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Study of Sickle Cell Syndromes in the Population of the Region of Batna

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***Abstract*—**Sickle cell anemia is a recessive genetic disease caused by the presence in the red blood cell, of abnormal hemoglobin called hemoglobin S. It results from the replacement in the beta chain of the acid glutamic acid by valin at position 6. Topics may be homozygous (SS) or heterozygous (AS) most often asymptomatic. Other mutations result in compound heterozygous: - Synthesis of hemoglobin C mutation in the sixth leucin codon (heterozygous SC); - ß-thalassemia (heterozygous S-ß thalassemia). SS homozygous, heterozygous SC and S- ß -thalassemia are grouped under the major sickle cell syndromes.

To make a laboratory diagnosis of hemoglobinopathies in a portion of the population in region of Batna, our study was conducted on 115 patients with suspected sickle cell anemia, all cases have benefited from hematological tests as blood count (count RBC, calculated erythrocyte indices, MCV and MCHC, measuring the hemoglobin concentration) and a biochemical test in this case electrophoresis CAPILLARYS HEMOGLOBIN (E).

The results showed:

27 cases of sickle cell anemia were found on 115 suspected cases, 73,03% homozygous sickle cell disease and 59,25% sickle cell trait. Finally, the double heterozygous S/C, represent the incidence rate of 3, 70%.

***Keywords*—**Hemoglobin, sickle cell syndromes, laboratory diagnosis

I. Introduction

SICKLE cell anemia is a genetic disorder of hemoglobin (Hb) that is transmitted as an autosomal recessive trait. The disease results from a mutation of the sixth codon of the gene beta globin. The mutation causes the synthesis of abnormal hemoglobin, HbS [1]. The polymerization of deoxygenated HbS to the state is causing a chronic hemolytic anemia and vaso-occlusive phenomena [2]. The disease is very common in populations of African sub-Saharan Africa. Because of recent population movements that characterize our time, it exists today on every continent.The objective of our work is to seek some sickle cell syndromes most frequently in the population of the region of Batna.

II. Materials and Methods

A. Patients and methods

Our study was conducted in the Central Laboratory of Hematology University Hospital of Batna and the biochemistry laboratory Batna, about 115 subjects suspected (or anemia detected during a whole some family survey) of both sexes and of different ages from different regions of the wilaya of Batna. The study lasted nine months from October 2009 to June 2010.

The starting point for the laboratory diagnosis is based on major tests and reproducible:

- Hematological tests: complete blood count with red blood cell count, erythrocyte constants calculated: (Mean corpuscular volume: MCV), (Mean corpuscular hemoglobin: MCH). The measurement of concentration of hemoglobin (Hb).
- Biochemical test: electrophoresis CAPILLARYS HEMOGLOBIN (E)

B. Statistical Analysis

Data were analyzed by Graph Pad Prism 5. The statistical method used is the t test to compare means for student samples. The results are expressed as mean ± sem. Statistical difference was considered significant when $P < 0.05$.

III. Results and Discussion

A. Distribution of patients under different types of sickle cell syndrome

Our results confirm the existence of different types of sickle cell syndromes in the population of the region of Batna (Table 1), 27 patients were found among 115 subjects suspected, 10 patients homozygous sickle cell (S / S), 16 sickle cell patients heterozygous (A / S), a patient with a double heterozygous S / C. Direct detection of the capillary at 415 nm is used to define the relative concentrations (percentage) of each fraction.

TABLE I
DISTRIBUTION OF PATIENTS UNDER DIFFERENT TYPES OF SICKLE CELL SYNDROMES

Type of disease	Number	Percentage %
Homozygous Sickle Cell Disease(S/S)	10	37,03
Sickle cell trait (A/S)	16	59,25
Double heterozygous(S/C)	01	3,70
Total	27	100%

Different variants of migration zones (Z1 to Z15 identified) are identified on screen and on the record results. The portion of the mouse on the name of an area leads display a tooltip showing the potential of hemoglobin variants migrating in this area.During the detection of these different sickle cell syndromes, the system represents CAPILLARYS HEMOGLOBIN (E) images by different electrophoretic hemoglobin abnormalities detected.

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B. Study of the electrophoretic profile in a homozygous sickle cell disease:

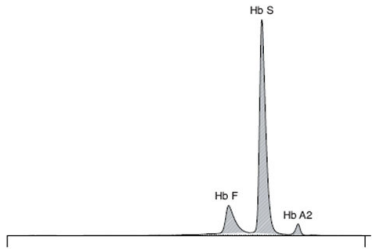

Fig. 1 Electrophoretic profile of a with homozygous sickle cell disease (HbS)

Sickle cell disease is caused by mutation of a glutamic acid beta chain (amino acid number 6) by a valin (neutral amino acid) [1]: it thus has an isoelectric point higher than in hemoglobin A [3]. It sover all negative charge is decreased the pH of the analysis: hemoglobin that migrates faster than hemoglobin A.

