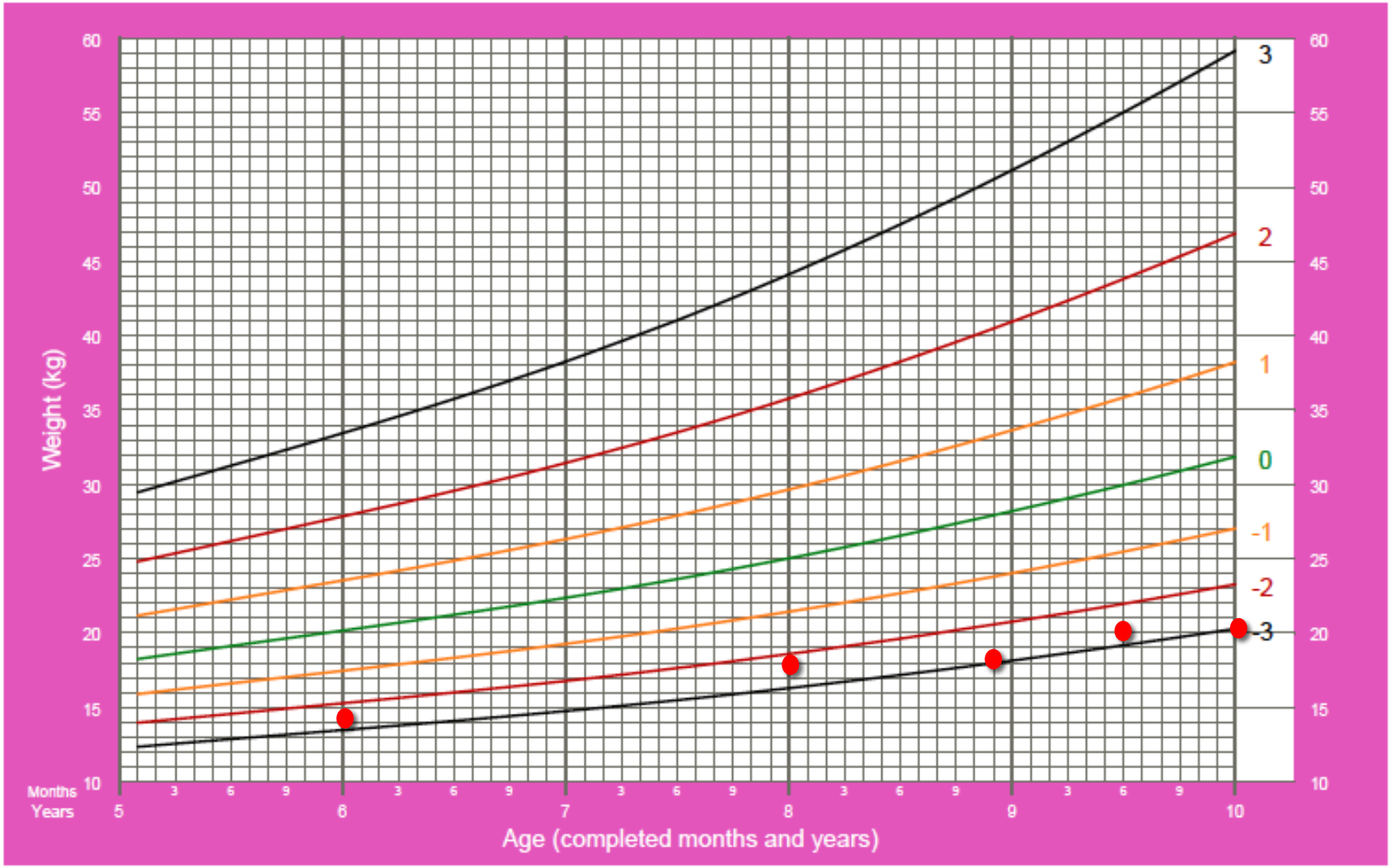


WHO Child Growth Standards

Weight-for-age GIRLS

5 to 10 years (z-scores)

