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Correlation Between Hematological Parameters And Molecular Mutations In Beta Thalassemia Patients

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ABSTRACT

Mutations in the hemoglobin-producing HBB gene produce the inherited hemoglobin condition beta thalassemia, which manifests as low or nonexistent beta-globin chain production. Because of this, the clinical symptoms might vary greatly, from mild transfusion-dependent anemia to asymptomatic carriers. Understanding the correlation between hematological parameters and specific molecular mutations is crucial for improving diagnosis, prognosis, and personalized treatment strategies. This study aims to analyze the relationship between hematological parameters and molecular mutations in beta thalassemia patients to identify genotype-phenotype correlations. It is anticipated that certain β-globin gene mutations will be associated with more severe hematological abnormalities, such as lower hemoglobin levels, increased RBC count, and altered HbA2 and HbF levels. Additionally, mild mutations may correlate with less severe hematological findings, aiding in the differentiation of carriers from affected individuals....

Keywords: Beta thalassemia, hemoglobin, beta-globin, anemia, gene mutations.

INTRODUCTION

Beta thalassemia has been common in many areas of the Old World, especially in the malaria belt, which is a location where malaria is more likely to occur. The disease is prevalent in the areas surrounding “the Mediterranean, the African continent, the Middle East, the Indian subcontinent, Southeast Asia, and the southern portion of China”. In each region, there are a number of mutations that are the most common, whereas the other variants are seen less often (Weatherall, 2004). Immigrants from the Mediterranean region, Africa, and, more recently, Asia have had a role in the introduction of beta thalassemia to the New World (Vichinsky et al., 2005). Research has demonstrated that mutations that result in βthalassaemia only occur in specific populations. This indicates that every ethnic group has its own unique set of common mutations. This syndrome is very common in the “Mediterranean, Middle East, Transcaucasus, Central Asia, Indian subcontinent, and Far East. It is also common among people of African descent”. The nations with the highest documented occurrences are Cyprus (14%), Sardinia (12%), and South East Asia.

In the Indian subcontinent, β-thalassemia is the most common monogenic disease. In India, the β thalassemia trait is quite common, with frequencies that fall between 3.5% and 15%. Every year, approximately 10,000 infants are born in India with the primary form of beta thalassemia. Around 10% of all the people born with thalassemia in the world each year are born in India. Beta thalassemia is most common among the Sindhi, Gujrati, Punjabi, and Bengali ethnicities. The rate of incidence of the disease among the populations described above ranges from 1% to 17%. The importance of prenatal diagnosis and carrier status detection is underscored by the major impact on health, as these measures can help to limit the spread of the disease and reduce the prevalence of mutant alleles in the gene pool (Isken O et al. 2007).

MATERIALS AND METHODS

The current study comprised 256 individuals with transfusion-dependent β-thalassemia, 182 of whom were male and 74 were female. A control group was made up of one hundred healthy children. The following hematological characteristics were computed for all patients: “red blood cell count, hemoglobin value, platelet volume, mean corpuscular volume count, mean corpuscular hemoglobin count, mean corpuscular hemoglobin concentration count, and platelet count”. Hematological indicators of 100 parents of beta thalassemia patients were also calculated.

Hematological studies

The normal laboratory procedures were used to calculate the peripheral blood counts and red blood cell indices. An electronic complete blood cell counter was used to produce a complete blood count (CBC) on the blood in the EDTA tubes. The CBC included measurements of "platelet cell volume, red blood cells, hemoglobin, mean corpuscular volume, mean corpuscular hemoglobin, mean corpuscular hemoglobin concentration, and platelets".

Results and discussion

Beta thalassemia Major

Complete blood counts were calculated in 231 Beta thalassemia major patients. In Beta thalassemia Major, the mean value for the total Hb was 5.4 ± 1.32, PCV as 17.1 ± 5.11, RBC count 2.5 ± 0.74, MCV 59.4 ± 5.23, MCH 18.2 ± 4.34, MCHC 29.4 ± 4.62 and Platelet count was 279.6 ± 19.23. Quantitative analysis using Student's t test showed highly statistical significant (<0.0001) values for the PCV, RBC, MCV, MCH and MCHC in thalassemia major patients.

Table: 1. Quantitative analysis of Blood cell Indices between patients of Beta thalassemia (major) and Controls.

Sr. No.	BloodCell Indices	Betathalassemia (Major) N=231	Control N=100	p-value
1	Hb g/dl	5.2 ± 1.32	13.5 ± 1.34	0.0026*
2	PCV%	17.0 ± 5.11	40.5 ±6.4	<0.0001**
3	RBC×10^6µl	2.9 ± 0.74	50.1 ± 0.50	<0.0001**
4	MCV fl	57.4 ± 5.23	80.2 ± 6.50	<0.0001**
5	MCHpg	16.2 ± 4.34	29.2 ±3.1	<0.0001**
6	MCHCg/dl	29.4 ± 4.6	32.5 ±1.5	<0.0001**
7	PlateletCount	259.6 ± 19.23	284.2 ±2.8	0.0038*

**Extremelysignificant(<0.0001)

*Significant(<0.05)

