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PREPONDERANCE IN UNTIPICAL HAEMOGLOBIN VARIENTS AND BLOOD GROUPS IN UTTARAKHAND

Physiology

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ABSTRACT

Context: Untipical haemoglobinopathies are considered as atypical disorder occurs due to genetic disproportion within alpha and beta chains of amino acids order make changes. **Aims and Objectives:** A total of 933 cases were subjected in this study which were further studied for gene frequency. **Material and Methods:** Initiating with CBC (Complete blood cell count) doubtful cases were subjected to detailed study by HPLC (high performance liquid chromatography) for confirmation of Thalassemic cases. Ultimately gene occurrency analysis by ARMS (Amplification Refractory Mutation System) by PCR (Polymerace Chain Reaction). **Results:** A sum total of 933 subjects, aged between 01-30 yrs were studied and it depicts 3.8% prevalence. B-thalassemia trait was screened. The frequencies with respect to ABO is shown as B>0>AB>A. The amplicons which were analysed after gel electrophoresis were screened for ARMS, PCR, which IVS1-5 (G-C) is maximum for almost 55.0% and lowest is Codon 41/42 N (NCTT) 7.5%. **Conclusion:** As such patients requires repeated blood transfusion to availability of maximum type of affected Blood group is the time of need for availability to with blood banks. Further genetic studies will definitely help in effective for further pharmaceutical companies.

KEYWORDS

1. INTRODUCTION

The beginning elaborations started in 1930 on Tunisians people accumulating date of relationship of blood groups (ABO) system with altered haemoglobinopatheis.[1-4] This cumulative work with multiple speculations along with altered haemoglobinopatheis was initial of its kind.[4]

Further many observers has developed more precise method for the detection of β-thalassemia mutations in South East Asia, based on PCR generated restriction sites which are very synonymous to β-thalassemia mutations and later on practised in ruling out pathological mutations in mitochondrial DNA.[5] These modifications are observed at positions IVS1-5, IVS1-1 of β-globin gene.[6-8]

2. MATERIAL AND METHODS

These participants were children, married or pre-married candidates, patients with familiar, unfamiliar, doubtful or having family history, clinically suspicious or low haemoglobin drop patients which were mention by the physician and some may be self-participants.

2.1 Number of cases

A total of 933 cases were included in the study for detection of Thalasemic cases. Out of these only positive thalasemic cases were further analyzed for gene frequency]

2.2 Diagnostic criterion

By performing complete Blood Cell Counts; By HPLC (high performance liquid chromatography) for the detection and confirmation of Thalassemic cases; by (Bio-Rad D-10) haemoglobin testing system. By analysis by ARMS (Amplification Refractory Mutation System); PCR (Polymerase Chain Reaction) for gene frequency distribution. Isolation of DNA from whole blood which was preserved in anticoagulant (EDTA) by using the already commercially available kit (QIA and DNA blood midi kit 100 samples)

3. RESULTS

The total number of 933 cases included in the population we found blood group B (315) was observed to be more frequent and least O (144). Among Rh positive male, blood group B (195), and in female, blood group A (90) was most prevalent blood group (Table 1).

While observing the frequency of ABO Rh blood group in abnormal haemoglobin variants, which were accounted as 36 cases. Within these blood group B (14) is observed maximally and blood group AB (6) is least (Table 2).

The frequency of mutation detected in abnormal haemoglobin variants are 36 in number. The mutation IVS 1-5 (G→C) M (55.0%) is maximum and frequently observed and the least in codon 41/42 N (TCTT) is (7.5%).

If we segregate the abnormal haemoglobinopathies according to gender then we observe the males (25) are more sufferer then females (11) (Table 4).

So far as we analyse abnormal haemoglobinopathies distribution according to age and sex wise when we observe that more cases in males observed during 0 to 10 yrs and then subsequently decreases but in females the least between the age 16 to 30 yrs, but maximum between 0 to 10 yrs. (Table 5).

4. DISCUSSION

The sequel of abnormal haemoglobinopathies are moreover autosomal recessive disorders and are genetically inherited through one or both parents who might be the carrier or suffering in another presentable form with the disease.[9,10]

An aggregate of 933 patients were analysed for abnormal haemoglobin variants, ABO & rhesus blood groups ranging from 01 to 30 years. Out of the total 933 subjects 627 were males and 306 were females.