World Health Organization

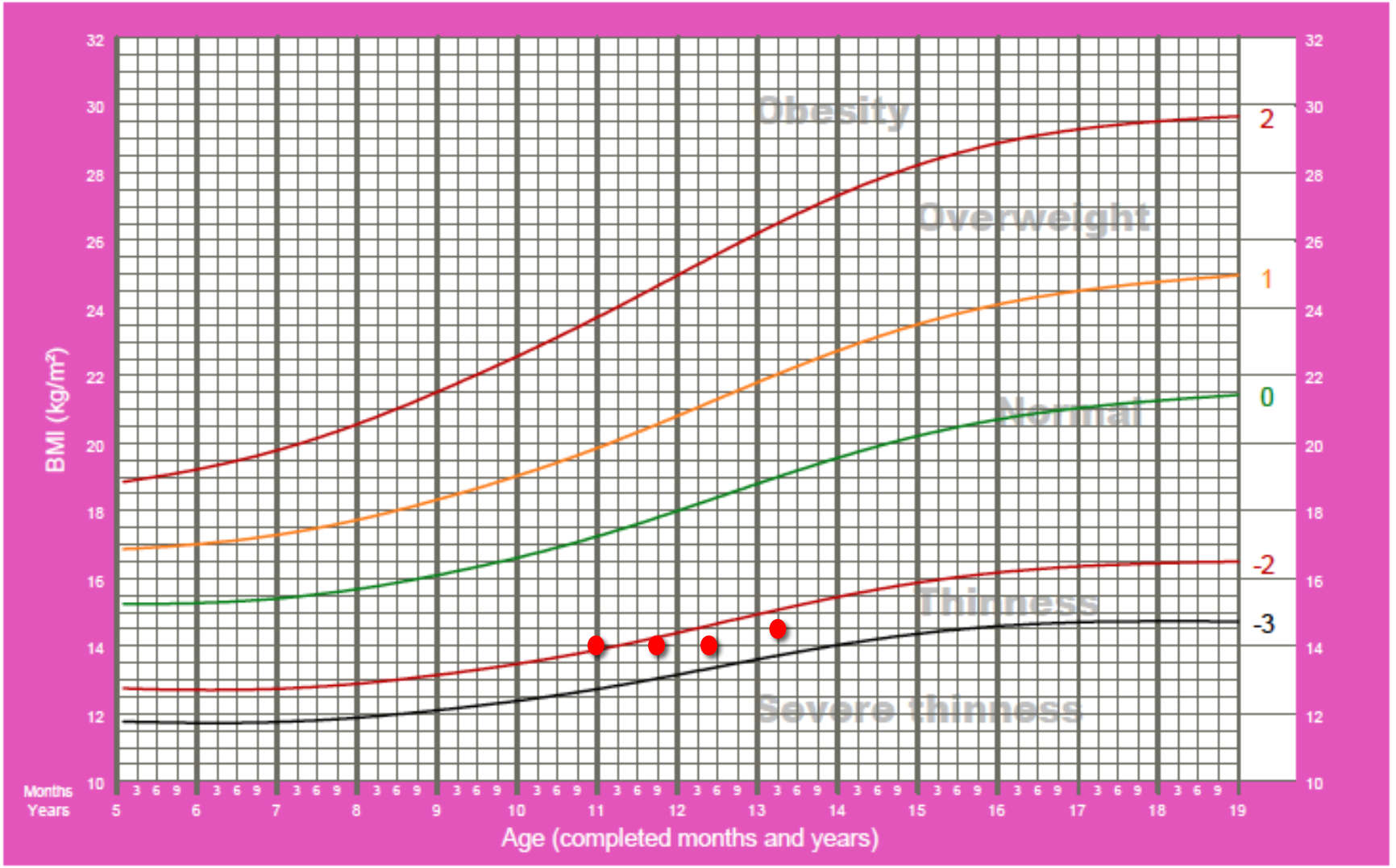


2007 WHO Reference

BMI-for-age GIRLS

5 to 19 years (z-scores)

