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Prevalence of β-thalassemia in anemic children referred to City Medical Complex in Kabul City in 1401

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ABSTRACT

Introduction: β-thalassemia is caused by a defect in the synthesis of the β-chain of the hemoglobin molecule. Depending on the extent of the genetic aberration, there are several classifications of β-thalassemia, namely β-thalassemia minor, intermediate, and major. Among these, β-thalassemia major represents the most severe manifestation of this disorder.

Materials and Methods: This descriptive investigation was carried out in a cross-sectional manner. The data pertaining to the research were obtained from the archived records of anemic children who were referred to City Medical Complex Hospital in Kabul city for an HB-Electrophorisis test during the initial six months of 1401. The analysis of the data was performed utilizing descriptive statistics and the software SPSS version 22.

Results: The data presented in the study revealed that out of the total sample size of 216 children diagnosed with thalassemias, 37 individuals were identified as having β-thalassemias, accounting for approximately 17.6% of the cases. Further analysis of the β-thalassemia subgroup indicated that the majority of cases (66.7%) were classified as β-thalassemia major.

Conclusion: The study found that 17.6% of children with anemia were diagnosed with β-thalassemia, with the majority being thalassemia major. The prevalence is highest among children under 6, with symptoms onset around 6 months and survival until 10. β-thalassemia minor has the highest occurrence, possibly due to its hereditary nature passed down from parents. Traditional family marriages in society often lack awareness and knowledge about this condition, affecting the prevalence of β-thalassemia.

Keywords: Thalassemia, β -halassemia, Alpha thalassemia, Hemoglobin.

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1. Introduction

In 1925, a pediatrician named Thomas Colley introduced the term thalassemia. Colley's observations focused on children primarily belonging to Italian families or immigrants from the Mediterranean countries nearby. These children exhibited severe anemia, enlarged spleens, and deformities in their facial and skull bones (1). These children exhibited severe anemia, enlarged spleens, and deformities in their facial and skull bones. Consequently, Colley named this condition thalassemia. The term itself is derived from two components: "thalasa," meaning sea, and "omiya," meaning blood. Hence, thalassemia denotes a blood disease that originates from regions surrounding the sea (2). Thalassemia is a genetic disorder characterized by a reduction or absence of globin chains, which are essential components of hemoglobin. This reduction is caused by mutations in the genes responsible for the production of hemoglobin, leading to impaired oxygenation in affected individuals (3).

As the most prevalent hereditary disease worldwide, thalassemia affects both males and females equally. The alpha and β genes play a crucial role in the synthesis of the hemoglobin chain (4). The classification of thalassemia is based on the specific reduction or absence of each hemoglobin chain. Alpha thalassemia, located on chromosome 16, is one of the most prevalent hemoglobin disorders globally and is inherited through four lupus genes. On the other hand, β-thalassemia, whose coding gene is situated on chromosome 11, is a hemoglobin disorder characterized by anemia that is passed down from parents to children. B-thalassemia is commonly found in individuals from the Mediterranean region (5).

The β-globin chain's production rate is either reduced or absent in β-thalassemia, a condition first defined by Cooley in 1925. This deficiency primarily leads to microcytic and hypochromic anemia, as well as a wide range of syndromic forms. The severity of the anemia determines the classification into β-thalassemia major, minor, or intermediate (6, 7).

2. Material and methods

The research was carried out at the City Medical Complex Kart-e-Parwan hospital in Kabul city during the first half of 1401. It was a descriptive-cross-sectional study that included all anemic children referred to the hematology department of the hospital. The samples were collected using a simple (convenience) method, and the research sample was taken from the patients based on the hospital database. The sample size for this study was 216 children with hemoglobin levels ranging between 5 and 12 mg/dL. The collected data includes the registry number, age, sex, and type of Hb level. Hemoglobin electrophoresis is carried out utilizing the D10 automatic machine, a dual system that effectively separates hemoglobins within a mere 10 minutes.

