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 hemoglobinic 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. Antalgics, antipyretics, non steroid anti-inflammatory drugs, antibiotics, blood transfusion, folic acid supplementation and hydratation 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