

Usefulness of CBC in the Diagnosis of beta Thalassemia Trait (BTT)

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Routine CBC		
WBC		9.38
RBC	H	6.68
HGB		13.0
HCT		40.7
MCV	L	61.0
MCH	L	19.4
MCHC	L	31.9
CHCM	L	30.8
CH		18.6
RDW	H	15.6
HDW		2.88
PLT		228
MPV	L	7.1

Fig.1A

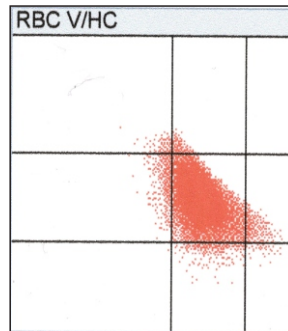


Fig.1B

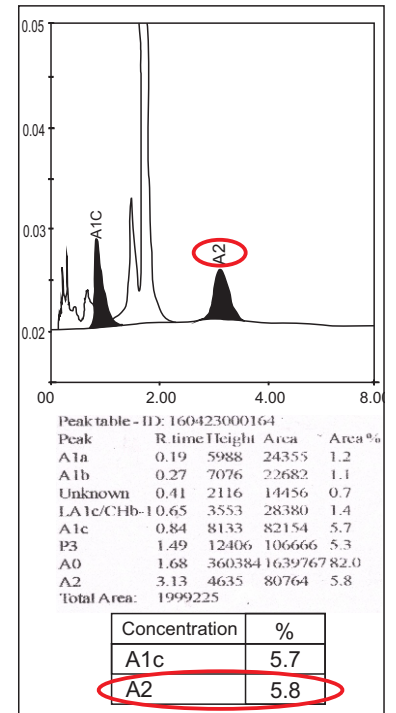
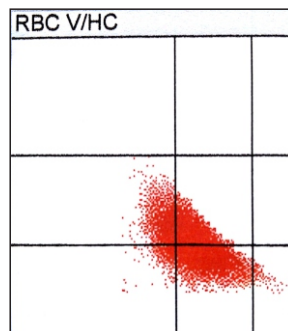


Fig.1C

Fig. 1 (A-C) Patient with BTT. Typical CBC values are seen (A) with a normal RBC graph (top image in B) versus a comma shaped RBC graph of BTT (bottom image in B). The HPLC chromatogram (C) shows an elevated A2

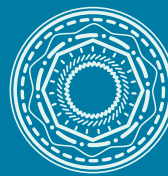
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Beta-thalassemias are a group of autosomal recessive, hereditary blood disorders characterized by abnormalities in the synthesis of the beta chains of hemoglobin. Three forms have been described: thalassemia major, intermedia and minor.

Thalassemia major individuals present in their early life with severe anemia, growth retardation, pallor, jaundice, hepatosplenomegaly, leg ulcers, and skeletal changes. They need regular transfusion therapy which can cause iron overload related complications including endocrine complication.

Thalassemia minor or beta thalassemia trait (BTT) is clinically asymptomatic but some subjects may have mild to moderate anemia.

Though thalassemia minor is clinically innocuous, its diagnosis is crucial because when both parents are carriers, there is a 25% risk at each pregnancy of having children with thalassemia major.

*At a glance*

- Beta-thalassemia trait (BTT) is important to diagnose so that people are aware of the possible genetic risks in children
- A simple CBC can give pointers to possible BTT
- The diagnosis is made by Hb electrophoresis, which shows raised HbA2 levels and confirmed if necessary using genetic analysis.

Diagnosis

This is based on hematologic and molecular genetic testing.

- ① CBC - the basic investigation is an invaluable tool in diagnosing typical cases of BTT (Fig. 1A).

The informative parameters include

- Hemoglobin - 10-12 g/dl
 - RBC count - usually > than 5.2 million/cu mm
 - MCV - 65-80 fl
 - MCH - 20-25pg
 - MCHC - nearly normal
 - Red cell Distribution Width (RDW) - this parameter is useful to distinguish BTT from iron deficiency anemia (IDA), which is the commonest differential diagnosis for a microcytic hypochromic blood picture. This parameter which indicates the degree of anisocytosis, is increased in IDA, whereas it is normal/near normal in BTT.
- ② Mentzer index. This is defined as mean corpuscular volume per red cell count. An index value less than 13 suggests thalassemia trait and more than 13 suggests iron deficiency.
 - ③ Cell counters like Advia provide graphs (Fig. 1B) that is very useful in suggesting the diagnosis.
 - ④ Iron studies and ferritin levels are usually normal and hemoglobin electrophoresis reveals increase of HbA2 levels (4-8%) (Fig. 1C).
 - ⑤ The pitfalls in carrier identification are coexistent iron deficiency, coexistent alpha thalassemia, delta thalassemia, and silent carriers.
 - ⑥ Confirmation of diagnosis is done by molecular genetic analysis.

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Owner, Printer & Publisher: Dr. Bhavin Jankharia

Published at: Dr. Jankharia's Imaging Centre

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