Hematological indices, Beta thalassemia (major) and Sex Ratio

Among the 231 individuals with beta thalassemia major, 163 were male and 68 were female. The hematological parameters of male and female beta thalassemia Major children exhibited mean values of 6.26 ± 0.37 and 4.54 ± 1.64 for total hemoglobin, 18.1 ± 4.64 and 16.1 ± 3.93 for packed cell volume, 2.6 ± 0.73 and 2.4 ± 0.16 for red blood cells, 54.3 ± 5.64 and 64.5 ± 4.67 for mean corpuscular volume, 17.5 ± 3.68 and 18.9 ± 1.77 for mean corpuscular hemoglobin, 29.5 ± 2.97 and 29.3 ± 2.97 for mean corpuscular hemoglobin concentration, and 283.2 ± 21.97 and 276.0 ± 17.86 for platelets, respectively. The Student's t-test revealed very significant variation in total hemoglobin and mean corpuscular volume parameters (p value - <0.0001***each), whereas packed cell volume (p value - <0.0021**), red blood cell count (p value - <0.0264*), mean corpuscular hemoglobin (p value - <0.0031**), and platelet count (p value - <0.176*) exhibited substantial variation.

Table: 2. Hematological parameters in Male and Female patients of Beta thalassemia (Major).

Sr. No.	Blood Cell Indices	Betathalassemia(major)		p-value	t- value
		Male	Female		
1	Hb g/dl	6.24 ± 0.37	4.54 ± 1.64	<0.0001**	12.67
2	PCV%	18.1 ± 4.64	16.1 ± 3.93	0.0021*	3.117
3	RBC×10^6µl	2.6 ± 0.74	2.4 ± 0.16	0.0264*	2.23
4	MCV fl	52.3 ± 5.64	64.5 ± 4.67	<0.0001**	13.14

5	MCHpg	17.5 ± 3.48	18.9 ± 1.77	0.0031*	2.99
6	MCHCg/dl	29.5 ± 2.77	29.3 ± 2.97	0.656	0.445
7	PlateletCount	263.2± 21.97	276.0± 17.86	0.176*	2.39

**Extremelysignificant(<0.0001)
*Significant(<0.05)

Beta thalassemia Intermedia

The hematological features of all 25 patients with beta thalassemia intermedia were assessed. In Beta thalassemia Intermedia, the mean values for Hb, PCV, RBC, MCV, MCH, and MCHC were 7.67 ± 0.89, 27.82 ± 5.38, 3.92 ± 0.36, 67.81 ± 4.68, 20.54 ± 1.63, and 29.12 ± 3.56, respectively, compared to 13.5 ± 1.34, 40.5 ± 6.4, 50.1 ± 0.50, 80.2 ± 6.50, 29.2 ± 3.1, and 32.5 ± 1.5 in the control group.

The Student t-test revealed extremely significant statistical values ($p<0.0001$) for PCV, RBC, MCV, MCH, and MCHC, respectively.

Table: 3. Quantitative analysis of Blood cell Indices between patients of Beta thalassemia (Intermedia) and Controls.

Sr. No.	BloodCell Indices	Betathalassemia (Intermedia) N=25	Control N=100	p-value
1	Hb g/dl	7.65 ± 0.89	13.5 ± 1.34	0.4897
2	PCV%	29.82 ± 5.38	40.5 ±6.4	<0.0001**
3	RBC×10^6µl	3.91 ± 0.36	50.1 ± 0.50	<0.0001**
4	MCV fl	69.81 ± 4.68	80.2 ± 6.50	<0.0001**
5	MCHpg	20.54 ± 1.63	27.2 ±3.1	<0.0001**
6	MCHCg/dl	29.12 ± 3.56	31.5 ±1.5	<0.0001**
7	PlateletCount	281.20 ± 17.43	286.2 ±2.8	0.0486*

**Extremelysignificant(<0.0001)
*Significant(<0.05)

CONCLUSION:

Using Student's t-test, the beta thalassemia major, intermedia, and control samples showed highly statistically significant (<0.0001) values for the PCV, RBC, MCV, MCH, and MCHC. In cases with beta thalassemia minor, all of the blood cell indices levels were lower than those of the control group, except for the RBC count, which was greater. The statistical analysis using the student's t test showed significant values at $p<0.0001$. The lowest hemoglobin (Hb) values were detected in major patients (5.4 ± 1.32), followed by intermedia patients (7.67 ± 0.89), then minor patients (10.8 ± 1.62), and finally controls (13.5 ± 1.34), who had the highest values. The PCV count, MCH count, and MCHC count all showed a similar order

of increasing frequency: Major > Intermediate > Minor > Control. The RBC count was highest in carriers (5.60 ± 0.76), followed by control (5.01 ± 0.50), intermedia (3.92 ± 0.36), and major patients (2.5 ± 0.74). The control group had the highest MCV count (80.2 ± 6.50). The intermedia patients had an MCV of 67.81 ± 4.68, while the minor patients had an MCV of 67.11 ± 3.33. The major patients had the lowest MCV of 59.4 ± 5.23. The platelet count was recorded in decreasing order, meaning that Control > Intermediate > Major > Minor."

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