The underlying mechanism employed in this process is chromatography. The patient's EDTA blood is utilized, and its formula is represented as HBF+HBA2-100=HBA1. Through the use of electrophoresis, various forms of hemoglobin can be quantitatively determined. Hemoglobin electrophoresis can be performed using either an acidic citrate agar buffer or an alkaline cellulose acetate buffer. In the more commonly used alkaline system, hemoglobin molecules are placed in the alkaline buffer of cellulose acetate, which possesses a net negative charge. Consequently, they migrate towards the positive electrode of the electrophoresis system. This method is known for its rapidity and reproducibility (8). The data were analyzed using SPSS 24 statistical software. The data derived in this study are presented using mean (SD), standard deviation (SD), frequency (F), and percentage (P).

3. Results

A total of 1224 children visited the hematology department of City Medical Complex, out of which 216 children with low hemoglobin levels underwent electrophoresis testing for the diagnosis and determination of the cause of anemia. A total of 216 children with low hemoglobin levels were examined using electrophoresis to diagnose and determine the cause of anemia. Among these children, 37 (17.5%) were diagnosed with β-thalassemia, while no cases of alpha-thalassemia were detected. The remaining anemic children were found to have different types of microcytic hypochromic anemias (Table 1). The gender-based frequency distribution of β - thalassemia reveals that 53.5% of males have β-thalassemia minor, while 46.5% of females have the same condition. In terms of β-thalassemia intermediate, 58.7% of males are affected, compared to 41.3% of females. For β-thalassemia major, the percentage is 66.7% in males and 33.3% in females. Additionally, among those who are carriers and have normal health, 62.7% are males and 37.3% are females (Table 1). In contrast, β-thalassemia major is predominantly observed in children under 6 years old, with a percentage of 88.9%. In children aged 6–12 years old, the prevalence drops significantly to 11.1%. Interestingly, no cases of thalassemia major were observed in children aged 12–18 years (Table 2). Among children under 6 years old, 44.2% have β-thalassemia minor, 50% have β-thalassemia intermedia, and 88.9% have β-thalassemia major.

For children aged 6–12 years, the percentages are 30.2% for β-thalassemia minor, 26.1% for β-thalassemia intermedia, and 11.1% for β-thalassemia major. In children aged 12–18 years, 25.6% have β-thalassemia minor, 29.3% have β-thalassemia intermedia, and no cases of β-thalassemia major were observed. Among children who are carriers (heterozygous) and have normal hemoglobin levels, the distribution is 50% for children under 6 years old, 33.9% for children aged 6–12 years, and 16.1% for children aged 12–18 years (Table 3). Furthermore, the findings indicated that β-thalassemia minor constituted 50.7% of the cases, intermedia accounted for 24.5%, major comprised 17.5%, and silent carrier represented 7.4%. Nevertheless, the predominant classification among thalassemia cases was minor (Table 4).

Table 1: Prevalence of anemia and thalassemias in children

	Number	Frequency
Anemia	1224	100%
β-thalassemia	216	17.6%
α-thalassemia	0	0%

4. Discussion

β-thalassemia is an inherited blood disorder that affects the production of hemoglobin, the protein responsible for carrying oxygen in red blood cells throughout the body. It is classified into two types: thalassemia major (also known as Cooley's anemia or transfusion-dependent thalassemia) and thalassemia intermedia (a non-transfusion-dependent form) (9). Beta thalassemia exhibits varying prevalence rates among diverse populations and geographic regions. It is particularly common in Mediterranean countries such as Greece, Italy, and Cyprus, as well as in Southeast Asia and specific areas of Africa. In these regions, the carrier frequency can reach as high as 10-15% in certain populations (10).

The prevalence of β-thalassemia in anemic children can vary depending on the population and region being studied. In this study, we found that 17.6% of children with anemia were diagnosed with beta-thalassemia. Among these, β-thalassemia major was the most common type, while gender was not associated with beta-thalassemia. Additionally, β-thalassemia major had the highest prevalence in children under 6 years old, as the symptoms of this disease start around 6 months of age, and these children have a lifespan until the age of 10.

Furthermore, based on electrophoresis parameters, β-thalassemia minor had the highest percentage, indicating its hereditary nature and transmission from carrier parents. Gursel, Orhan, et al. revealed that the male gender represented a majority, comprising over 50% of the cases. β-thalassemia major was the predominant type, accounting for 98.97% of the cases, while β-thalassemia intermedia accounted for 3.21%. Notably, the age group under 3 years exhibited the highest prevalence of β-thalassemia major, with 40 individuals affected (11). Dilip Kumar's study conducted in Bengaluru, India, in 2019 uncovered the prevalence of beta thalassemia among 85 individuals. The study primarily focused on children aged 12 and below, revealing that 62.5% of those affected were male, while the remaining 37.5% were female (12).

