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A STUDY OF PREVALENCE OF HEMOGLOBIN E AND GLUCOSE 6-PHOSPHATE DEHYDROGENASE DEFICIENCY AMONG ETHNIC KARBI TRIBAL POPULATION IN A TERTIARY CARE CENTRE IN NORTH – EAST INDIA

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ABSTRACT: Introduction: Hemoglobin E (HbE) disorders and glucose-6-phosphate dehydrogenase (G6PD) deficiency are two important red cell genetic abnormalities highly prevalent in malaria-endemic regions of Southeast Asia. The Karbi tribe, one of the major ethnic groups of Northeast India, is considered to have a significant burden of these disorders; however, data on their prevalence remains limited. This study aimed to determine the prevalence of Hb E disease, HbE trait, and G6PD deficiency among Karbi tribal individuals attending a tertiary care centre in Northeast India. **Methods:** A total of 600 Karbi tribal patients were enrolled in this cross-sectional study facility based study. Demographic details including age, gender, and sub-clan distribution were recorded. High performance liquid chromatography was done for Hemoglobin variant and G6PD by a semi-automated analyzer. Data were analyzed for prevalence patterns across age groups, gender, and sub-clans. **Results:** Hemoglobin E trait was found in 84 subjects (14%) and Hemoglobin E disease in 12 individuals (2%). G6PD deficiency was observed in 36 individuals (6.0%). Females showed a higher prevalence of HbE trait and disease, but the difference was statistically not significant. Males showed a statistically significant higher frequency of G6PD deficiency compared to females (4.33% vs 1.67%). Sub-clan analysis revealed Timung sub-clan had slightly higher distribution of HbE trait and G6PD deficiency. Coexistence of Hemoglobin E trait and G6PD deficiency was found in 6 individuals (1.0%). **Conclusion:** This study highlights a high prevalence of HbE trait and a considerable burden of G6PD deficiency in the Karbi population visiting the tertiary care center. The findings underline the importance of routine screening and genetic counseling in this community.

INTRODUCTION: Hemoglobinopathies and red cell enzymopathies are of significant public health concern, affecting millions, particularly in Southeast Asia and Northeast India.

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Hemoglobinopathies are a group disorders that result from either a qualitative structural abnormality of the hemoglobin (Hb) molecule or decreased synthesis of globin chains (thalassemia) [1, 2].

In Southeast Asia, with a reported prevalence of around 30–45%, hemoglobin E (HbE; β26Glu-Lys) predominates [3]. The prevalence soars to as high as 60% in parts of Thailand, Laos, and Cambodia, peaking at more than 70% in certain ethnic populations of Thailand [4].

The heterozygous state, known as Hb E trait is generally asymptomatic, while the homozygous hemoglobin E (HbEE) presents with mild anemia, splenomegaly, and episodes of hemolysis [5]. As per a theory proposed by Haldane more than 60 years ago, a study suggested that higher rates of hemoglobinopathies, particularly high HbE allele frequencies are more consistent in malaria-infected areas [6, 7].

Red blood cell enzymopathies are a group of genetic disorders that alters intraerythrocytic metabolism [8, 9]. The most common inherited red cell enzymopathy in humans is Glucose 6 Phosphate Dehydrogenase (G6PD) deficiency. It is estimated that more than 400 million people of the global population, around 4.9% has G6PD deficiency [10]. It is more prevalent in Africa, Asia, Europe and the Mediterranean region, areas which are malaria is currently endemic or was endemic in the past [11].

In India, the distribution of G6PD deficiency is very heterogeneous and the prevalence of G6PD deficiency ranges 0.3 to 30.7% per cent in various regions [12]. The clinical spectrum of G6PD deficiency may range from mostly asymptomatic individuals to chronic non-spherocytic hemolytic anemia. Patients with G6PD deficiency may also present with neonatal jaundice or acute hemolytic anemia on exposure to exogenous agents (acute infections, drugs or fava beans) [13].

The Karbi tribe, one of the major indigenous communities of Assam, resides predominantly in the Karbi Anglong and West Karbi Anglong districts. A study carried out by ICMR showed a high density of malarial parasites in Karbi Anglong [14].

Due to geographical isolation and endogamous practices, genetic disorders may show distinctive prevalence patterns within this population. However, there is a paucity of data regarding the burden of inherited hematologic conditions among the Karbi people.

