Metabolic Neonatal Emergencies
Diagnosis and Management

Essay

Submitted for Partial Fulfillment of Master Degree in Pediatrics

By

Enas Hamza El-Sherbiny

MBBCh

Supervisors

Prof. Dr. Mohamed Ashraf Abd Elwahed

Professor of Pediatrics

Ain Shams University

Dr.Ola Galal Badr Eldin

Lecturer of Pediatrics

Ain Shams University

Ain Shams University

Faculty of medicine

2009

بسم الله الرحمن الرحيم

سبحانك لا علم لنا إلا
ما علمتنا إنك أنت
العليم الحكيم

(سورة البقرة آية ٣٢)

Acknowledgement

First of all, thanks to Allah the most merciful and the most beneficent for giving me the strength to complete this work and for giving me such a wonderful supporting family. I am greatly honored to express my endless gratitude to ***Prof. Dr. Mohamed Ashraf Abd Elwahed*** *Professor of Pediatrics Ain Shams University. who kindly offered me the generous encouragement ,support, sincere concern and directions ,really I am lucky to be under his supervision.*

Words fail me when I come to express endless gratitude to ***Dr.Ola Galal*** *Lecturer of Pediatrics Ain Shams University for her help and expert supervision. She has been very kind ,generous with scientific advice and a strong motivating force to produce an accurate work. and without her help achievement of this work wouldn't be possible. for here I will remain humbly grateful.*

Dedication

To my mother,my words stand short of my supreme gratitude for every thing she offered me throughout my lif .God bless her.

To my father's soul, mercy be upon him.

My deepest hearty feeling to my husband for his unlimited support,active cooperation and patience,which give me a great motivate throughout this work .

To my lovely daughter and son ,I love you

List of contents

Page

List of abbreviations

ACTH	Adrenocorticotropic hormone
AD	Autosomal dominant
AF	Aminotic fluid
AL	Argininosuccinic lyase deficieny
alkyl DHAP	Alkyl Dihydroxyacetonephosphate acyltransferase
AR	Autosomal recessive
AS	Argininosuccinic acidemia
ATP	Adenosine triphosphate
BCKD	Branched-chain alpha-keto acid dehydrogenase
BUN	Blood urea nitrogen
CBC	Compelet blood count
Cbl	Cobalamin
CDG	Congenital disorders of glycosylation
CDGS	Carbohydrate-deficient glycoconjugate syndromes
CG-MS	Chromatography-mass spectrometry
CK	Creatine kinase
CNS	Central nervous system
CoA	Coenzyme A
CPS	Carbamyl phosphate synthetase
CPT	Carnitine palmitoyltransferase
CSF	Cerebrospinal fluid
D-BP	D-bifunctional protein
DHAPAT	Dihydroxyacetonephosphate acyltransferase

DHCR7	7-dehydrocholesterol-delta 7-reductase
DTRs	Deep tendon reflexes
EMG	Electromyographic
ER	Endoplasmic reticulum
ERG	Electroretinogram
ERT	Enzyme replacement therapy
ETF	Electron transfer flavoprotein
FAO	Fatty acid oxidation disorders
FDPase	Fructose 1,6-diphosphatase
FFP	Fresh-frozen plasma
GABA	Gama amino butyric acid
GAD	Glutamic acid decarboxylase
GALT	Galactose-1-phosphate uridyltransferase
GCSF	Granulocyte colony–stimulating factor
GDP-mannose	Guanosine diphosphomannose
GFR	Glomerular filtration rate
GSD	Glycogen storage disease
HF	Hydrops fetalis
HHH	Hyperammonemia-hyperornithinemia-homocitrullinemia
HPA	Hyperpipecolic academia
HSCT	Hematopoietic stem cell transplant
IDMs	Infants of diabetic mothers
IEF	Isoelectrofocusing
IEM	Inborn error of metabolism

IEMs	Inborn errors of metabolism
Ile	Isoleucine
IRD	Infantile Refsum disease
ISSD	Infantile sialic acid storage disease
IUGR	Intrauterine growth restriction
IVA	Isovaleric acidemia
L/P	lactate/pyruvate
LCHAD	Long-chain 3-hydroxyacyl-CoA dehydrogenase
LCHAD,	long-3-hydroxyacyl-CoA dehydrogenase
LEU	Leucine
LPI	Lysinuric protein intolerance
LSD	Lysosomal Storage Diseases
MCA	Multiple congenital anomalies
MCAD	Medium-3-hydroxyacyl-CoA dehydrogenase
MELAS	Mitochondrial encephalopathy myopathy, lactic acidosis, and stoke like episodes
MERRF	Myclonus epilepsy, with ragged-red fibers
Met	Methionine
MMA	Methylmalonic acidemia
MPS	Mucopolysaccharidoses
MR	Mental retardation
MRI	Magnetic resonance imaging
MRS	Magnetic resonance spectroscopy
MSUD	Maple syrup urine disease

MTP	Mitochondrial trifunctional protein
NAGS	N-Acetylglutamate synthetase
NALD	Neonatal adrenoleukodystrophy
NG	Nasogastric
NGT	N-acetylglucosaminyltransferase
NICCD	Neonatal intrahepatic cholestasis caused by citrin deficiency
NKH	Nonketotic hyperglycinemia
NP-C	Niemann-Pick disease type C
NPD	Niemann-Pick disease
NTBC	(2-(2-nitro-4-trifluoromethylbenzoyl)-1,3-cyclohexanedione
OTC	Ornithine transcarbamylase
PBD	Peroxisomal biogenesis disorders
PDC	Pyruvate dehydrogenase complex
PMI	Phosphomannose isomerase
PMM	Phosphomannomutase
PPA	propionic acidemia
PRBCs	packed red blood cells
RCDP	Rhizomelic chondrodysplasia puncta
SCHAD	Short -3-hydroxyacyl-CoA dehydrogenase
SCOT	Succinyl-CoA: 3-ketoacid CoA transferase
SGA	Small-for gestational age
SIDS	Sudden infant death syndrome

SLOS	Smith-Lemli-Opitz syndrome
THAN	Transient hyperammonemia of the newborn
THR	Threonin
TPGS	alpha-tocopherol polyethylene glycol succinate
TTN	Transient tyrosinemia of the newborn
UCD	Urea cycle disorders
UDP	Uridine diphosphate
US	United States
VAL	valine
VLCFA	Very long chain fatty acid
ZWS	Zellweger syndrome

List of tables

List of figures

Introduction

More than 500 inborn errors of metabolism (IEMs) have been identified. Individually rare IEM are relatively common collectively and early diagnosis and treatment may reverse acute symptoms and prevent chronic damage. Accurate diagnosis can help in future family planning **(Yuen, 2004).**

IEM present a particular challenge for the general paediatrician and most generalists, as they have a little experience with their diagnosis and management. In addition, the clinical presentation of the diseases often mimics common acquired conditions, especially infections, intoxications, and some nutritional deficiency disorders. The difficulty is increased as the clinicians often think about the chemical physiology of IEM as most textbooks are organised biochemically according to different aspects of metabolism **(Clarke, 2004).**

Aim of the work

The aim of this review was to demystify the clinical challenge in suspection, diagnosis and treatment of IEMs by presenting a stepwise approach that facilitates the early recognition of IEMs and guides further investigations.