Letter to the Editor

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Did the Proband Have Thalassemia Intermedia or Severe Thalassemia Trait?

Ağır Talasemi Taşıyıcılığı mı? Talasemi İntermedia mı?

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To the Editor,

I enjoyed reading Bilgen et al.'s case report, *The effect of HBB: (c*+96T>C (3'UTR+1570 T>C) on the mild β-thalassemia intermedia pheonotype*, in the recent issue of the Journal (2011; 28: 219-222). I congratulate the authors for exploring at the molecular level at least one of the thalassemia minima that fits well with the present clinical thalassemia nomenclature. Based on their clinical description, I would not consider the proband as thalassemia intermedia, as until 20 years of age he did not require blood transfusions [1]. The mother had thalassemia trait, the father had thalassemia minima, and the proband clinically had severe thalassemia trait, as he had mild hepatosplenomegaly and was compound heterozygote (thalassemia trait[+] and carrier of HBB: C*+9,6 T>C mutation).

I would like to suggest the following arbitrarily modified nomenclature for clinical thalassemia syndromes:

Thalassemia major (according to transfusion requirement):

a) Severe: transfusion interval less than 1 month;

b) Moderate: transfusion interval more than 1 month.

Thalassemia intermedia (according to transfusion requirement):

a) Moderate: transfusion interval less than 1 year;

b) Mild: transfusion interval more than 1 year.

Thalassemia trait:

a) Severe: with mild hepatosplenomegaly;

b) Mild: without hepatosplenomegaly;

Thalassemia minima: absence of clinical and hematological findings.

Sincerely

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Conflict of Interest Statement

The authors of this paper have no conflicts of interest, including specific financial interests, relationships, and/or affiliations relevant to the subject matter or materials included.

References

1. Bilgen T, Canatan D, Arıkan Y, Yeşilipek A, Keser İ. The effect of HBB: c.*+96T>C (3'UTR +1570 T>C) on the mild b-thalassemia intermedia phenotype. 2011; 28: 219-222
2. Özsoylu Ş: Thalassemia trait as thalassemia intermedia. Am J Hematol 2001; 67: 218 (letter)

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Reply

First of all we thank Professor Dr. Özsoylu for his contribution and interest in our report. Intermedia is a beta thalassemia group characterized by severity varying from thalassemia major to asymptomatic carriers. It seems that according to clinical classification it is possible to describe our patient as severe thalassemia trait rather than mild thalassemia intermedia. On the other hand, the primary reason we consider our patient as thalassemia intermedia is that he is compound heterozygous for 3'UTR+1570 T>C and Cod 8(-AA) beta-globin gene mutations. We think mild thalassemia intermedia is the appropriate description of our patient, considering that he has two different mutations on his 2 different copies of the beta globin gene, as well as mild phenotypic findings. We think that the description of severe thalassemia trait is more suitable for patients with only 1 beta globin gene mutation with clinical findings in addition to the thalassemia minor phenotype.

Again, we appreciate your letter, as it afforded us the opportunity to explain in greater detail our rationale for the use of genetic data in addition to clinical criteria in the classification of thalassemias.

Sincerely,

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