

## Self assessment<sup>☆</sup>

### Questions

#### Case 1

Andrew, an 11-year-old boy presents to a Rapid Access Paediatric Clinic with an elevated ferritin.

He had experienced tiredness and exhaustion following exercise for the past 3 months and had presented to his GP. There is no history of haemochromatosis or iron ingestion. He suffers from occasional lower abdominal pain with loose stools. Routine bloods carried out by the GP were normal apart from an elevated ferritin.

There is no pertinent past medical history. His two brothers and mother, and possibly his maternal grandmother, have congenital cataracts.

On examination, everything was normal apart from evidence of cataracts and mild eczematous changes to his skin.

His most recent blood tests reveal a ferritin of 2001.5 ug/L and an iron level of 15.2 umol/L.

Q1. Which of the following is NOT true regarding ferritin? (Choose ONE)

- It is an acute phase reactant
- Reference ranges are lower for females than for males
- It serves to store iron in a non-toxic form in the body
- Low serum ferritin is a very specific test for iron-deficiency anaemia
- Low serum ferritin is a very sensitive test for iron-deficiency anaemia

Q2. Which of the following is a potential cause of an elevated ferritin level? (Choose TWO)

- Leukaemia
- Infection
- Crohn's Disease
- Hookworm infestation
- Coeliac Disease

Q3. Which of the following is NOT a recognised complication of a raised ferritin level? (Choose ONE)

- Testicular atrophy
- Hypothyroidism
- Cardiomyopathy
- Diabetes
- Liver Cirrhosis

Q4. What is the inheritance mechanism and associated gene mutation of hereditary hyperferritinaemia cataract syndrome? (Choose ONE)

- Autosomal Dominant – FTL
- Autosomal Recessive – H63D
- X-linked Dominant – C282Y
- X-linked Recessive – ATP7B

#### Case 2

George is a 5-year-old boy arriving in the emergency department by ambulance with both his parents. He presents with a two-day history of general malaise, with intermittent abdominal pain. He has not been eating or drinking since last night, and has not passed urine since the early hours of this morning. On examination, he is afebrile, his abdomen is soft and mildly tender, and his smile is slightly asymmetrical. He is mildly tachycardic and tachypnoeic. He appears distressed, though he is alert and oriented. His neck is turned to the left and is painful when moved, and he has reduced tone, power and absent reflexes in his right limb. The remainder of the neurological exam is normal. George's height is on the 99th percentile and his weight is on the 66th percentile for his age. He was born at term by normal vaginal delivery, and has no significant past medical history and his immunisations are up to date.

An MRI of George's cervical spine is taken (Figure 1).

Q1. What investigation is most appropriate in this case? (Choose ONE only)

- Serum vitamin B12
- Full body CT scan
- Spinal angiography
- Lumbar Puncture
- Spinal cord biopsy

A lumbar puncture is performed. The procedure is atraumatic and cerebrospinal fluid obtained from this is clear, with the exception of showing an increased white blood cell count (7 WBC/mm<sup>3</sup>).

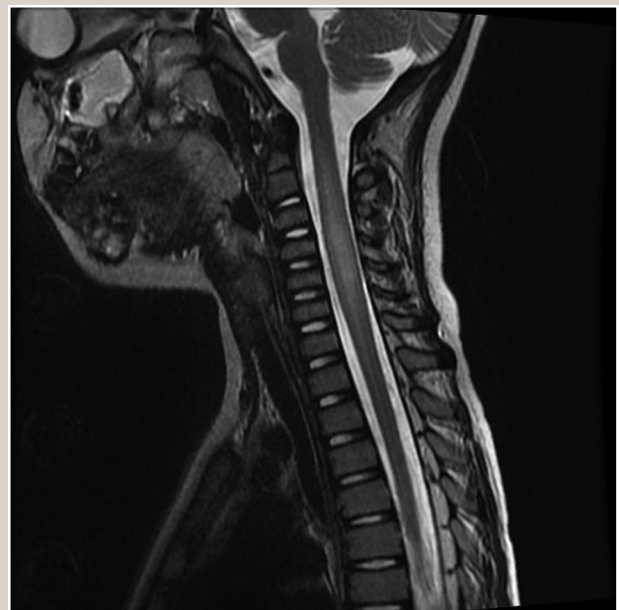


Figure 1 MRI scan of the cervical spine.

