# Primary amenorrhoea with ulcerative colitis

| HOSP # | | WARD | |
|---|---|---|---|
| CONSULTANT | | DOB/AGE | 15 y girl |

## Abnormal Result

This patient was discussed at a combined Endocrinology / Chemical Pathology meeting.

Total bilirubin: 281 umol/L

## Presenting Complaint

The patient was a candidate for a liver transplant, but was referred to the endocrinology department for the short stature and primary amenorrhoea prior to surgery.

## History

She was diagnosed with ulcerative colitis in 2016 (@ 12y age) and primary sclerosing cholangitis. Breast development started in 2018 (@14 years), but no menstrual cycles started ever since.

She has one younger sister which is well currently at 4 y age.

Birth weight was 3.8 kg.

Medication

Patient was receiving steroids and sulfasalazine intermittently.

For portal hypertension she is also receiving furosemide and

spironolactone

Vitamin D supplements are also given

# Examination

Height (114cm) for age: <3rd percentile

Weight 35 kg

Breasts well developed – Tanner IV,

No armpit hair growth, parse pubic hair – Tanner II

# Laboratory Investigations

| Test | Result |
|---|---|
| Total bili (umol/L) | 281 H |
| Conj bili (umol/L) | 246 H |
| ALT (U/L) | 58 H |
| AST (U/L) | 151 H |
| ALP (U/L) | 524 H |
| GGT (U/L) | 65 H |
| TSH mIU/ml | 1,74 |
| Free T4 (pmol/L) | 16,4 |
| Free T3 (pmol/L) | 2,8 L |
| FSH (IU/L) | 8,2 |
| LH (IU/L) | 6,2 |
| E2 (pmol/L | 462 |
| Prog (nmol/L) | 0.9 |
| Prolactin (ug/L) | 15,4 |
| INR | 2.09 |

| | |
|---|---|
| IGF-1 (ug/L) 107.8 – 541.5<br>Tanner stages:<br>Boys Girls<br>Stage I 63 – 271 ug/L 71 – 394 ug/L<br>Stage II 114 – 411 ug/L 122 – 508 ug/L<br>Stage III 166 – 510 ug/L 164 – 545 ug/L<br>Stage IV 170 – 456 ug/L 174 – 480 ug/L<br>Stage V 161 – 384 ug/L 169 – 400 ug/L | **23.5** |

Table 1 – Results

# Other Investigations

## Histology (Colonoscopy)

MICROSCOPIC:
Right, transverse and left colon:
Sections show large bowel type mucosa with maintained crypt architecture with no cryptitis or crypt abscess formation noted. No significant increased intra epithelial lymphocytes or subepithelial collagen deposition is present. The lamina propria shows normal inflammatory cells with no giant cells, granulomas, infective organisms, viral inclusions, epithelial atypia or malignancy identified. Colon mucosa morphologically within normal limits

Rectum:
Sections show large bowel mucosa with preserved crypt architecture and increased chronic inflammation in the lamina propria. Active inflammation is absent. There is no evidence of granulomas, viral inclusions, parasites or dysplasia. Non-specific increase in chronic inflammation in the lamina propria.

The other proposed additional examination is a pubic ultrasound to evaluate the ovaries, fallopian tubes and uterus.

It was also proposed that IGF binding protein 3 be measured, as low levels may yield IGF-1 shorter biologically active.

# Final Diagnosis

Primary amenorrhoea most likely due to a physiological delay. Although the pelvic ultrasound hasn’t been done at the time of writing, the low IGF-1 likely indicates a low growth due to chronic systemic disease – see other possible aetiologies below.

# Take Home Message

Amenorrhea can be a condition resulting from dysfunction of the hypothalamus, pituitary, ovaries, uterus, or vagina.

The most common aetiologies include:

- Gonadal dysgenesis, including Turner syndrome – 43%
- Müllerian agenesis (absence of vagina, sometimes with absence of uterus) – 15%
- Physiological delay of puberty (constitutional delay of puberty, chronic systemic disease, acute illness) – 14%
- Polycystic ovary syndrome (PCOS) – 7%
- Isolated gonadotropin-releasing hormone (GnRH) deficiency – 5% (possible selection bias)
- Transverse vaginal septum – 3%
- Weight loss/anorexia nervosa – 2%
- Hypopituitarism – 2%