

**SCREENING FOR BIOTINIDASE DEFICIENCY
IN NEONATES AND SOME PATIENTS
WITH NEUROLOGIC DISORDERS**

THESIS

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قَالُوا سُبْحَانَكَ لَا عِلْمَ لَنَا إِلَّا مَا
عَلَّمْتَنَا إِنَّكَ أَنْتَ الْعَلِيمُ الْحَكِيمُ
سُبْحَانَكَ اللَّهُ الْعَلِيمُ
البقرة - ٢٢



*I am greatly indebted
to my parents and husband
and deep thanks to
my children*

***THIS THESIS HAS NOT BEEN SUBMITTED FOR A
DEGREE AT THIS OR ANY OTHER UNIVERSITY.***

RANDA MOHAMED SALAH EL-DIN ABD EL-SALAM

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ABBREVIATIONS

Ala	Alanine
Arg	Arginine
BPAB	Biotinyl-p-aminobenzoate
C.T.	Computerized tomography
CoA	Co enzyme A
CRM	Cross-reacting material
Cys	Cysteine
CyS-Scy	Cystine
DNPH	Dinitrophenyl hydrazine
E. Coli	Escherichia Coli
EDTA	Ethylene diamine tetracetate
Glu	Glutamic acid
Gly	Glycine
HCL	Hydrochloric acid
His	Histidine
IEF	Isoelectric Focusing gel electrophoresis
Leu	Leucine
LMCD	Late onset, biotin responsive, multiple carboxylase deficiency
MCD	Multiple carboxylase deficiency
N.R.C	National Research Center
PABA	P-aminobenzoic acid

phe	Phenylalanine
Pro	Proline
S & S	(Schleucher and Schuell) filter paper card
S.D.	Standard Deviation
Ser.	Serine
STLC	Serum thin layer chromatography
Thr	Threonine
Trp	Tryptophan
Tyr	Tyrosine
ULTC	Urine Thin layer chromatography
Val	Valine
\bar{X}	Arithmetic means

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CHAPTER I

A) INTRODUCTION