<sup>☆</sup> This article contains cases from previous self assessments.

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Q2. Considering the history, and results of the LP, what is the most likely interpretation of the MRI? (Choose ONE only)

- Primary schwannoma neurofibrosarcoma
- Subacute combined degeneration of spinal cord
- Transverse myelitis
- Syringomyelia
- Extradural spinal metastasis
- Anti-MOG associated encephalomyelitis
- Viral meningitis
- Normal appearance of the cervical and thoracic spinal cord

Q3. Which of the following has not been found to cause this condition? (Choose ONE only)

- Vitamin B12 deficiency
- Multiple sclerosis and neuromyelitis optica spectrum disorders
- Viral Meningitis
- Hepatitis B Vaccination
- Paraneoplastic syndromes

Q4. What are the top three management priorities for this patient?

- Vitamin B12 supplements
- Referral to paediatric neurosurgery
- IV antiviral, antibiotic and corticosteroid therapy
- Follow up in outpatient department
- Commence anticoagulant therapy
- Transfer to Paediatric Intensive Care and monitor respiratory function
- Referral to paediatric oncology
- Urinary catheterisation
- Nasogastric tube insertion

### Case 3

Emily, a 15 year old girl presents to paediatric outpatients with her mother. Her mother noticed Emily's left shoulder blade appeared to protrude more than her right. Pain and neurological features are both absent.

Emily has not started her periods yet and only in the past 3–4 months appears to have begun her growth spurt.

On examination Emily appears comfortable at rest. There are no neurocutaneous stigmata. A lateral thoracolumbar curvature towards the left hand side is noted. On performing the Adam's test where the child bends forward, marked discrepancy between protrusion of the ribs is apparent, being more prominent on the left side. There appears to be no discrepancy in her leg length.

Q1. What else should part of the examination? Choose ONE answer.

- Beighton score
- Kellgren–Lawrence grade
- Mirel's score
- DASH (Disabilities of arm, shoulder & hand) Score

Emily is diagnosed to have scoliosis and after consulting a paediatric neurosurgeon further investigations to assess the severity are arranged pending review in the neurosurgical outpatient clinic.

Q2. Which of the following is Cobb's angle measured on? Choose ONE answer.

- X-ray PA Spine
- X-ray AP spine
- X-ray Lateral Spine
- MRI spine

Q3. Skeletal immaturity is a poor prognostic factor for the progression of scoliosis. Which two of the following scales evaluates skeletal growth potential? Choose TWO answers.

- Risser grade
- Ann Arbor Staging
- Cobb scale
- Tanner staging

Q4. Emily and her mother wonder what the further management by the neurosurgical team is going to entail.

1 Surgery is recommended in adolescents with a Cobb's angle greater than what value? Choose ONE answer.

- 20°
- 25°
- 30°
- 45°
- 60°

### Answers

#### Case 1

Answers:

- E
- A and B
- B
- A

In the setting of iron-deficiency anaemia, ferritin is the most specific lab test available. However, it is not a very sensitive test as ferritin levels may be increased by infection or chronic inflammation as ferritin is an acute phase reactant. In their study, Kis et al. showed that using a ferritin value of  $\leq 100$   $\mu\text{g/L}$  had a sensitivity of 64.9% and a specificity of 96.1% for the detection of iron deficiency.

Elevated ferritin can have effects on a plethora of organ systems: it can cause cirrhosis of the liver; it can affect the heart and cause a congestive cardiomyopathy and conduction defects; it can affect the endocrine system and cause diabetes mellitus sometimes requiring insulin and also cause a lowered gonadotrophin secretion and rarely reduced ACTH secretion. However, thyroid function is usually preserved in children with raised ferritin. Pulmonary hypertension, opportunistic infections and arthropathies are also seen more commonly in patients with raised ferritin.

Common causes for an elevated ferritin in a child are malignancy and infection. Ferritin is an acute phase reactant and, as such, levels increase during infection and in haematological malignancies. In haemophagocytic lymphohistiocytosis (HLH), ferritin levels are typically in excess of 1000  $\mu\text{g/L}$ . However, coeliac disease commonly results in reduced ferritin, due to reduced iron absorption. Similarly, inflammatory bowel disease such as Crohn's and Ulcerative Colitis may cause a reduced ferritin level due to malabsorption. Hookworm infestation can lead to blood loss (commonly over 100 ml/day) and thus a low ferritin level.

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