Screening for Various Haemoglobinopathies to Observe Prevalence and Patterns in Population of District Datia (M.P.), India

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Abstract

Purpose: Haemoglobinopathies are the most common inherited disorder of Red Blood Cells across globe and one of the major public health problems in many regions of India. Variation in ethnic and regional prevalence is observed. The prevalence and pattern of haemoglobinopathies in Datia region has not been reported limiting the knowledge of diseases profile in this region. Present work showed the broad view of various haemoglobinopathies in the local area and population at risk. Efforts were made to identify prevalence of Sickle cell anaemia, β -thalassemia major and G-6-P-D deficiency among patients attending various OPDs of District Hospital, Datia, Madhya Pradesh, India.

Methods: All anaemia patients referred by Govt. hospitals/facilities (Datia District hospital) exhibiting anaemia were screened for various haemoglobinopathies. A total of 605 cases exhibiting anemia received from September 2017 to March 2020 at the Model Rural Health Research Unit (MRHRU), Datia, M.P. were tested for different haemoglobinopathies. Samples were received from District Hospital Datia. The samples were tested by solubility and cellulose acetate electrophoresis and HPLC and G6PD deficiency.

Results: A total 605 patients were screened for haemoglobinopathies from District Hospital, Datia. It was found that 13 cases (2.14%) were of β thalassemia trait and other rare variants followed by 4 cases (0.66%) of sickle cell trait. A total of 3 cases (0.5%) of β thalassemia major was observed in total samples of 605.

Conclusion: Very low prevalence rate of Sickle cell anaemia and thalassemia was observed. Large scale screening in the region is required for identifying the true burden.

Introduction

Haemoglobinopathies are considered the commonest genetic defect globally¹. Datia is the district headquarters of Datia district (a sub-rural region), 69 km from Gwalior and is surrounded by state U.P., India on north side. The haemoglobinopathies are one of the most neglected disease² especially in rural settings. The health and medical facilities are mainly provided by the District medical hospital and disease profile is unknown here. The screening of haemoglobinopathies in Datia region has not been reported earlier. This study was aimed to screen the patients having severe anaemia from district hospital Datia for sickle cell disease, β -thalassemia major and G6PD deficiency. Further, this study was mandated to provide supportive care and management of sickle cell through counselling and prevent birth of future affected babies.

Material & Methods

All anaemic patients (<11 gm/dL haemoglobin) and other suspected of haemolytic disorder referred by Govt. hospitals/facilities were referred to Model Rural Health Research Unit, Datia laboratory for diagnosis. The samples were obtained after taking written and understood consent (informed consent

form) from patients and under the age of 18 years of patients were taken from their parent/legal guardians. After obtaining the informed consent about 2 mL intravenous blood, each free from blood transfusion was collected in ethylene diamine tetra acetic acid (EDTA) vials as anticoagulant. In total, 605 samples were screened for various haemoglobinopathies. Presence of sickle haemoglobin (HbS) was tested by solubility test^{3,4}, confirmation of genotype was done by Cellulose acetate membrane electrophoresis^{3,7} at pH 8.6. Quantification of HbA2 and HbF was performed by CE-HPLC⁷. β thalassemia was investigated by CE-HPLC. Parental analysis was done to confirm genotype. Screening for G6PD deficiency was performed by DPIP decolourization method^{8,9}. The study was carried out in accordance with Helsinki declaration. This project was part of the service provided by MRHRU Datia. A post facto ethical clearance was taken to publish the collected data and also to elaborate this on-going project. Statistical analysis is done by using excel, the categorical analysis is presented as percentages. The datasets used and/or analysed during the current study available from the corresponding author on reasonable request.

Results

In total screened 605 patients, 485 (80.2%) were females and 120 (19.8%) were males. Age of majority of participants (49.3%) was in the age group of 21–30 years followed by the 0-10 years group (22.3%), 11-20 years group (19.0%) and 31- above years group (9.4%). Caste-wise distribution revealed that 48.6% patients were from Other Backward Classes (OBC), 31.4% from Scheduled Caste (SC) communities followed by 14.9% general category, Scheduled Tribe (ST) constituted only 1.65% of the samples screened also 3.5% did not disclose their caste category. Six patients were Blood Transfusion dependent. Four subjects showed the presence of HbS (HbAS) (Sickle Cell Carrier) and 13 subjects were β -thalassemia trait (Table 1) and three cases were observed of thalassemia major. No G6PD deficiency individuals were identified in total tested 605 samples (Table 1). The prevalence of β -thalassemia major and β -thalassemia minor were found to be 0.5% and 2.14% respectively (Table 1), also the prevalence of sickle cell trait found was 0.66%. All β -thalassemia major were young children (Table 3), as were reported to hospital early due to difficulty they face in normal living. Analyses of β -thalassemia trait by age showed more adults in their 21-30 age group and too majority being females (Table 2).