World Health Organization

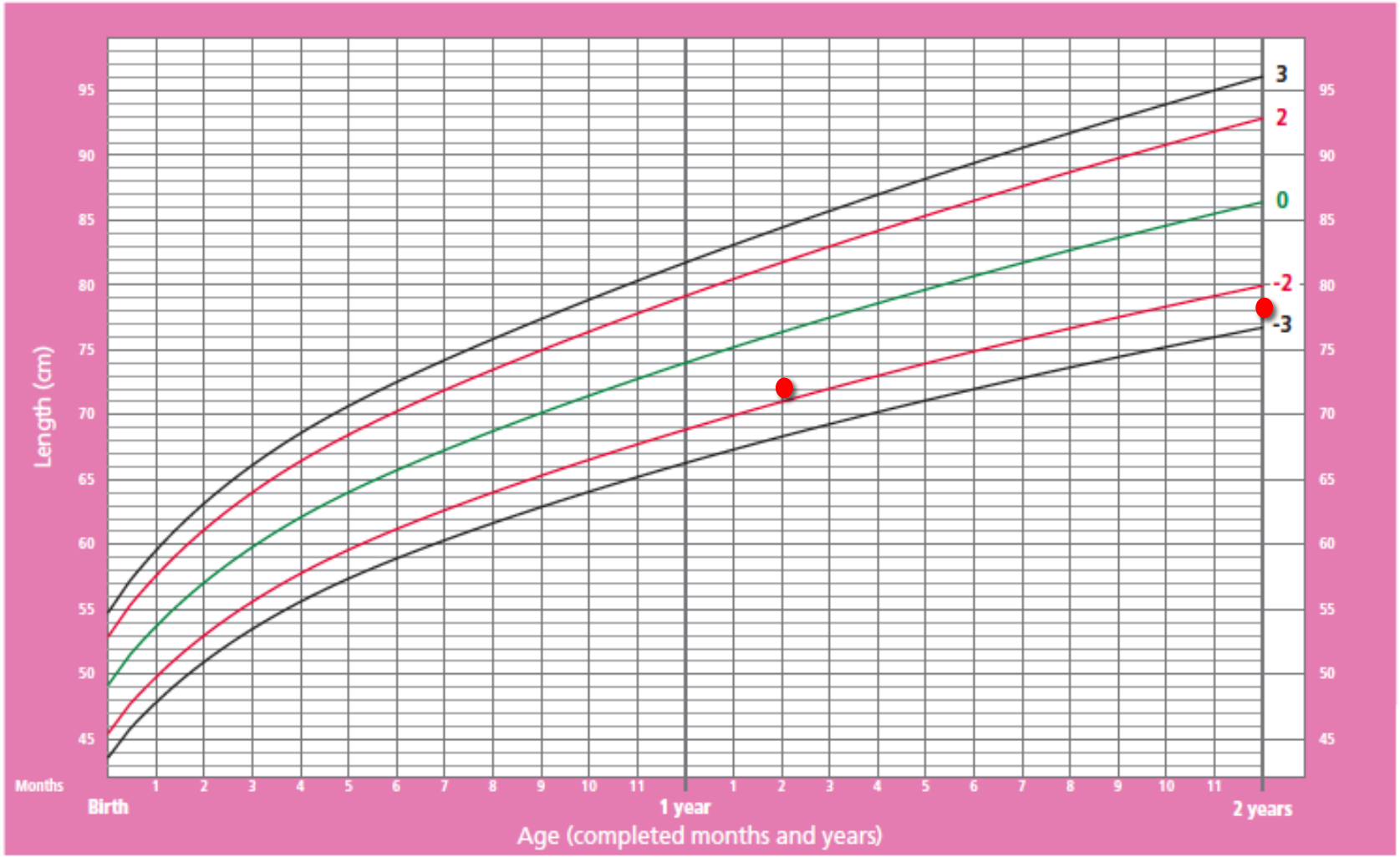


2007 WHO Reference

Length-for-age GIRLS

Birth to 2 years (z-scores)