This study aims to fill this critical knowledge gap by evaluating the prevalence of HbE (disease and trait) and G6PD deficiency among the Karbi tribal population and providing baseline data to inform screening and counseling initiatives. There is an urgent need to develop integrated hierarchical core facilities to manage such diseases.

MATERIALS AND METHODS: The cross-sectional study was carried out in ethnic tribal Karbi population visiting Diphu medical college and Hospital (DMCH), Diphu, Karbi Anglong, Assam for a period of two years from January 2023 till December 2024 with due permission from the Institutional Ethics Committee through Letter No-DMCH/EC/2022/105. Written, informed consent was taken from all adult subjects. In case of minors, written informed consent from the parent/guardian was taken and assent from the minors. Karbi identity and sub clan was confirmed by self report. With 95% confidence interval and at 4% margin of error, the required minimum sample size was calculated to be 595, rounded off to 600. Consecutive sampling was done and all eligible subjects were enrolled. Pregnant women and individuals receiving blood transfusion within three months were excluded from the study.

Hemoglobin E was analysed by collecting peripheral blood in an EDTA vial using High-Performance Liquid Chromatography (HPLC) in Biorad D10. Hb E trait was considered with Hb E levels < 40%. Hb E disease was considered with Hb E levels between 70% and 90%. Quantitative screening of G6PD deficiency was detected with the help of a semi-automated analyzer (SD Biosensor Healthcare Pvt Ltd). Quantitative G6PD deficiency was defined by enzyme activity levels below <30% of the adjusted male median (5-15 U/g Hb).

Statistical Analysis: Microsoft Excel (Office 2019, Microsoft Corporation, Redmond, Washington, USA) was used to create the database, and GraphPad Prism version 9.00 (GraphPad Software, Inc., California, USA) was used for statistical analysis. Nominal variables are expressed as percentages and the number of cases, while continuous variables are presented as mean ± standard deviation. A p-value < 0.01 indicated high statistical significance, while a two-tailed p-value < 0.05 was deemed statistically significant.

RESULTS AND OBSERVATION: Following all relevant inclusion and exclusion criteria, samples from 600 study participants was collected and

analysed. Of these 600 study participants, 312 were male and 288 females with a male: female ratio of 1.08:1. The highest number of participants was found in the age group of 11-20 years for both males and females. The mean age of male participants was 27.62±15.97 years, and the mean age of female participants was 27.93±15.65 years.

TABLE 1: AGE AND GENDER-WISE DISTRIBUTION OF STUDY PARTICIPANTS (N = 600)

Age Group (years)	Male (n=312)	Female (n=288)	Total
1 – 10	52	44	96
11 – 20	70	64	134
21 – 30	62	58	120
31 – 40	50	52	102
41 – 50	44	40	84
51 – 60	34	30	64
Total	312	288	600

Out of 312 males, 268 (85.89%) were found to have normal HPLC pattern. Hemoglobin E trait was found in 38 (12.17%) study subjects and Hemoglobin E disease in six (1.92%). Among females, 236 (81.94%) were found to have normal HPLC pattern. Hemoglobin E trait was found in 46 (15.97%) study subjects and Hemoglobin E disease in six (2.08%). A higher prevalence of HbE disease and trait was observed in female participants. But prevalence of both HbE trait and HbE disease does not show any statistically significant difference between male and female participants as per Fisher's Exact Test. When both genders were considered together, normal hemoglobin HPLC pattern was found in 504 individuals with a prevalence of 84%. Hemoglobin E trait was found in 84 subjects (14% prevalence) and Hemoglobin E disease was found in 12 individuals with a prevalence of 2%.

TABLE 2: DISTRIBUTION OF HEMOGLOBIN E DISORDERS (HBE TRAIT & DISEASE)

Hemoglobin Pattern	Male (n=312)	Prevalence (%)	Female (n=288)	Prevalence (%)	Total (N = 600)	Prevalence (%)
Normal HPLC pattern	268	85.89%	236	81.94%	504	84.0%
HbE Trait	38	12.17%	46	15.97%	84	14.0%
HbE Disease (EE)	6	1.92%	6	2.08%	12	2.0%

Prevalence of G6PD Deficiency: G6PD deficiency was observed in 36 individuals (6.0%), with 26 (4.33%) males and 10 (1.67%) females affected. Fisher's Exact Test (Two-tailed) p value was found to be 0.0197. The prevalence was significantly higher among males ($p < 0.05$), consistent with the X-linked inheritance pattern. The Odds Ratio (OR) for G6PD deficiency in this study was found to be 2.53.