Table 1 depicts the dispersal of the blood group (ABO) & Rhesus (D) between study subjects. Blood group B was observed as the majority frequent 315 while blood group O was least frequent 144. Here we observe the same frequency in earlier studies.[11,12]

In Rh (D) blood typing 665 was Rh positive and 268 was Rh negative. Amongst Rh positive male blood group B (195) was observed as the most prevalent blood group proceeding ahead by blood group A (129), AB (70), O (64).

Between Rh positive female, blood group A (90) was most common followed by blood group B (57), AB (50) and O (10).

The frequencies pattern with respect to ABO can be shown by B > A > AB > O in males and A > B > AB > O in females. This study is almost synonymous to earlier studies.[13,14]

Table 2 shows that blood group B is observed maximally with 38.81% followed by blood group O (30.5%), AB (16.6%) & A (13.8%). Out of these Rh + were 35 (97.2%) & Rh- is 1 (2.7%).[13,14]

Table 3 depicts the spectrum of β-Thalassemia mutations in north india population (Uttarakhand) within this study a total of 36 β-Thalassemia alleles have been observed out of 933 individuals in North India population after screening ARMS PCR, the amplicons were subjected for gel electrophoresis with 1.6% as agarose. The product were visualized under UV transmitter for the DNA bands. Screening for 04 different types of β-Thalassemia mutations were observed i.e. IVS 1 – 5 (G-C)M, as most common,[15] followed by Fr 8/9 (+G)M, 619 bp deletion and codon 41/42 N (TCTT), at 285 bp, 215 bp, 242bp and 439 bp respectively. [16,17] The amplicons which were subjected for gel

electrophoresis after screening by ARMS PCR were IVS 1-5 (G-C)M (55.0%), Fr 8/9 (+G)M (17.5%), 619 bp depletion (10%) and codon 41/42 N(TCTT) (7.5%) respectively.[16,17] The earlier studies to their connection of its frequency in β-Thalassemia trait reported in Gujrat (10-15%), followed by Sindh (10%), Punjab (6.5%), Tamil Nadu (2.4%).[18-20]

Table 4 indicates the gender wise distribution of haemglobinopathics. Here we can easily observe that the number of males are more 25 (69%) then females 11 (30.5%) who were found as positive thalassemic cases.

This shows that such mutations in thalassemic patients are more prevalent in males then females.

Table 5 depicts that males shows more features and incidence during the age between 0 to 5 and 6 to 10 years parallarly, but decreases on increasing age. This years, be because of less life survival rate after the age of 10 years.

Table 1: Dissemination of ABO and Rh blood group in the study population (n=933)

Blood group	Male		Female		Total		Total
	Rh+	Rh-	Rh+	Rh-	Rh+	Rh-	
A	129	36	90	35	219	71	290
B	195	34	57	29	252	63	315
AB	70	46	50	18	120	64	184
O	64	53	10	17	74	70	144
Total	452	169	207	99	665	268	933

Table 2: Frequency of ABO Rh blood group in Abnormal haemoglobin varients (n=36)

Variables Blood Groups	No. Observed	Prevalence %
A	5	13.88
B	14	38.88
AB	6	16.66
O	11	30.55
Rhesus (Rh)		
D+	35	97.22
D-	1	2.77

Table 3: Frequency of mutation detected in abnormal haemoglobin varients (n=36)

Mutation Detected	No. of Patients Detected with Mutation	Frequency (%)	Amplified Product Size (bp)
IVS 1-5 (G→C)M	22	55.0	285
619 bp detection	4	10.0	242
Fr 8/9 (+G) M	7	17.5	215
Codon 41/42 N (TCTT)	3	7.5	439

Table 4: Gender wise distribution of haemoglobin

Gender	No. of Cases
Males	25
Females	11

Table 5: Age and sex wise distribution of hemoglobinpathies

Males		Females	
Year	No.	Year	No.
0-5	9	0-5	4
6-10	9	6-10	4
11-15	4	11-15	0
16-20	1	16-20	1
21-25	1	21-25	1
26-30	1	26-30	1

5. CONCLUSION

These varied type of abnormal haemoglobinopatheis with particular to thalassamia are the greatest factor of genetic mutational anamoly, which has eventually lead to wide spread public health disorder, therefore because clinical importance after birth.

Elaborate study of varied haemoglobinopathies and their screening will definitely be a center stone while observed their occurrences with every region of the state along with the data shoring with regional research center – precise by formulating a data of blood groups in relation to abnormal haemoglobinopathies may furnish details about the available of human blood during emergencies, and also enlightens the possibility of future burden of disease.

6. Source of Funding

None.

7. Conflict of Interest

None.

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