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# β-globin variants present in Western Sudan

## Abstract

**Objective:** A variety of observations of the hemoglobin D (Hgb D) phenotype has occurred in association with family studies of patients with sickle cell disease. Very little is known about the occurrence and prevalence of the Hgb D variant and its impact on blood profiles among Sudanese. This study was aimed at determining the percentage of Hb D in North Darfur State, West Sudan, whose population has been shown to have hemoglobin-S (Hb S) disease.

**Methods:** From December 2017 to August 2018, this descriptive community-based investigation was conducted. Six hundred and sixty-six (666) people were randomly selected to participate in this study. With each participant's there was verbal consent. A questionnaire was designed to collect personal details. 5 mL of venous blood was gathered in EDTA containers. The Hb D variant was checked using Sebia Minicap Automated Capillary Electrophoresis System- USA and frequency was calculated using version 21.0 of the software package for social science (SPSS).

**Result:** The prevalence of Hb variants was as follows: AD=0.6%, SS=2%, AS=10.5 %, and AA=86.9%.

**Conclusion:** The prevalence of Hb D variant was 4 (0.6 %) in 666 participants from four western Sudanese tribes, beside reported of Hb SS and Hb AS.

**Keywords:** Hb variants, HbD, north Darfur, Sudan

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**Abbreviations:** NN, normal Hb; AS, heterozygote of hemoglobin S; SS, homozygote of hemoglobin S; DD, homozygote of hemoglobin D; AD, heterozygote of hemoglobin D

## Introduction

Hemoglobin (Hb) is a tetrameric protein made up of two alpha (α) and two beta (ß) globin chains with heme as a prosthetic group. Its principal function is to carry oxygen to the tissues and return $CO_2$ to the lungs. Adult hemoglobin (Hb A) is the most common type, with two α and two β chains. The most common type of hemoglobin found at birth is fetal hemoglobin (Hb F), which is made up of two α and two γ chains.[1]

Approximately 7% of the world's population has mutations in genes that code for hemoglobin chains. These genetic mutations can affect the outcome at which globin chains are formed, triggering thalassemia, or they can change the composition of the molecule, resulting in hemoglobin variants.[2] Hemoglobin variants are generally the result of single amino acid substitutions induced by point mutations in globin chain genes, ending in a tetramer of various physicochemical properties.[3] The globin Gene Server database (http://globin.cse.psu.edu/) contained 1198 hemoglobin variants before September 2014. The majority of the hemoglobin variants listed may not elicit symptomatic clinical manifestations; but, in certain circumstances, such as Sickle cell hemoglobin, they can be linked to relevant pathophysiology. Hb S is the most frequent hemoglobin variant in the world; its clinical outcome is severe in homozygous or in association with other relatively common hemoglobinopathies, such as beta-thalassemia, Hb C or Hb D.[4] The most common pathological Hb variants worldwide in descending order of prevalence are Hb S, HbE, HbC, and Hb D. All of these hemoglobins have single amino acid substitutions in the β chain. At position 6 of the β chain, Hb S has a valine instead of glutamic acid and in the similar position Hb C had a mutation in glutamic acid instead of lysine. At position 26 of the β chain, lysine is replaced for glutamic acid in Hb E. Additionally, in Hb D, glutamine is substituted for glutamic acid at position 121 of the β-globin chain,[5] resulting in Hb D disease which is an autosomal recessive condition consequential in mild hemolytic anemia. The heterozygous variant (Hb AD) is more popular than the homozygous type (Hb DD), which is uncommon.[6]

## Materials and methods

Between December 2017 and August 2018, a cross-sectional, prospective community-based study was implemented in Northern Darfur. The aim of the study was to figure out which Hb variants existed in North Darfur. Six hundred and sixty-six (666) people were randomly selected from all parts of north Darfur after granting their informed consent, with represent of all the major Northern Darfur tribal communities. Direct structural interviewing and a questionnaire were used to gather information. Demographic details such as age, sex, ethnicity (tribe), and family history were recorded. The State Ministry of Health approved ethical clearance. Sebia Minicap Automated Capillary Electrophoresis System- USA was used to identify Hb variants in five mL of venous blood (with K2-EDTA) from each respondent.

## Results

Six hundred and sixty-six individuals were participating in the present study, with an average age of 21.3 years. Of these 55% were females with an average age of 21.7 ± 18.3 years and the remaining 45% were male with an average age of 20.9 ± 15.4 years (Table 1). The respondents came from different tribal groups, who constitute the majority of the Northern Darfur tribes (Table 2). The study showed the prevalence of Hb variants AA; 86.9%; Hb SS=2%; Hb AS=10.5% and Hb AD=0.6%.

