بسم الله الرحمن الرحيم

شبكة المعلومـــــات الجامعية

التوثيق الالكتروني والميكروفيلم

شبكة المعلومات الجامعية

جامعة عين شمس

التوثيق الالكتروني والميكروفيلم

﴿ قـــسـم ﴾

نقسم بالله العظيم أن المادة التي تم توثيقها وتسجيلها
علي هذه الأفلام قد اعدت دون آية تغيرات

يجب أن

تحفظ هذه الأفلام بعيداً عن الغبار

في درجة حرارة من 15 – 20 مئوية ورطوبة نسبية من 20-40 %

To be kept away from dust in dry cool place of
15 – 25c and relative humidity 20-40 %

بعض الوثائق الأصلية تالفة

بالرسالة صفحات

لم ترد بالأصـــل

GENETIC SCREENING FOR G6PD MEDITERRANEAN MUTATION IN SOME EGYPTIAN CHILDREN WITH G6PD DEFICIENCY

Thesis

Submitted in partial fulfillment in Master Degree in

Clinical and Chemical Pathology

By

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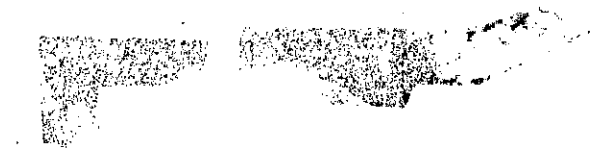

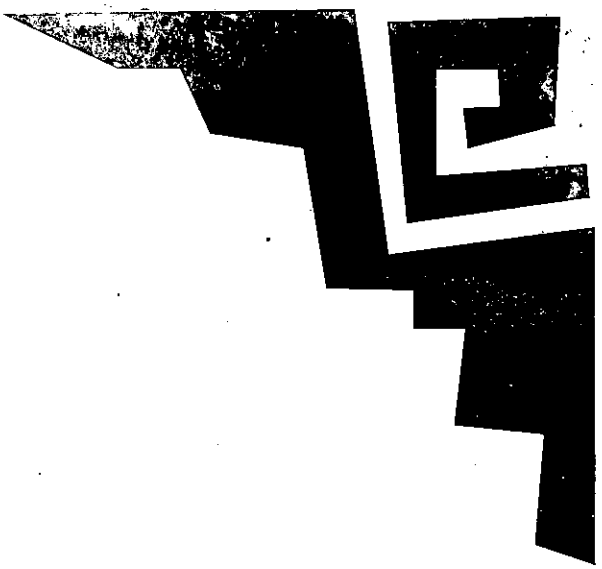

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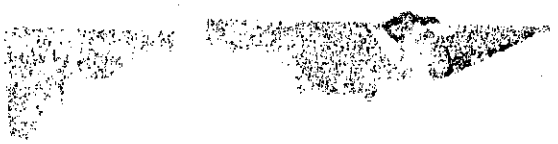

To :

MY FAMILY

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ABSTRACT

Glucose-6-phosphate dehydrogenase deficiency, the most common human enzymopathy, is characterized by a wide clinical, biochemical and molecular heterogenity. This study was conducted on a group of 21 Egyptian pediatric G6PD deficient patients. Quantitative assay of G6PD enzyme and estimation of its electrophoretic mobility was performed prior to molecular analysis. The frequency of the G6PD Mediterranean mutation at nucleotide $563^{C\rightarrow T}$ among the studied group was investigated. Exons 6 and 7 of the G6PD gene were amplified using PCR reaction followed by digestion of the amplified fragment using the Mbo II restriction enzyme. The $563^{C\rightarrow T}$ mutation was found in only 28.6% (6/21) of studied cases. G6PD variants present in Egypt, thus require further molecular characterization through analysis of the whole G6PD coding sequence.

Key Words *:*

G6PD - haemolytic anaemia - favism