Table 2: Prevalence of different types of beta thalassemia in children based on their gender.

Thalassemia	Gender	Frequency	Percent
Minor	Male	23	53.5
	Female	20	46.5
	Total	43	100.0
Intermediate	Male	27	58.7
	Female	19	41.3
	Total	46	100.0
Major	Male	6	66.7
	Female	3	33.3
	Total	9	100.0
Silent Carrier	Male	74	62.7
	Female	44	37.3
	Total	118	100.0

Sajjad Afrouz and colleagues conducted a study in 2016 in Kohgiluyeh, Iran, which identified 150 individuals with beta-thalassemia. The study focused on individuals aged 20–30 years, with 51.4% being male and 48.6% being female (13). β-thalassemia can give rise to various complications, such as stunted growth and development, issues with the heart, liver, and spleen, problems with the endocrine system, blood clotting, and osteoporosis (14). It is crucial to conduct regular physical examinations, perform blood tests, and refrain from taking iron supplements in order to provide proper and continuous care for children affected by β-thalassemia. Additionally, seeking genetic counseling can prove advantageous in managing the condition effectively (15). Treatment options for this condition may include the potential administration of blood transfusions and the surgical removal of the spleen. Additionally, healthcare professionals may recommend daily doses of folic acid as part of the prescribed treatment plan (16). β-thalassemia poses a significant global health burden, particularly in regions with a high carrier frequency. The management of β-thalassemia involves the regular administration of blood transfusions and iron chelation therapy to prevent iron overload (17).

Table 3. The prevalence of β-thalassemias in children varies based on age.

Thalassemia		Frequency	Percent
Minor	0-6	19	44.2
	6-12	13	30.2
	12-18	11	25.6
	Total	43	100.0
Intermediate	0-6	23	50.0
	6-12	12	26.1
	12-18	11	23.9
	Total	46	100.0
Major	0-6	8	88.9
	12-18	1	11.1
	Total	9	100.0
Normal	0-6	59	50.0
	6-12	40	33.9
	12-18	19	16.1
	Total	118	100.0

Table 4: Prevalence of various types of beta thalassemia was determined based on diagnostic parameters determined by electrophoresis.

	Number	Frequency
Silent carer	16	7.4%
β-thalassemia minor	109	50.4%
β-thalassemia intermedates	53	24.5%
β-thalassemia major	38	17.6%
Total	261	100%

Despite advancements in treatment, complications can still arise, including ocular manifestations such as refractive errors, abnormalities in the retina, and dysfunction of the tear film (18). Healthcare professionals should be well-informed about the potential ocular implications of β-thalassemia in children with anemia and should regularly monitor their eye health (19). Early detection and intervention can play a crucial role in preventing or minimizing the impact of ocular complications on visual function (20).

5. Conclusion

In this investigation, it was discovered that 17.6% of children with anemia had beta-thalassemias, and among them, thalassemia major was the most common, while β-thalassemia was found to be independent of gender; furthermore, β-thalassemia major was most prevalent in children under 6 years old due to early onset symptoms and shorter lifespan, and in terms of electrophoresis parameters, β-thalassemia minor had the highest occurrence due to its hereditary nature and transmission from parents, which is particularly significant in traditional societies with family marriages and limited knowledge.

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Conflict of interest

We declare that we have no conflict of interest.

