Clinical Case Presentation

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History

• A 29-year-old Icelandic man with a previous history of ulcerative colitis and anemia.
• Presents to his family doctor with a 3-4 week history of tiredness, headache and increasing shortness of breath.
Past Medical History

• Ulcerative Colitis
  – Diagnosed 9 years ago.
  – Treated effectively with mesalazine 1000mg x2.

• Anemia
  – Diagnosed 3 years ago.
    • Hemoglobin 115 g/L [134-171 g/L]
    • MCV 70 fL [80-97 fl]
  – Put on iron supplements, ferrous sulfate 100mg x2.
  – Describes headache if missed intake of iron.
Review of Systems:
• No bloody stool.
• Black stool.
  – Associates with iron supplement.
• Otherwise negative.

Family History:
• Maternal grandfather passed away fifty years ago from leukemia.
• Paternal grandfather has colitis.
• **Social history**
  – Works as car mechanic. Has a wife and two sons.

• **Medications**
  – Mesalazine 1000mg x2/day
  – Ferrous sulfate 100mg x2/day
  – Naproxen 500mg p.r.n.
Examination

- Normal vital signs.
- General appearance: Very pale.
- CV: Mid-systolic ejection murmur on heart auscultation.
- Abdomen: Soft and nontender. No organomegaly.
- Rectal examination: Black stool. Fecal occult blood test positive.

Examination otherwise unremarkable.
<table>
<thead>
<tr>
<th>Test</th>
<th>Result</th>
<th>Reference range</th>
</tr>
</thead>
<tbody>
<tr>
<td>WBC count</td>
<td>7.3 x10⁹/L</td>
<td>4-10 x10⁹/L</td>
</tr>
<tr>
<td>Hemoglobin</td>
<td>57 g/L</td>
<td>135-175 g/L</td>
</tr>
<tr>
<td>Hematocrit</td>
<td>0.175</td>
<td>0.395-0.515</td>
</tr>
<tr>
<td>MCV</td>
<td>66.9 fl</td>
<td>80-96 fl</td>
</tr>
<tr>
<td>MCH</td>
<td>21.8 pg</td>
<td>26-31 pg</td>
</tr>
<tr>
<td>Platelet count</td>
<td>458 10⁹/L</td>
<td>150-400 10⁹/L</td>
</tr>
<tr>
<td>ESR</td>
<td>27 mm/hour</td>
<td>&lt;15 mm/hour</td>
</tr>
<tr>
<td>Reticulocyte count</td>
<td>4.3%</td>
<td>0.610-2.24%</td>
</tr>
<tr>
<td>S-Iron</td>
<td>38.9 µmol/L</td>
<td>10-30 µmol/L</td>
</tr>
<tr>
<td>Ferritin</td>
<td>663 µg/L</td>
<td>15-400 µg/L</td>
</tr>
<tr>
<td>Albumin</td>
<td>41</td>
<td>38-50 g/L</td>
</tr>
<tr>
<td>B-12</td>
<td>281.3 pmol/L</td>
<td>200-800 pmol/L</td>
</tr>
<tr>
<td>Folate</td>
<td>20.11 nmol/L</td>
<td>7.0-39.0 nmol/L</td>
</tr>
</tbody>
</table>
Differential diagnosis?
Diagnostic Testing

• Gastroscopy
  – Mild esophagitis. Duodenal biopsy was normal.

• Colonoscopy
  – Distal proctitis. Biopsy from the ileum was normal.

• CT abdomen
  – No pathology.

• Capsular endoscopy
  – Signs of a small jejunal angiodyplasia. Otherwise normal.
Treatment

- RBC transfusion
  - 6 units of RBCs in 7 days $\rightarrow$ Hb 106 g/L [134-171 g/L]

  After discharge his Hb level kept declining

- In the next 3 months he received 13 units of RBCs and he was also given iron IV (saccharated iron oxide)

  Remained anemic despite the transfusions
Still anemic and the ferritin level was very high!

