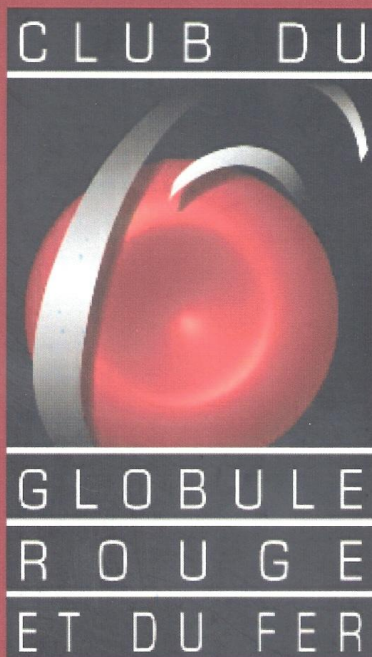


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G6PD DEFICIENCY IN A DIABETIC PATIENT WITH A TURNER SYNDROME

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G6PD deficiency is a genetic disorder linked to X chromosome (Xq28); males and homozygous females express complete deficiency, while in heterozygous females, enzymatic expression is variable and dependent on X lyonisation. We report a case of a Mauritanian G6PD deficient diabetic patient with a Turner syndrome.

Patients and Methods. D.C. a 13 year-old girl was investigated for short stature and growth velocity decrease, obesity, delayed puberty. The assessment found low growth hormone level, hypogonadism, normal thyroid state, noninsulin-dependent diabetes. A karyotypic analysis, completed by fluorescent in situ hybridization (FISH) was performed, as well as HbA1c determination, study of hemoglobin (Hb) and G6PD activity assay. The sequencing of G6PD gene exons was carried out on PCR products by ABI 3130.

Results. Cytogenetic analysis showed a 46,X,i (X)(q10) karyotype corresponding to an isochromosome for the q arm of X chromosome. HbA1c level (5.4%; N: 4-6) was discordant with the patient glycemic status. G6PD activity was reduced (<1UI/g Hb; N≥8). Hb profile was normal. G6PD gene sequencing showed two mutations 376G/202A (African variant A-) and 949A (G6PD Kerala-Kalyan, Indian variant).

Discussion. In this African diabetic patient, HbA1c level shows a discrepancy with its glycemic state and has prompted us to seek a cause of underestimation of the Hb glycation, explained by high G6PD deficiency. Because of the presence of an isochromosome Xq and a normal X chromosome (trisomy q arm and monosomy for p arm), the patient presents 3 G6PD deficient genes including two identical on one chromosome, situation compatible with both mutations identified and enzyme activity. HbA1c level, an index of diabetes follow-up, has to be used with caution in this G6PD deficient diabetic patient.

SOCIAL ENVIRONMENT AND EDUCATION OF CONGOLESE SICKLE CELL DISEASE PATIENTS: A SURVEY IN KINSHASA AND LUBUMBASHI

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We report the case of 288 patients with sickle cell disease (SCD) followed up clinically in Kinshasa (two groups of 92 and 116 patients) and in Lubumbashi (80). The mean age was of 11.4 years (6-20) and the sex ratio 1.2.

Most of them (58.6%) was from a family with 6 to 10 members and live in houses of 2-3 compartments and without ceiling. Two or more children use the same bed and only 11.2% use mosquito nets impregnated with insecticide. Most of them go to school on foot, 14.3% of them do it for more than 3 km. These conditions of the patients with SCD, associated to the clinical aspects affect negatively their school performances. Indeed, 86.3% in Lubumbashi and 65.2% in Kinshasa have not finished the elementary school and only 1.1% arrived at the university. Actually, the program of community education coupled with the early medical care seems to contribute to a better school application of the affected infants and allow them to be accepted in the society.

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SICKLE CELL DISEASE: THE BENIN EXPERIENCE

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Introduction. With 7 millions inhabitants, 3% population growth, Benin is considered as the most affected by hereditary hemoglobin abnormalities amongst all Benin Gulf countries. Hemoglobin (Hb) Hb S prevalence is 24% versus 9% for Hb C. The rate of sickle cells disease (SCD) in the population is 4%.

Management of the disease. SCD management began in the seventies in free out-patients consultations. Analgesics, antipyretics, non steroid anti-inflammatory drugs, antibiotics, blood transfusion, folic acid supplementation and hydration are usual treatments. SCD management got improvement since the National University Hospital (CNHU) created the Haematology Service in 1988. The SCD patients represent 65% of the work load and they consume 35% of blood collected by the Cotonou blood transfusion centre. Out of 519 patients hospitalized in two years, 63.6% were Hb SS and 14.8% Hb SC. Hb SS subjects were mainly hospitalized for anaemic syndrome (40.6%), hyperalgic attacks (34.5%) and infections (20.9%). Sustained follow-up increased patients' life quality. This results in a need of addressing their training, occupation and marriage issues. In 1991, patients were charged for the medical consultation; then paying professional services progressively reduces (60%) the Haematology Service attendance rate from 1991 to date.