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Reference

1. Fotzi I, Pegoraro F, Chiocca E, Casini T, Mogni M, Veltroni M, Favre C. Case Report: Clinical and Hematological Characteristics of εγδβ Thalassemia in an Italian Patient. Frontiers in Pediatrics. 2022 Mar 17;10:839775.
2. De Sanctis V, Elsedfy H, Soliman AT, Elhakim IZ, Soliman NA, Karimi M, Elalaily R. The diagnostic approach to central adrenocortical insufficiency (CAI) in thalassemia. Mediterranean Journal of Hematology and Infectious Diseases. 2016;8(1).
3. Lee YK, Kim HJ, Lee K, Park SH, Song SH, Seong MW, Kim M, Han JY. Recent progress in laboratory diagnosis of thalassemia and hemoglobinopathy: a study by the Korean Red Blood Cell Disorder Working Party of the Korean Society of Hematology. Blood research. 2019 Mar 31;54(1):17-22.
4. Alaithan MA, AbdulAzeez S, Borgio JF. A comprehensive review of the prevalence of beta globin gene variations and the co-inheritance of related gene variants in Saudi Arabians with beta-thalassemia. Saudi medical journal. 2018 Apr;39(4):329.
5. Ekwattanakit S, Riolueang S, Viprakasit V. Interaction between Hb E and Hb Yala (HBB: c. 129delT); a novel frameshift beta globin gene mutation, resulting in hemoglobin E/β0 thalassemia. Hematology. 2018 Feb 7;23(2):117-21.
6. Jahangiri M, Rahim F, Saki N, Saki Malehi A. Application of Bayesian Decision Tree in Hematology Research: Differential Diagnosis of β-thalassemia Trait from Iron Deficiency Anemia. Computational and Mathematical Methods in Medicine. 2021 Nov 9; 2021:1-0.
7. Jahangiri M, Rahim F, Saki N, Malehi AS. Differential Diagnosis of β-thalassemia Trait from Iron Deficiency Anemia: Application of Bayesian Decision Tree.
8. Westermeier R. Electrophoresis in practice: a guide to methods and applications of DNA and protein separations. John Wiley & Sons; 2016 May 16.
9. Zhou Y, Cao Y, Fang Z, Huang K, Yang M, Pang G, Zhao J, Liu Y, Luo J. Research on the clinical factors of cardiac iron deposition in children with β-thalassemia major. European journal of pediatrics. 2023 Nov 8:1-8.
10. Eida RA, Elbedewy TA, Mabrouk MM, Elnassr NM. Prevalence of metabolic syndrome in β-thalassemia major adult patients in Tanta University Hospitals.
11. Gursel O, Kurekci AE, Tascilar E, Ileri T, Altun D, Tapan S, Kurt I, Kocaoglu M, Aydin A, Okutan V, Ozcan O. Premature atherosclerosis in children with β-thalassemia major. Journal of pediatric hematology/oncology. 2012 Nov 1;34(8):630-4.
12. Kumar D, Kinikar AA. Clinical Profile of Children with Beta-Thalassemia. Asian Journal of Clinical Pediatrics and Neonatology¦ Volume. 2019 Oct;7(4):42.
13. Afrooz, Kepati, Mohammad Amin, Zol-Adl, Mohammad, Sangtarash, Awadpour, Azizi, Ghale Gulab, Persai, Zafar. Investigating the prevalence of thalassemia and comparing the average blood indices in marriage candidates of Kohgiluyeh and Boyar Ahmad provinces with the type of thalassemia, 2013. Armaghane knowledge. 2016 Apr 10;21(1):84-94
14. Ravikumar Y, Koonyosying P, Srichairatanakool S, Ponpandian LN, Kumaravelu J, Srichairatanakool S. In Silico Molecular Docking and Dynamics Simulation Analysis of Potential Histone Lysine Methyl Transferase Inhibitors for Managing β-thalassemia. Molecules. 2023 Oct 25;28(21):7266.
15. Vishnevskia-Dai V, Sella King S, Lekach R, Fabian ID, Zloto O. Ocular manifestations of leukemia and results of treatment with intravitreal methotrexate. Scientific reports. 2020 Feb 6;10(1):1994.
16. Amra AA, Aldy F, Lubis B, Rahman E. The Effect of Anthropometry on Refractive Error and Ocular Biometry in Children with β Thalassemia Major. Open Access Macedonian Journal of Medical Sciences. 2021 May 14;9(T3):64-7.
17. Jeong J, Barra L. The use of anti-platelet and/or anticoagulant agents in the prevention of large vessel vasculitis-associated ischemic complications: a meta-analysis. Open Journal of Rheumatology and Autoimmune Diseases. 2014 May 4;2014.
18. Galanello R, Origa R. Beta-thalassemia. Orphanet journal of rare diseases. 2010 Dec;5:1-5.
19. Cao A, Galanello R. Beta-thalassemia. Genetics in medicine. 2010 Feb 1;12(2):61-76.
20. Taher AT, Musallam KM, Cappellini MD. β-thalassemias. New England Journal of Medicine. 2021 Feb 25;384(8):727-43.