World Health Organization

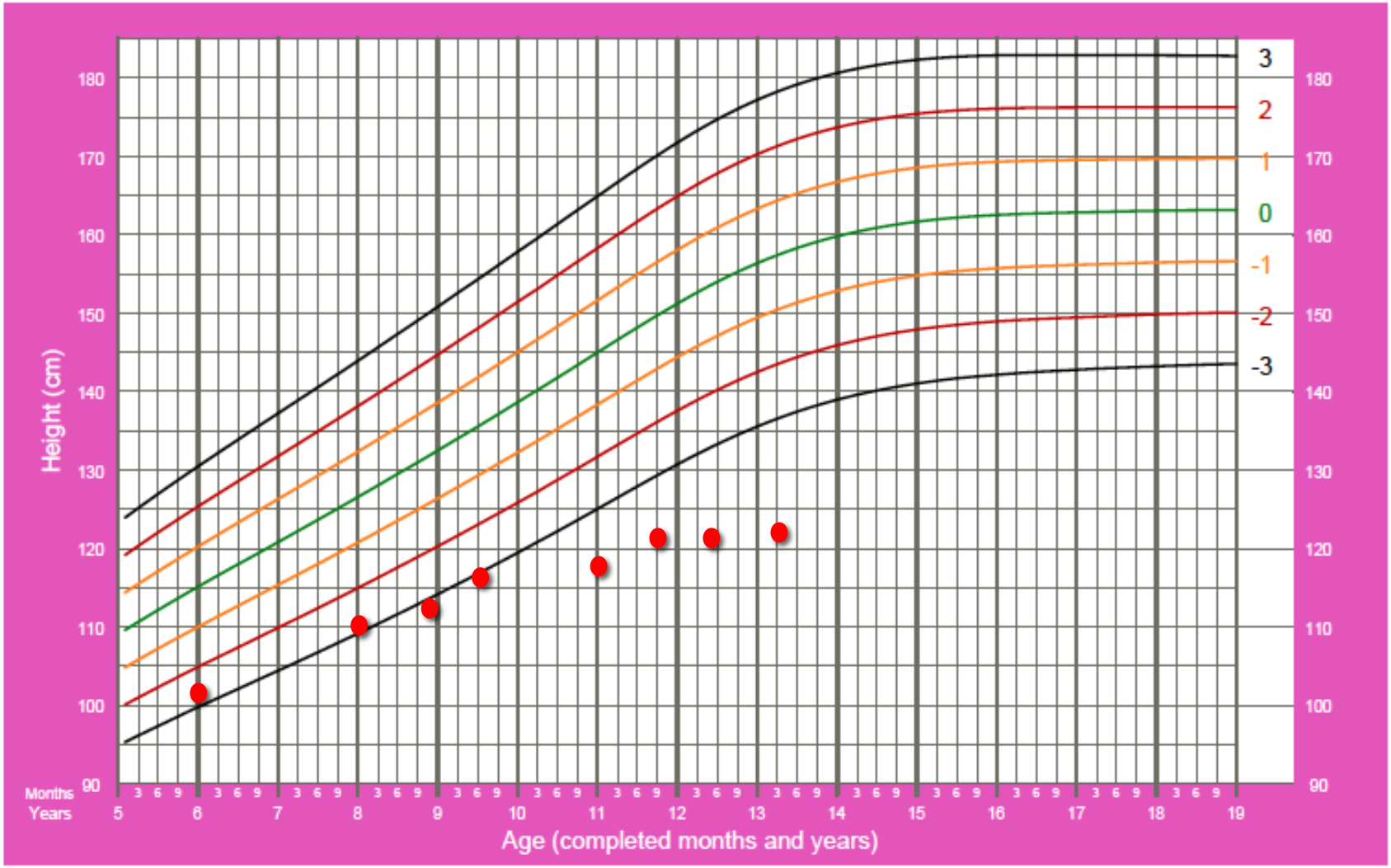


WHO Child Growth Standards

Height-for-age GIRLS

5 to 19 years (z-scores)

World Health Organization



2007 WHO Reference

Age 13 years…..

- Stunted
- Wasted
- Delayed puberty
- Dietary intake: attempting low fat high / protein options, with increased MCT. Cannot tolerate palatability of formula
- Diarrhea, with up to 5 watery stools a day
- **Family:**

Mother: phenotypically Noonan syndrome, PS (surgery) and short stature

Older Brother: phenotypically Noonan syndrome, PS

Father: CP with dyskinesia

Examination

- Noonan phenotype
- Pre-tibial and sacral edema
- Cardiac examination in keeping with pulmonary valve stenosis with pulmonary valve regurgitation
- No CCF
- Abd: distended but soft, without HSM

Investigations

- TP 30 / Alb 17
- IGs ↓
- Hb normal, WCC normal, Lymphs 0.62 (↓), Plts normal
- Stool mcs normal
- CMV PCR negative
- LFTs normal
- S-Mg 0,57 (↓)
- Steatocrit 4
- Alpha-1 antitrypsin 0,61
- Vit D 4,5 (↓)
- Vit A 43,1
- Vit E 7,9 (↓)
- TTG negative (extended spectrum investigations pending)

Investigations

- Bone age: 9 years (below 3rd centile for chronological age)
- Bone mineral density decreased
- FSH 1,6
- LH 0,2
- 17B Oestradiol <75

Secondary hypogonadism

Management

- Commenced on exclusive Monogen® feeds at correct caloric requirement + Liquigen (high energy medium chain emulsion)
- PEG inserted
- Supplements: Vit D, E, B12; Centrum, SlowMag, Spiractin, Calcium

- After 4 weeks:

TP 63 / Alb 40

Mg normalised

Lymphocytes 0,97 (1-4)

