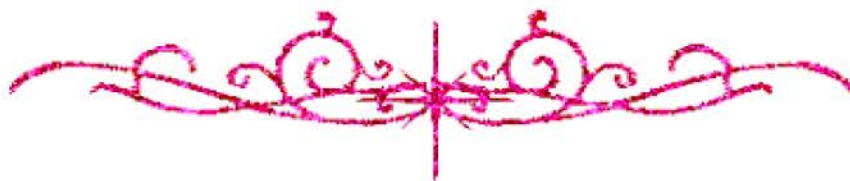


بسم الله الرحمن الرحيم





شبكة المعلومات الجامعية التوثيق الالكتروني والميكروفيلم



جامعة عين شمس

التوثيق الإلكتروني والميكروفيلم

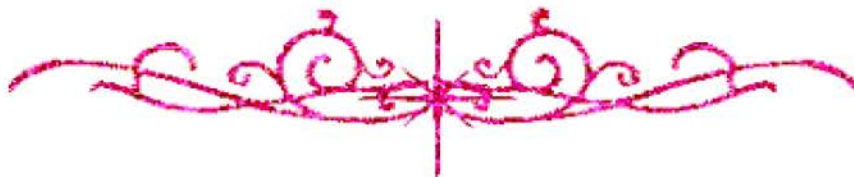
قسم

نقسم بالله العظيم أن المادة التي تم توثيقها وتسجيلها
علي هذه الأقراص المدمجة قد أعدت دون أية تغيرات



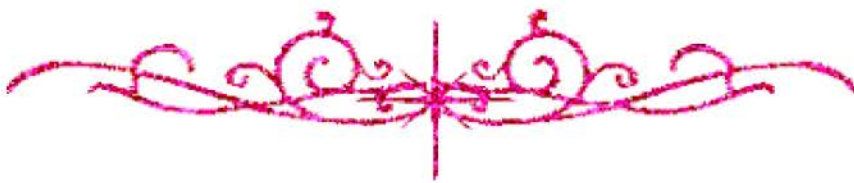
يجب أن

تحفظ هذه الأقراص المدمجة بعيدا عن الغبار



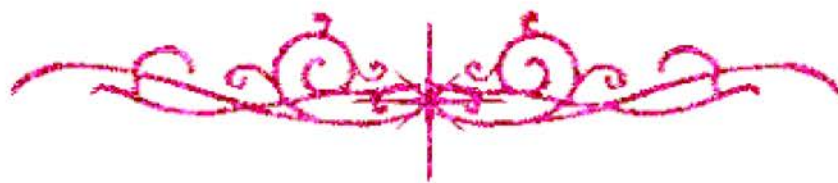


بعض الوثائق الأصلية تالفة





بالرسالة صفحات
لم ترد بالأصل



THE DIFFERENTIAL EFFECT OF MANAGEMENT COMPONENTS AMONG CLASSIC PKU PATIENTS



Thesis

68964

*Submitted For The Partial Fulfilment Of Master Degree In
Medical Human Genetics*

616.042
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2002

بسم الله الرحمن الرحيم

"سبحانك لا علم لنا إلا ما علمتنا

إنك أنت العليم الحكيم"

صدق الله العظيم

سورة البقرة آية ٣٢

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List of Abbreviations

AD	Autosomal Dominant
AR	Autosomal Recessive
ASUMGC	Ain Shams University Medical Genetics Center
BBB	Blood brain barrier
BH ₄	Tetrahydrobiopterin
cDNA	Complementary Deoxyribonucleic acid
DGGE	Denaturing gradient gel electrophoresis
DHPR	Dihydropteridine reductase
DNA	Deoxyribonucleic acid
EEG	Electroencephalogram
F	Female
FH	Family history
GTP	Guanosine triphosphate
HPA	Hyperphenylalaninemia
IEM	Inborn Errors of Metabolism
IQ	Intelligence Quotient
Kb	Kilobase
KD	Kilodalton
LNAA	Large Neutral Amino acids
M	Male
MC	Microcephaly
MR	Mental retardation
mRNA	Messenger ribonucleic acid
NPN	Non protein nitrogen
PAH	Phenylalanine hydroxylase
Pat	Paternal
PCR	Polymerase chain reaction

Phe	Phenylalanine
PKU	Phenylketonuria
PPH4S	6-pyruvyl tetrahydropetrin synthetase
RFLP	Restriction fragment length polymorphism
SDC	Strict Dietary Control
SDS	Standard Deviation Score
Se	Selenium
TH	Tyrosine hydroxylase
Tyr	Tyrosine
VNTR	Variable number tandem repeat

Approximate conversions for phenylalanine concentrations

$$1\text{mg /dL} = 60\ \mu\text{mol/L}$$