TABLE 3: DISTRIBUTION OF G6PD DEFICIENCY

Gender	Males	Females	p value
G6PD deficiency (n)	26	10	$p < 0.05$
Prevalence (%)	4.33	1.67	(0.0197)

Distribution of Hemoglobin E Disease, Trait, and G6PD Deficiency among Karbi Sub-clans: The distribution of HbE disease, HbE trait and G6PD deficiency was further analysed among the five major sub clans of the Karbi tribe.

Among 190 Engti participants, 20 (10.52%) had HbE trait, 4 participants had HbE disease (2.10%) and 9 participants (4.73%) had G6PD deficiency. Among 160 Terang participants, 18 (11.25%) had HbE trait, 3 (1.87%) participants had HbE disease and 8 participants (5%) had G6PD deficiency. Among 90 Enghee participants, 15 (16.67%) had HbE trait, 3 participants had HbE disease (3.33%) and 6 participants (6.67%) had G6PD deficiency.

Among 90 Teron participants, 16 (17.78%) had HbE trait, 1 participant had HbE disease (1.11%) and 7 participants (7.78%) had G6PD deficiency. In the Timung sub clan, 15 participants (21.42%) tested positive for HbE trait, 1 for HbE disease (1.42%) and 7(10%) for G6PD deficiency.

TABLE 4: DISTRIBUTION OF HEMOGLOBIN E DISEASE, TRAIT, AND G6PD DEFICIENCY AMONG KARBI SUB-CLANS

Sub-clan	Hemoglobin E Trait (n)	Hemoglobin E Disease (n)	G6PD Deficiency (n)	Total (n)
Engti	20	4	9	190
Terang	18	3	8	160
Enghee	15	3	6	90
Teron	16	1	7	90
Timung	15	1	7	70
Total	84	12	36	600

Combined Hemoglobin E Disorders and G6PD Deficiency: Coexistence of Hemoglobin E trait and G6PD deficiency was found in 6 individuals (1.0%), while no cases of combined HbE disease and G6PD deficiency were observed.

TABLE 5: G6PD DEFICIENCY BY GENDER AND COEXISTENCE WITH HBE DISORDERS

Condition	Male (n=312)	Female (n=288)	Total (N = 600)	Prevalence (%)
G6PD Deficient (any Hb status)	26	10	36	6.0%
HbE Trait + G6PD Deficiency	4	2	6	1.0%
HbE Disease + G6PD Deficiency	0	0	0	0.0%
Normal Hb + G6PD Deficiency	22	8	30	5.0%

DISCUSSION: Hemoglobinopathies E, both disease and trait are autosomal recessive inherited disorders, primarily affecting the globin moiety of the Hb molecule. In the past, these hemoglobinopathies exhibited limited distribution, confined to tribal populations, particularly in maralia endemic regions [6.7]. Tribal population of India constitutes approximately 8.5 per cent of the total population of India1 [15]. In India, the tribal population is a heterogeneous group, each being distinctive in their genetic make-up and geographical identity. These tribals can be broadly divided into several groups (i) Tribal populations in the North-East, (ii) Tea Garden tribal populations, (iii) Tribal populations in central India, (iv) Tribal populations in western India, (v) Tribal population in eastern parts of Odisha and Andhara Pradesh, and *(vi)* Tribal populations in south India [16]. This distinction is important because there are some variations in the nature, composition and clinical severity of various haemoglobinopathies in different tribal areas of the country. Our study was confined to the ethnic tribal population in Diphu, Karbi Anglong.

In a study carried out in West Bengal spanning 10 years and examining 119,336 samples, the prevalence of HbE trait was found to be 3.02% and HbE disease was found to be 0.34% [17]. In this study of 600 ethnic Karbi tribal participants from Northeast India, a higher prevalence rate of Hemoglobin E trait (14.0%) and Hemoglobin E disease (2.0%) was found. Our study reflected recognized patterns across the region, where HbE is notably common due to historic selective pressures from malaria. In some areas of Northeast India, carrier rates of HbE may reach as high as 55% [18]. Our observed figures, while somewhat lower, remain consistent with the broader regional distribution and suggest a significant genetic health concern.