Weight 23,5kg (increase of 1kg)

Edema resolved

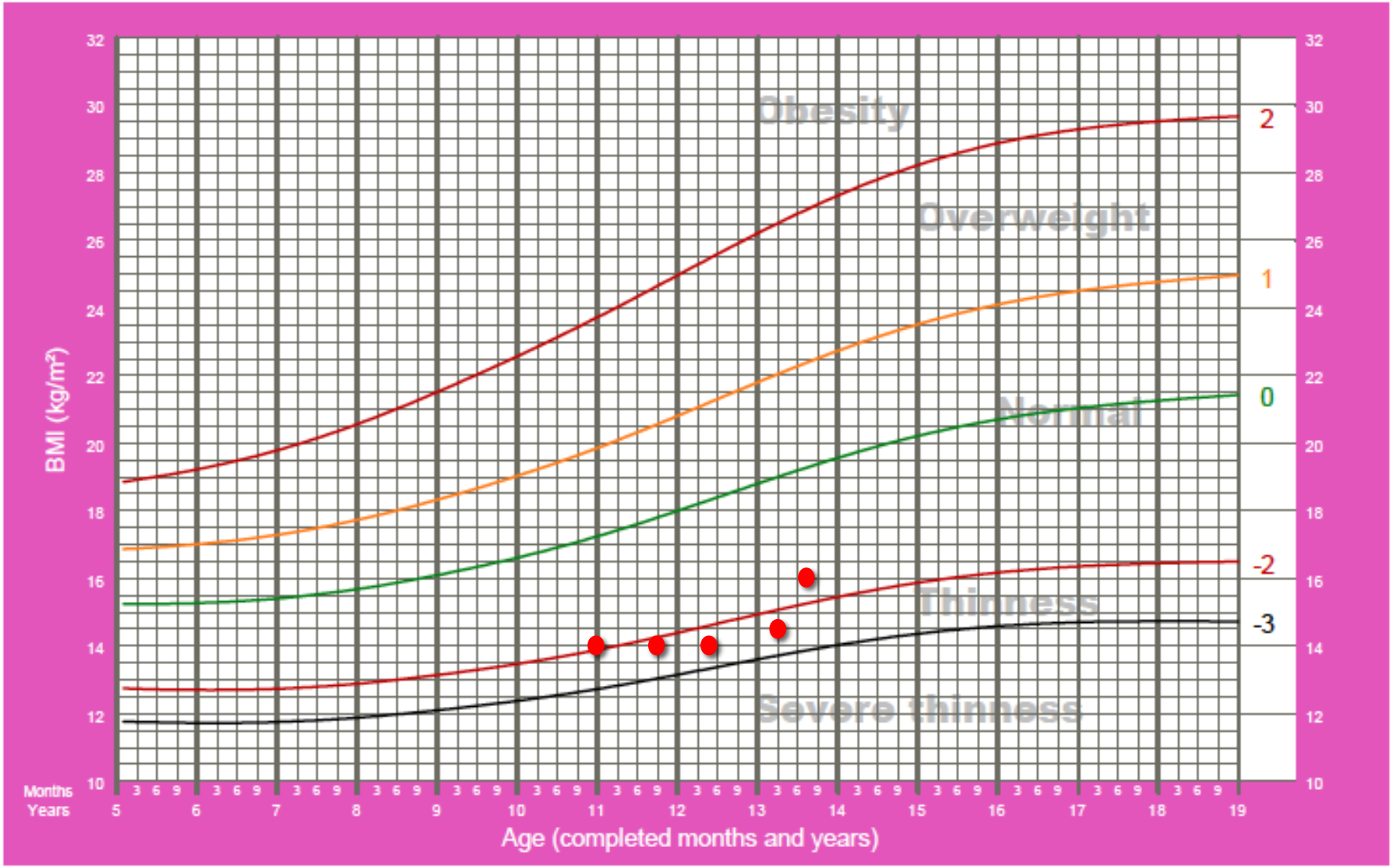
Stools decreased to daily / every other day: normal

- Continued on 8 weeks exclusive Monogen feeding via PEG

BMI-for-age GIRLS

5 to 19 years (z-scores)

