Shared molecular basis, diagnosis, and co-inheritance of alpha and beta thalassemia

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Shared molecular basis, diagnosis, and co-inheritance of alpha and beta thalassemia

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TO THE EDITOR: We have read the article entitled “Molecular basis and diagnosis of thalassemia” by Lee et al. that was recently published in Blood Research [1]. We want to express our congratulations to the author for successful publication, and we would also like to provide some input.

The authors described the molecular underpinnings of alpha and beta thalassemia separately in this article. Every facet of the illnesses was addressed, including diagnostic criteria for both, yet there was no mention of their common molecular basis or co-inheritance.

We would like to emphasize this issue by pointing out that other published manuscripts have reported on both types of thalassemia. In a study in Pakistan, in order to establish the prevalence and investigate the spectra of alpha thalassemia gene deletions in patients with beta thalassemia, Shahid et al. concluded that alpha thalassemia coexists with beta thalassemia major [2]. This study led to the significant finding that alpha thalassemia deletions (–alpha 3.7, –alpha 4.2) are also the common co-inherited deletions found in beta thalassemia major; however, this link has not been thoroughly researched. Guvenc et al. investigated the association of alpha and beta-thalassemia genotypes using the reverse hybridization technique, and demonstrated that the alpha thalassemia mutation is co-inherited with sickle cell anemia. These authors also concluded that interactions between the alpha and beta globin chains may produce moderate to severe phenotypes depending on the molecular defects involved [3]. Both types of thalassemia occur at such high frequencies that it is not uncommon for individuals to inherit the alpha thalassemia trait from one parent and the beta thalassemia trait from the other [4]. In addition, alpha and beta globin genes are inherited on two different chromosomes (the alpha globin gene is on chromosome 16 and the beta globin gene is on chromosome 11). The diagnosis of individuals with both alpha and beta thalassemia is not routinely performed for the individual types, and the simultaneous presence of alpha and beta thalassemia does not appear to interfere with ascertaining beta thalassemia carrier state in routine hematological screening tests.
[4]. Although restriction endonuclease DNA mapping can detect contemporaneous inheritance of both types of thalassemia, definitive screening criteria are still needed.

The co-inheritance of alpha and beta thalassemia is quite rare; according to Li et al., in the Chinese population, the chances of finding an individual with co-inheritance of both is 1:1000 [5]. The co-occurrence of both alpha and beta thalassemia reduces the severity of the disease; therefore, these patients present with only mild to moderate symptoms of anemia and are less likely to be found in a hospital-based population [6].

**Conclusion**

These findings imply that a diagnostic criterion for screening patients with concurrent alpha and beta thalassemia should be developed. Furthermore, prenatal testing and genetic counseling should be undertaken in regions where alpha and beta thalassemia are more common in order to build a controlled and preventative environment that could lead to a lower incidence of co-inheritance of the two diseases.

**Authors’ Disclosures of Potential Conflicts of Interest**

No potential conflicts of interest relevant to this article were reported.
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