Table 1
Distribution of patients according to
haemoglobinopathy found in current study:

No. of patients (out of 605)	Haemoglobinopathy
4	Sickle cell trait
13	β -thalassemia trait
3	thalassemia major
0	G6PD deficiency

Table 2
Analyses of β -thalassemia trait by age, gender, cast and category distribution.

β -thalassemia trait	Age	Gender	Cast	Category
1.	21-30 years	Female	Jha	OBC
2.	11-20 years	Female	Rajput	OBC
3.	21-30 years	Female	Sharma	Gen
4.	21-30 years	Female	Tilwani	Gen
5.	21-30 years	Male	Prajapati	SC
6.	21-30 years	Female	Prajapati	SC
7.	31 and Above	Male	Kukreja	Gen
8.	21-30 years	Female	Kukreja	Gen
9.	0-10 years	Male	Kukreja	Gen
10.	31 and Above	Male	Sindhi	Gen
11.	31 and Above	Female	Sindhi	Gen
12.	21-30 years	Female	Sindhi	Gen
13.	21-30 years	Female	Sindhi	Gen

Table 3
 β -thalassemia major by age, gender, cast and category distribution:

β -thalassemia major	Age	Gender	Cast	Category
1.	0-10 years	Male	Prajapati	SC
2.	0-10 years	Female	Kukreja	Gen
3.	11-20 years	Male	Sindhi	Gen

Discussion

The present study provides an overview on the presence of haemoglobinopathies in patients attending the district hospital, Datia. In the present study it was observed that the majority of the patients attending the clinic were females, mainly because of all suspected haemolytic anaemia/anaemia cases were referred by Gynaecology Department of District Hospital. The reason for the high proportion of the female patients may be due to commonly prevalent anaemia though the disease is not sex-linked¹⁰. Unlike sickle cell anaemia, β -thalassemia major is detected early among infants and as they require blood transfusion. Due to presence of tribes in Datia region it was expected that sickle cell disease could be prevalent, but

results were contradictory^{11,12,13}. The general prevalence of sickle cell anaemia and β -thalassemia trait lies between 1-44% and 3-17% respectively¹³. In this study a prevalence of β -thalassemia minor was found to be 2.14% and sickle cell trait 0.66% respectively in Datia District. The found cases have been provided by supportive care as after identification of such cases, the parents were advised for screening of all members for haemoglobinopathies and antenatal screening in utero after conception. The 0% observance of G6PD deficiency might be due to the majority of the patients attending the clinic were females, mainly because of all suspected haemolytic anaemia/anaemia cases were referred by Gynaecology Department of District Hospital. As, G6PD deficiency is a genetic disorder and passed to the child from one or both parents. The X chromosome carries the defect that causes this deficiency, from the two sex chromosomes. Since, males have only one X chromosome, while females have two X chromosomes¹⁴. In males, one copy of the altered gene is enough to cause G6PD deficiency. However, in females, a mutation have to be present in both copies of the gene. Since it's less likely for females to have two altered copies of this gene, hence we are getting this kind of result in this study. Males are affected by G6PD deficiency much more frequently than females, and they are less reported to hospital. Limitations of the study are small sample size and it is hospital-based study. Community based studies are required for arriving at the true picture of the status of haemoglobinopathies in Datia District. Similarly, large scale studies are essential to identify G6PD deficiency among tribals and non-tribals to enable health authorities to carry out malaria control in the region¹⁴ as this region is reported to carry high incidence of *Plasmodium vivax* infection.

Approval by the appropriate ethics committee for research: The study was approved by the institutional ethics committee of ICMR- National Institute of Research in Tribal Health, Jabalpur, India via letter no. NIRTH/IE C/01/31/2021

Declarations

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The manuscript has been approved by the Publication Screening Committee of ICMR-NIRTH, Jabalpur and assigned with the number ICMR-NIRTH/PSC/31/2021

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