In the technique CAPILLARYSHEMOGLOBIN(E) in alkaline buffer, hemoglobin S migrates between fractions A and A2.

C. Change in number of red blood cells (106 / mm ³) in homozygous sickle cell disease

The results obtained show that there is a statistically significant decrease red cell counts (P <0.05) in patients (3.898 ± 0.2045) by contribution to controls (5.009 ± 0.1773) (see Table II). According [4] the solubility of the oxygenated form of hemoglobin S is identical to that of normal hemoglobin.

However as the deoxygenated hemoglobin S was less soluble than hemoglobin A. It can then was polymerized by a cooperative process and allow the stiffening, clumping and red cell sickliness.

D. Variation of the concentration of hemoglobin (g / dl) in patients homozygous sickle cell disease

TABLE II
VARIATION IN THE NUMBER OF RED BLOOD CELLS IN PATIENTS WITH HOMOZYGOUS SICKLE CELL DISEASE

State / Bioassay	Normal Control	SICK
Many of the red blood cells(106/ mm3)	5,009 ± 0,1773	3,898 ± 0,2045***

P*** <0.05 , mm 3 = millimeter cube

The results obtained show that there is a very highly significant decrease (p <0, 0001) the concentration of hemoglobin in these patients (6.910 ± 0.238) (see Table III) by contribution to controls (14.81 ± 0.1977). According [5]. The molecular mechanism is replaced by a valin for glutamic acid at position 6 of the globin chain that results in abnormal hemoglobin S (Hb S) causes dehydration of cells red and failure of their deformability (sickle cell) linked to the polymerization of HbS molecules in low oxygen.

E. Change in mean Corpuscular Volume (MCV) and mean Corpuscular Hemoglobin (MCH) (pg) in Patients Homozygous Sickle Cell Disease

TABLE III
VARIATION IN THE CONCENTRATION OF HEMOGLOBIN (G / DL) IN PATIENTS HOMOZYGOUS SICKLE CELL DISEASE

State / Bioassay	Normal Control	SICK
Concentration of Hb (g / dl)	14,81 ± 0,1977	6,910 ± 0,238 ***

P*** <0.0001 , g = gram, dl = deciliter.

The results obtained allow to observe a statistically significant decrease in mean corpuscular volume (p <0.0001) in these patients (62.88 ± 0.1256) by contribution to controls (91.24 ± 1.328) (see Table IV). Abnormalities functional membranes accompany sickling. Tostson was the first to show an increase in passive permeability of monovalent cations Na + and K + during deoxygenating of acute sickle cell blood.

With out changing neither the water content nor the total amount of monovalent cations [6].Our results show a significant decrease (P <0.0001) of the mean corpuscular hemoglobin in these patients (24.41 ± 0.5789) by contribution to controls (29.83 ± 0.4793) (see Table IV). Increasing the concentration of HbS favors the polymerized state, increases the proportion of polymerized hemoglobin in a given pO2 at the expense of free molecules [8].

F. Study of Electrophoretic Profiles in a Sickle Cell Trait

TABLE IV
CHANGE IN MEAN CORPUSCULAR VOLUME (MCV) AND MEAN CORPUSCULAR HEMOGLOBIN (MCH) (PG) IN PATIENTS HOMOZYGOUS SICKLE CELL DISEASE

State / Bioassay	Normal Control	SICK
MCV (pg)	29,83 ± 0,4793	24,41 ± 0,5789***
MCH (fl)	91,24 ± 1,328	62,88 ± 0, 1256 ***

P*** <0.0001 , pg = picogram.,fl = femtoliter.

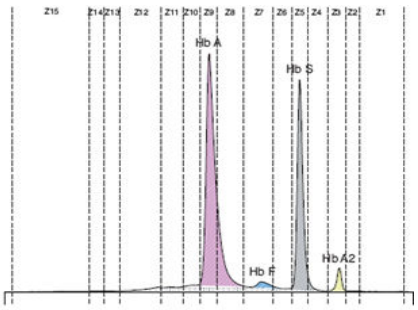

Fig. 2 Electrophoretic profile with varying heterozygous (HbS)

The sickle cell trait is characterized by the presence of Hb A [2]. It is then important to quantify the various fractions A, A2 and S [8] “Fig 2.”

G.Study of electrophoretic profile in a double heterozygous (S / C)

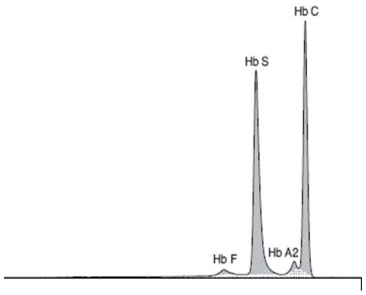

Fig. 3 Electrophoretic profile of a double heterozygous S / C

Compound heterozygotes (SC), in which the hemoglobin electrophoresis showed 50% and 50% Hb C HbS "Fig 3." combined with microcytic or normocytic anemia, moderate [9].

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