**Table 1** Gender and average age of the research group

| Gender | Frequency | Percent % | Mean age (years) |
|---|---|---|---|
| Male | 297 | 45 | 20.9 ± 15.4 |
| Female | 369 | 55 | 21.7 ±18.3 |
| Total | 666 | 100 | - |

**Table 2** Percentage of Hb variants in the research group

| | | Hemoglobin variants | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| | | AA | | AS | | SS | | AD | |
| Locality | Total | Frequency | % | Frequency | % | Frequency | % | Frequency | % |
| ALFASHER | 195 | 160 | 82.1 | 26 | 13.3 | 6 | 3.1 | 3 | 1.54 |
| SARF-OMR | 15 | 13 | 86.7 | 2 | 13.3 | 0 | 0 | 0 | 0 |
| KALMENDO | 15 | 13 | 86.7 | 2 | 13.3 | 0 | 0 | 0 | 0 |
| ALMALHA | 17 | 17 | 100 | 0 | 0 | 0 | 0 | 0 | 0 |
| ALKOMA | 19 | 14 | 73.7 | 3 | 15.7 | 1 | 5.3 | 1 | 5.3 |
| OMBARO | 26 | 25 | 96.2 | 1 | 3.8 | 0 | 0 | 0 | 0 |
| ALTEWSHA | 12 | 8 | 66.7 | 3 | 25 | 1 | 8.3 | 0 | 0 |
| ALWAHA | 33 | 30 | 90.9 | 3 | 9.1 | 0 | 0 | 0 | 0 |
| UM-KADAD | 14 | 14 | 100 | 0 | 0 | 0 | 0 | 0 | 0 |
| ALLAYEED | 33 | 33 | 100 | 0 | 0 | 0 | 0 | 0 | 0 |
| ALSERAF | 21 | 20 | 95.2 | 1 | 4.8 | 0 | 0 | 0 | 0 |
| DAR-ALslam | 41 | 27 | 65.9 | 11 | 26.8 | 3 | 7.3 | 0 | 0 |
| TAWELA | 56 | 49 | 87.5 | 7 | 12.5 | 0 | 0 | 0 | 0 |
| KUTOM | 40 | 38 | 95 | 2 | 5 | 0 | 0 | 0 | 0 |
| MALET | 17 | 17 | 100 | 0 | 0 | 0 | 0 | 0 | 0 |
| KORMA | 52 | 47 | 90.4 | 5 | 9.6 | 0 | 0 | 0 | 0 |
| KABKABYA | 10 | 9 | 90 | 1 | 10 | 0 | 0 | 0 | 0 |
| ALTENA | 33 | 30 | 90.9 | 3 | 9.1 | 0 | 0 | 0 | 0 |
| KARNOY | 17 | 15 | 88 | 0 | 0 | 2 | 12 | 0 | 0 |
| Total | 666 | 579 | 86.9 | 70 | 10.5 | 13 | 2 | 4 | 0.6 |

## Discussion

HbD - known as Punjab or Los Angeles - is underexplored, especially in Sudan, where this recent study about its prevalence was carried out. Moreover, Hb D presents in considerable geographic distribution and is associated with Hb S, forming a heterozygous composite with peculiar clinical severity. Allocation of Hb D-Punjab in Punjab region, Northwest Indian and Gujarat region with an estimated frequency of 2.0%.[7] Globally Hb D is occurs in variety of countries include Italy,[8] Turkey,[9] Belgium,[10] Austria,[11] Xinjiang province, northwestern China,[12,13] Caucasian individuals in England and the United States,[6] British family of Spanish and Austrian extraction,[14] a mulatto child of an English mother[15] and in the American Indian.[16] The percentage of worldwide prevalence of Hgb D was recorded in the American Negro 0.4%, the Algerian Moslems 2.0% and Sikhs of North-Central India 2.0.[17]

In our current study 83 (12.5%) of participants had the genotype Hb SS and Hb AS, from these cases, 70 (10.51%) had the AS gene and the remaining 13 (1.9 5%) had the SS gene. 4 (0.6%) had Hb AD in four tribes of Northern Darfur state. This abnormality was the first time to be recorded in Northern Darfur state combined with prevalence of Hb SS and Hb AS. Gibreel et al.[18] reported that 2 (3.39%) % with Hb D and 12 (20.34%) showed patterns consistent with sickle cell disease (SS) of 209 participants in Eastern Beja tribes- Sudan; this with our resent study indicating that Sudan is a diverse country with a complex population originating from different ethnic groups created from intermarriage and social interaction status. The most important discover was that a number of reports of the Hb D trait have appeared in connection with family studies of patients with sickle cell- Hb SS disease[6,14] which was in line with our current study.

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## Conclusion

The prevalence of HbD variant was 4 (0.6 %) in 666 participants from four western Sudanese tribes, beside reported of Hb SS and Hb AS.

## Recommendations

Further exploration into Hb D variants in Sudan is needed to understand the various forms of Hb variants in diverse ethnic and locations.

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## Conflicts of interest

The authors declare no conflicts of interest.

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