=> Referral to a hematologist
First visit to the hematologist

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</tr>
</thead>
<tbody>
<tr>
<td><strong>WBC count</strong></td>
<td>6.0 x10⁹/L</td>
<td>4-10.5 x10⁹/L</td>
</tr>
<tr>
<td><strong>RBC count</strong></td>
<td>3.12 10¹²/L</td>
<td>4.30-5.80 10¹²/L</td>
</tr>
<tr>
<td><strong>Hemoglobin</strong></td>
<td>80 g/L</td>
<td>134-171 g/L</td>
</tr>
<tr>
<td><strong>Hematocrit</strong></td>
<td>0.237 l/L</td>
<td>0.39-0.50 l/L</td>
</tr>
<tr>
<td><strong>MCV</strong></td>
<td>76.1 fl</td>
<td>80-97 fl</td>
</tr>
<tr>
<td><strong>MCH</strong></td>
<td>25.6 pg</td>
<td>26.5-33.5 pg</td>
</tr>
<tr>
<td><strong>MCHC</strong></td>
<td>337 g/L</td>
<td>318-358 g/L</td>
</tr>
<tr>
<td><strong>RDW</strong></td>
<td>20.1 %</td>
<td>10.6-13.2 %</td>
</tr>
<tr>
<td><strong>Platelet count</strong></td>
<td>529 10⁹/L</td>
<td>150-400 10⁹/L</td>
</tr>
<tr>
<td><strong>S-Erythropoietin</strong></td>
<td>17.7 U/L</td>
<td>3.3 - 16.6 U/L</td>
</tr>
<tr>
<td><strong>Reticulocytes</strong></td>
<td>59.7x10⁹/L</td>
<td>25 – 140 x 10⁹/L</td>
</tr>
<tr>
<td><strong>S-Iron</strong></td>
<td>41 µmol/L</td>
<td>10-33 µmol/L</td>
</tr>
<tr>
<td><strong>Total iron binding capacity</strong></td>
<td>41 µmol/L</td>
<td>40 - 70 µmol/L</td>
</tr>
<tr>
<td><strong>Ferritin</strong></td>
<td>1863 µg/L</td>
<td>30-400 µg/L</td>
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</tbody>
</table>
Further Testing

• Hemoglobin electrophoresis
  – Normal

• He was tested for three mutations in the HFE gene that is related to hereditary hemochromatosis
  – Negative
Peripheral Blood Smear
Bone Marrow Smear

(Prussian blue stain)
Case summary: A 29-year-old Icelandic man with a previous history of ulcerative colitis. He has microcytic anemia and has iron overload.

What is the most likely diagnosis?
Sideroblastic Anemia

- The presence of ring sideroblasts on bone marrow examination
- The iron deposition in the erythroblast’s mitochondria is in perinuclear distribution
Sideroblastic Anemia

• Acquired
  – More common
  – Known causes (eg, ethanol, copper deficiency, hypothermia, drugs (Isoniazid, Linezolid and Chloramphenicol)).

• Congenital

Our case: Laboratory results 10 years ago showed Hb 138 g/L and MCV 74 fL

=> Congenital form more likely than the acquired form
=> May respond to Pyridoxine (vitamin B6)

Reference: Bottomley SS. Causes of congenital and acquired sideroblastic anemias. UpToDate.
Bottomley SS. Clinical aspects, diagnosis, and treatment of the sideroblastic anemias. UpToDate. [cited 2015 19 May].
Our case

• Pyridoxine 100 mgx1
  – Good response
  – Three months later Hb is 139 g/L without blood transfusions.

• Treatment of iron overload
  – Repeated phlebotomy done during next 18 months
  – 19 units of blood were removed
Learning point

In Northern Europe microcytic anemia is not always caused by iron deficiency!
Takk fyrir!