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GENOTYPE AND PHENOTYPE CORRELATION IN GLUCOSE-6- PHOSPHATE DEHYDROGENASE DEFICIENCY

Thesis

Submitted for partial fulfillment
Of M.D degree in Clinical and Chemical Pathology

By

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جامعة القاهرة / كليه الطب
الدراسات العليا

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الطبيب / ريهام عبد العليم محمد حسن
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تحت عنوان : باللغة الانجليزية : Genotype and Phenotype Correlation in Glucose-6-phosphate dehydrogenase deficiency

: باللغة العربية : علاقة التوصيف الجيني بالتوصيف الإكلينيكي في مرضى نقص إنزيم الجلوكوز-6-فوسفات ديهيدروجينيز

بناء على موافقة الجامعة بتاريخ ٢٦/ ٢ / ٢٠٠٦ تم تشكيل لجنة للفحص والمناقشة للرسالة المذكورة أعلاه على النحو التالي :-

١. أ.د/ [illegible] أستاذ الباثولوجيا الإكلينيكية [illegible] عن المشرفين
٢. أ.د/ منى [illegible] القاضي أستاذ الأطفال كلية الطب [illegible] ممتحن داخلي
٣. أ.د/ [illegible] الشافعي أستاذ الباثولوجيا الإكلينيكية [illegible] ممتحن خارجي

بعد فحص الرسالة بواسطة كل عضو منفردا وكتابة تقارير منفردة لكل منهم انعقدت اللجنة مجتمعة في يوم الاثنين بتاريخ ٣ / ٤ / ٢٠٠٦ بقسم الباثولوجيا الإكلينيكية مدرج "ب" بكلية الطب – جامعة القاهرة وذلك لمناقشة الطالب في جلسة علنية في موضوع الرسالة والنتائج التي توصل اليها وكذلك الأسس العلمية التي قام عليها البحث .

قرار اللجنة : قبول الرسالة

توقيعات أعضاء اللجنة :-

المشرف الممتحن	الممتحن الداخلي	الممتحن الخارجي

ختم

ABSTRACT

G6PD: Glucose-6- phosphate dehydrogenase

Glucose-6- phosphate dehydrogenase deficiency, the most common human enzymopathy, is characterized by wide clinical, biochemical, and molecular heterogeneity. Over 400 variants of G6PD have been distinguished on the basis of biochemical characterization.

The aim of this study was to establish, whether a correlation existed between genotype and clinical phenotype in 30 G6PD deficient Egyptian pediatric patients, and to determine whether the presence of hematologic and molecular predictive factors could eventually condition the clinical phenotype, thus facilitating prevention and refining the prognosis.

We study the clinical and hematological data from the 30 patients.

Analysis for enzymatic activity and molecular typing were performed on all subjects, 19 subject (63.3%) showed a severe G6PD deficiency (residual enzymatic activity (REA) <10%) and 11 subjects (36.6%) showed a moderate G6PD deficiency (residual enzymatic activity (REA) =10-60%).

The Mediterranean variant was found in (46.7%), while the remaining (53.3%) (Non- Mediterranean) were negative to all 4 studied mutations (Mediterranean, Seattle, A- and Montalbano variants).

In our study, (14.2%) G6PD deficient patients with Mediterranean variant were asymptomatic, (85.7%) had an acute hemolytic crisis, of them (75%) hemolysis induced after ingestion of fava beans, while (16.6%) hemolysis induced after drug intake and (8.3%) hemolysis induced secondary to multiple reasons and in non-Mediterranean G6PD patients, (12.5%) were asymptomatic, (87.5%) presented by AHC, of them (92.8%) hemolysis induced after ingestion of fava beans and (7.1%) hemolysis induced by drug intake.

Enzymatic activity was shown to be a poor predictive parameter of acute hemolytic crisis and was not correlated with clinical features.

Subjects with Mediterranean variant had a more severe clinical phenotype which was not related to enzymatic activity.

Key words: G6PD deficiency, phenotype, genotype.

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