Letter to Editor

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Is There a Substantive Association between Turner Syndrome and Beta Thalassemia Major?

Hossein Neamatzadeh[1], Seyed Alireza Dastgheib[2], Reza Bahrami[3*]

[1] Mother and Newborn Health Research Center, Shahid Sadoughi University of Medical Sciences, Yazd, Iran
[2] Department of Medical Genetics, School of Medicine, Shiraz University of Medical Sciences, Shiraz, Iran
[3] Neonatal Research Center, Shiraz University of Medical Sciences, Shiraz, Iran

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Corresponding author:
Reza Bahrami

Email:
r.bahrami.neo@gmail.com

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Dear Editor,

The case report by Mirhosseini et al. explores the noteworthy association between Turner syndrome and β-thalassemia major, emphasizing the clinical manifestations and the potential genetic factors that may clarify the relationship between these two rare conditions. While the findings warrant further consideration, a more detailed evaluation of the evidence and its implications is essential.

The authors examine rare comorbidity between Turner syndrome and β-thalassemia major, provide insights into their clinical characteristics and explore a potential genetic mutation in a transcription factor gene that could explain this association. Genetic testing is proposed to confirm this hypothesis. A particular case study illustrates the clinical features associated with Turner syndrome, such as short stature, triangular facial structure, low-set ears, webbed neck, and lordosis, alongside the diagnosis of β-thalassemia major. Notably, karyotype analysis revealed a 45, XO chromosomal configuration, typical of Turner syndrome, which underscores the chromosomal abnormalities that may interlink these disorders. The simultaneous presence of both conditions necessitates meticulous clinical management, focusing on growth and hormone development, and emphasizes the importance of individualized treatment protocols. The rarity of this association underscores the urgent need for further research to elucidate the genetic mechanisms and clinical implications involved.

The association reported in the Mirhosseini et al. study not only opens up intriguing research potentials regarding genetic and rare syndromic conditions but also sparks the necessity for a critical evaluation of the evidence presented in the paper. The indication of a possible genetic mutation as a contributing factor in this association is particularly compelling. Reference to a transcription factor gene suggests a complex genetic interplay that may underlie both conditions. Therefore, pursuing genetic testing to substantiate this potential link is crucial. Such inquiry could provide significant validation for the hypothesis and potentially reveal shared pathogenic pathways. However, until such testing is performed across more considerable and more diverse populations, the association remains largely speculative.

The study's foundation relies extensively on a single patient case report diagnosing both Turner syndrome and β-thalassemia major. While case reports can play a vital role in identifying novel associations, they cannot inherently establish causation or offer a generalized pattern. A solitary case may reveal an intriguing possibility but fails to furnish robust evidence applicable to a more extensive population. Consequently, while the case presented is captivating and serves as an initial prompt for further investigation, it cannot be definitive proof of a causal relationship between these two conditions.

Furthermore, the noted rarity of this association creates a puzzling situation. The uniqueness of this association engages interest and could fuel further inquiry. On the other hand, such limited instances raise fundamental questions about clinical relevance. If these cases are indeed infrequent, the implications of the findings may not hold for a broader population affected by these syndromes. Thus, a comprehensive investigation is warranted to determine whether this association is sufficiently widespread to demand attention in clinical practice or patient management.

In response to these considerations, the authors' appeal for further research to explore the underlying genetic mechanisms is prudent and essential. Additional studies, particularly those encompassing larger and more diverse cohorts, will provide insights not only into the prevalence of this association but also into the biological interplay that links these two conditions. Such exploration could reveal extra layers of complexity that could inform the understanding and management of individuals affected by Turner syndrome and β-thalassemia major.

Further research is needed to explore the potential link between Turner syndrome and β-thalassemia major, as initial findings from a single case report highlight this necessity. The term "association" refers to a relationship observed between two conditions in a patient, suggesting the need for deeper investigation. In contrast, "co-occurrences" indicate the presence of both conditions without implying causality, while "correlation" means a statistical relationship that warrants further exploration of underlying mechanisms. "Coincidence," on the other hand, signifies events occurring together by chance without any causal implications. Given the rarity of these occurrences and reliance on a sole case, the overall prevalence of this association remains unclear. Thus, additional studies are essential to determine the clinical significance and regularity of such cases, while approaching with appropriate caution in interpretation.

In summary, while the reported association between Turner syndrome and β-thalassemia major by Mirhosseini et al. presents a noteworthy finding that beckons further inquiry, it is imperative to scrutinize it through a critical lens 1. The dependence on a singular case report, the infrequent occurrence of the association, and the pressing need for extensive genetic investigations highlight the importance of exercising caution when interpreting these findings. Until comprehensive studies can either validate the connection or clarify the genetic mechanisms involved, these findings should be approached as tentative interpretations—acknowledging

their novelty but recognizing their limitations. Such a measured approach will better inform future research endeavors and clinical practices, ultimately enhancing the understanding and management of these complex conditions.

Conflict of Interest

The authors declare no conflicts of interest.

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Ethical Considerations

None.

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