Research. Traditional medicine, assessed through phytotherapy, seems to be only symptomatic. Other research topics concern psychosocial aspects, blood transmitted diseases, chronic complications, feasibility of physical activities as integration and self esteem factors. Documents are available to support fair information on SCD. SCD financial burden has been evaluated. A SCD Centre for children and pregnant women (1990) and a National Programme are available.

Associative activities. Associative activities are implemented to raise solidarity among patients, their relatives and the population. It's the main goal of Benin Sickle Cells Association (Association Béninoise de Lutte contre la

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Drépanocytose, ABLD) created in 1990 of which some achievements are: National Days of fight against SCD, information and systematic SCD screening campaigns, contribution to the federation of the National Sickle Cells Diseases Associations in Africa in 1996, called FALDA (Federation des Associations de Lutte contre la Drépanocytose en Afrique) including 14 countries. Lack of resources is the main bottleneck of ABLD. Conclusion. Sickle cells disease is still deadly. The only alternative in the absence of curative care is a precocious multidisciplinary and decentralized management of the disease and a tight follow-up of the patients.

SPLEEN SEQUESTRATION IN CHILDREN WITH SICKLE CELL DISEASE

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Introduction. Acute spleen sequestration is a major cause of morbidity and mortality in children with sickle cell disease. The aim of this study was to evaluate prevalence and risk factors of splenic sequestration in a cohort of Tunisian patients with sickle cell disease.

Patients and methods. We performed a retrospective study including 45 children among 194 patients with sickle cell anemia (20 homozygous SS, 25 double composite heterozygous S-β thalassemia). We defined splenic sequestration as an acutely enlarging spleen over 2 cm of steady state measurement with a fall in haemoglobin concentration over 20% of steady state with evidence of marrow activity (reticulocyte number >200000/mm³). All patient aged under 5 years was placed on short term transfusion program. Total surgical removal of the spleen was recommended after 2 acute splenic sequestrations and over the age of 5 years.

Results. The prevalence of splenic sequestration found in our study was 23%. The mean age and hemoglobin concentration during the first episode were respectively 41 months (range 3-124 months) and 4.9 g/dL (range 2.5-6.5 g/dL). Concomitant infection or painful episode were found in 7 cases (15%). 31 patients presented more than two episodes of spleen sequestrations (69%). 6 patients were placed on program of monthly transfusions (13%). Among these 6 patients 2 patients had at least a recurrence of splenic sequestration. Splenectomy was performed in 29 patients at a mean age of 5 years.

Conclusions. Splenic sequestration is a common finding in children with sickle cell disease. Surgical removal of the spleen remains the radical treatment after the age of 5 years.

DELAYED PUBERTY IN TUNISIAN THALASSEMIC PATIENTS

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Introduction. Delayed puberty in patients with thalassemia major has become an issue of interest in the last few years since life expectancy was increased by hypertransfusion and iron chelating therapy. The aim of this study was to evaluate the prevalence, risk factors and causes of delayed puberty in a cohort of Tunisian transfusion dependant thalassemic patients.

Patients and methods. Forty six patients (25 males, 21 females) took part in the study. For each patient were identified: Haematological parameters (age at first blood transfusion, first iron chelation, mean ferritinemia values). Blood sample was obtained from each patient in order to measure glycaemia, T₄, PTH, IGF₁ and hypothalamic-pituitary-gonadal function. MRI study was performed to evaluate iron overload in hypothalamic-pituitary area. Statistical analysis was carried out by using χ^2 method and Fisher's exact test.

Results. Among the 46 studied patients, 23(50%) developed delayed puberty. Delayed puberty is more frequent in boys: 15 of 25 boys (60%) than in girls: 8 of 21 girls (38%). Spontaneous induction of puberty was observed in 11 cases (4 females and 7 males). Hypogonadism was found in 9 patients. The risk factors for delayed puberty found in this study group were age of first iron chelation ($p=0.002$) and ferritinemia level ($p=0.03$).

Conclusions. Delayed puberty is very common in polytransfused thalassemic patients. Hypogonadism is the most frequent complication. Iron overload has been considered to be the major cause. It is mandatory to carefully monitor the pubertal development of these patients to detect abnormalities and to initiate appropriate iron-chelating agents before referring to substitutive hormones.

IDENTIFICATION OF A NOVEL DELETION-INSERTION IN THE β -GLOBIN GENE EXON 1 AT THE ORIGIN OF A MINOR β -THALASSAEMIA

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A minor β -thalassemia was diagnosed in a patient from Algeria in 1978. At the time, he presented a splenomegaly and a polycythemia (7.23.1012/L) without anemia (Hb 14 g/100 mL). There existed a microcytosis (61 fl) and a clear hypochromy (19.4 pg). Sideremy was normal. Hemoglobin electrophoresis of this patient showed the profile of a β thalassemia hétérozygote A/ β^0 : Hb