World Health Organization

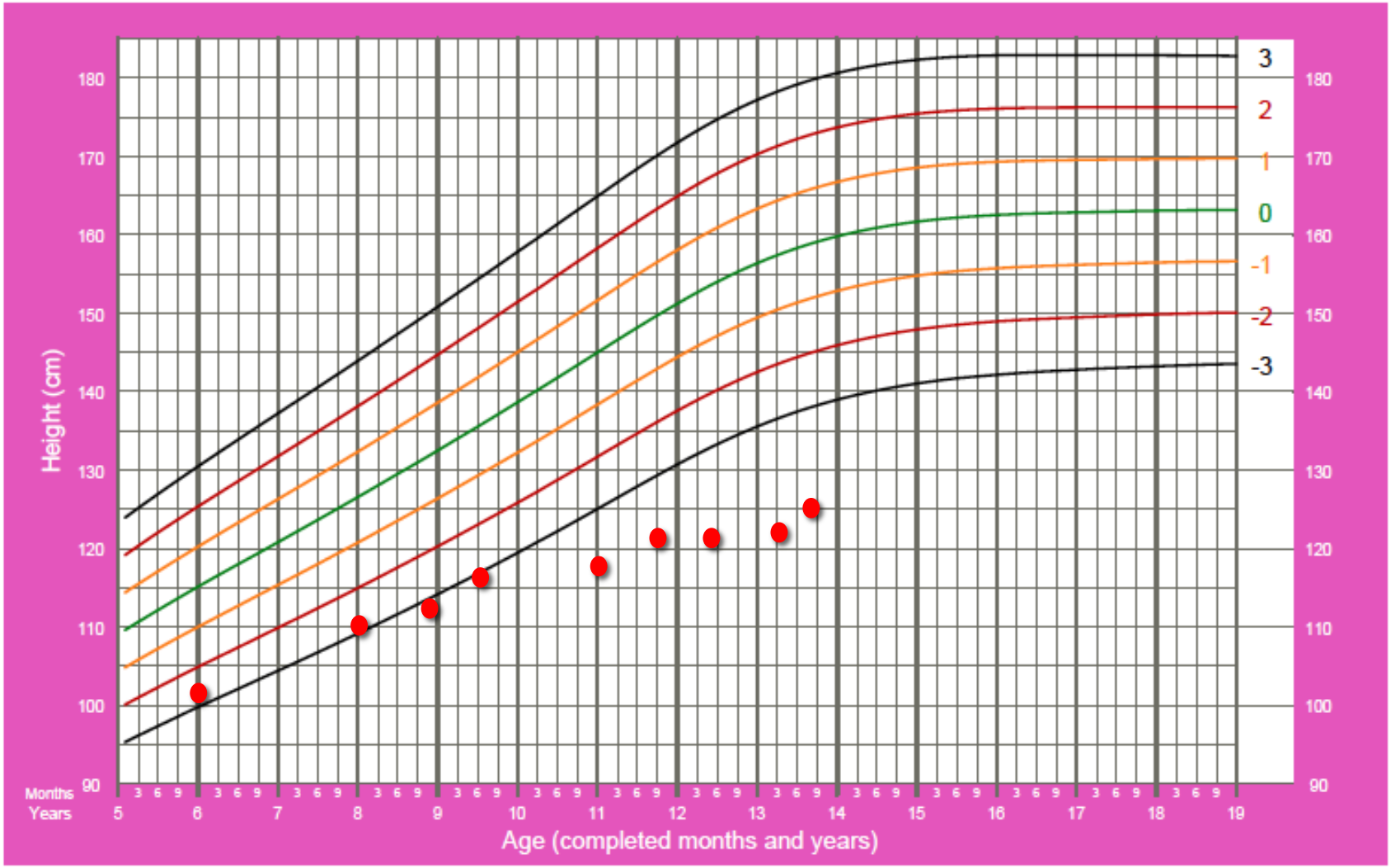


2007 WHO Reference

Height-for-age GIRLS

5 to 19 years (z-scores)

World Health Organization



2007 WHO Reference

- PEG was converted to a Mic-key successfully 2 weeks ago
- Begun with slow re-introduction of oral diet: high protein, fat-free, high MCT

- **Further challenges:**

✧Optimisation of height: Noonan's have 50/50 chance of GH deficiency. GH costly and not covered by medical insurance **+** cardiac risk of HOCM

✧Delayed onset of puberty: risk of estrogen therapy in terms of arresting length growth vs age at 13 years

✧Likely that short stature and delayed puberty are secondary to nutritional state. Allow 6mo of rehabilitation and if does not demonstrate adequate height growth and pubertal development consider investigations for GH +- estrogen therapy