Meanwhile, the G6PD deficiency prevalence of 6.0%, with 72% of cases in males—accords with established reports among Indian tribal groups. Nationwide, such tribal communities exhibit G6PD deficiency rates ranging from around 2.3% up to 27%, with an average near 7.7% [12]. Specifically in Northeast India, prevalence values commonly span between 1.9% to as high as 15–20% in some tribes. In the "Genetic Atlas of the Indian Tribes", published by ICMR, G6PD deficiency was reported to be present in all the tribal groups studied from North-East India. A very high frequency was observed in *Angami Nagas* (27.0%) from Nagaland followed by *Rabhas* (15.8%) and *Mikirs* (15.6%) from Assam [19].The pronounced male predilection mirrors the X-linked inheritance of G6PD deficiency.

Molecular studies have shown considerable genetic diversity in G6PD variants across India. For example, the G6PD Orissa and Mediterranean mutations are among the most prevalent comprising more than half of cases in some groups and are associated with varying levels of enzyme activity

and clinical severity [20]. While our study did not investigate molecular subtypes, these findings highlight the need for future genotyping to better understand susceptibility and clinical implications within the Karbi population.

Of particular interest in our cohort is the 1.0% co-occurrence of HbE trait and G6PD deficiency, with no observed combination of homozygous HbE disease and enzyme deficiency. In a study carried out in hundred unrelated healthy adults in Kachin, Myanmar co-inheritance of G6PD deficiency and HbE occurred at 23% [21]. Our study found considerably lower rates of co-inheritance. While limited by small numbers, this adds to a growing body of literature suggesting that co-inheritance, though rare, may influence disease severity particularly when overlapping hemolytic mechanisms are involved. In regions like Odisha, similar co-inheritance patterns have been implicated in increased neonatal morbidity and transfusion needs.

In malaria-endemic areas, prevalence of red blood cell disorders that are related to regional distribution of malaria are driven by selection pressure of malaria, and partially by migration. The evolutionary persistence of these disorders has been attributed to the selective advantage they confer against severe Plasmodium falciparum infection. Strong positive selection imposed by malaria has shaped the genetic architecture of many endemic populations, with hemoglobinopathies and G6PD deficiency leaving characteristic genomic imprints over generations [21, 22]. The malaria-resistant red-cell disorders such as G6PD deficiency and thalassemias are common in parts of Southeast Asia where malaria is endemic [23, 24, 25].

Although extensive literature exists on the protective roles of sickle cell hemoglobin (HbS) and hemoglobin C (HbC) against malaria, fewer studies have addressed the impact of HbE. However, several investigations in northeastern Myanmar and along the China–Myanmar border revealed high co-occurrence of HbE and G6PD deficiency, suggesting parallel adaptive responses to malarial pressure. Our study adds to this growing body of evidence by reporting similar trends in the Karbi population, thereby highlighting the importance of HbE in the malaria–host genetic interaction model. Clinically, the potential interactions between HbE and G6PD deficiency warrant careful evaluation. G6PD-deficient individuals are vulnerable to hemolytic crises triggered by infections, drugs (e.g., certain antimalarials), or oxidative stress. Coexistence with even mild hemoglobinopathies like HbE trait could compound this risk, especially in settings with limited access to healthcare or where antimalarial use is common.

Limitations of the study include absence of molecular characterization for the G6PD variants and lack of functional hemolysis assessments (such as enzyme activity assays). It was also limited to a facility based study with limited generalizability beyond the attending population. Functional assays of malaria protection were beyond the scope of this study.

Future directions should include molecular typing of HbE and G6PD mutations to better understand genetic diversity within the Karbi population. Longitudinal studies linking these genotypes with malaria incidence and clinical outcomes would further clarify the selective advantage hypothesis. Expanding research to other tribes of Northeast India could also shed light on the broader evolutionary and epidemiological dynamics of hemoglobinopathies in this malaria-endemic region.

Future investigations should also explore the clinical impact of co-inheritance on anemia severity, transfusion needs, and response to common hemolytic triggers.

CONCLUSION: In conclusion, our findings underscore a noteworthy prevalence of both HbE disorders and G6PD deficiency in the Karbi tribal population, with implications for newborn screening programs, genetic counseling, and safer therapeutic protocols particularly in malaria-endemic settings. Further molecular and longitudinal clinical studies are urgently needed to inform public health strategies and mitigate hemolysis-related morbidity in this vulnerable population.

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CONFLICTS OF INTEREST: None

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