Ask a Pathologist

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Question: My patient carries a diagnosis of chronic anemia and has been treated for iron-deficiency in the past with minimal to no improvement. How can I differentiate between iron-deficiency anemia and α-thalassemia?

Answer: Although both iron-deficiency anemia (IDA) and thalassemia can present with a microcytic hypochromic anemia on a complete blood count (CBC), a few tricks may push you towards more strongly considering thalassemia. These include:

1. Microcytic anemia with a low total red blood cell (RBC) count suggests IDA, while a normal to high total RBC count suggests thalassemia.
2. Microcytic anemia with normal RBC distribution width (RDW) suggests thalassemia trait.
3. In children, where an mean corpuscular volume (MCV) < 80 fL is more common, the Mentzer index can be useful:
    a. Mentzer index calculation = (MCV/RBC count); ratio > 13 suggests IDA, < 13 suggests thalassemia
    b. Note: if patient has both IDA and thalassemia, Mentzer index likely > 13

Iron studies should be ordered to rule out IDA. In thalassemia, they often show normal serum iron and serum ferritin levels. This is relevant, as iron supplementation plays no role in the treatment of thalassemia and can result in iron overload and secondary hemochromatosis. Alternatively, low iron does not rule out thalassemia and should be treated accordingly.
If still considering a hemoglobinopathy or thalassemia**,** the next step would be to order a **peripheral blood smear with review by a pathologist.** Target cell presence, in conjunction with the CBC findings, will suggest further workup requiring an **order for hemoglobin electrophoresis**. Although hemoglobin electrophoresis cannot diagnose α-thalassemia silent carriers (1 gene affected) or trait (2 genes), it is required to exclude other hemoglobinopathies, β-thalassemias, and, in infants, HbH (3 gene α-thalassemia) or Hb Bart's (4 gene α-thalassemia). If the hemoglobin electrophoresis results indicate normal findings, the work up for α-thalassemia is not complete and requires genetic testing. This typically requires an **order for an α-thalassemia panel** to identify the most common mutations associated with α-thalassemia**.**

References

1. Wahed, Amer and A. Dasgupta. Hemoglobinopathies and Thalassemias. *Hematology and Coagulation.* Elsevier; 2015: 57-74.