بسم الله الرحمن الرحيم

MONA MAGHRABY

شبكة المعلومات الجامعية

التوثيق الالكتروني والميكروفيلم

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جامعة عين شمس

التوثيق الإلكتروني والميكروفيلم

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MONA MAGHRABY

Assessment of Mothers' Knowledge towards Care of Their Children Suffering from Methylmalonic Acidemia (MMA)

Thesis

Submitted for Partial Fulfillment of the Requirements
for Master Degree in Pediatric Nursing

By

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Zeinab Megahed Ramadan

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LIST OF ABBREVIATIONS

Abbreviations	Full Term
ABC	ATP-Binding Cassette.
ABG	Arterial Blood Gases.
ACMG	American College of Medical Genetics.
ACSF3	Acyl-CoA Synthetase family member 3.
AdoCbl	Adenosyl-Cobalamin.
ADP	Adenosine DI Phosphate.
ALDH6A1	Aldehyde Dehydrogenase 6 family, member A1.
ALT	Alanine Aminotransferase.
AST	Aspartate Transaminase.
ATP	Adenosine Triphosphate.
Bilirubin T/U	Bilirubin Total vis Unconjugated.
C0	Free Carnitine 0.
C2	Acetyl Carnitine 2.
C3	Propionyl Carnitine 3.
C4	Buyeryl Carnitine 4.
C4DC	C4-Dicarboxylic Acylcarnitine.
C16	Hexaadecanoyl Carnitine.
CBC	Complete Blood Count.
cblA	Cobalamin A deficiency.
cblB	Cobalamin B deficiency.
cblC	Cobalamin C deficiency.
cblD	Cobalamin D deficiency.
cblX	Cobalamin IX deficiency.
CI–	Chloride.
CMAMMA	Combined Malonic and Methylmalonic Aciduria.
CNS	Central Nervous System.
CoA	Coenzyme A.
CSF	Cerebrospinal Fluid.
CVS	Chorionic villus Sampling.
DNA	Deoxyribonucleic Acid.
EC	E-Cadherin.
ECG	Electro- Cardio Gram.

Abbreviations	Full Term
EKG	German Spelling- Electrocardiogram.
ESRD	End-Stage Renal Disease.
D-MMCoA	D-Methylmalonyl-CoA.
DNA	Deoxyribonucleic Acid.
DXA	Dual-energy X-ray Absorptiometry.
FAO	Food and Agriculture Organization of the United Nations.
GC	Gas Chromatography.
GC-MS	Gas Chromatography and Mass Spectrometry.
GCSF	Granulocyte-Colony Stimulating Factor.
GFR	Glomerular Filtration Rate.
GH	Growth Hormone.
HCFC1	The light purple striped box indicates cblX deficiency.
HGMD	Human Gene Mutation Database.
IQ	Intelligent Quotient.
IQs	Intelligence Quotients.
K+	Potassium.
LHON	Leber Hereditary Optic Neuropathy.
LMBRD1	LMBR1 Domain Containing 1.
MA	Malonic Acid.
MCEE	Methylmalonyl-CoA Epimerase.
MCM	Methylmalonyl-CoA Mutase.
MCR	Methylmalonyl-CoA Racemase.
MeCbl	Methyl-Cobalamin.
MMA	Methylmalonic Acidemia.
MMAA	Metabolism Of Cobalamin Associated A.
MMAB	Metabolism Of Cobalamin Associated B.
MMACHC	Metabolism Of Cobalamin Associated C.
MMADHC	Metabolism Of Cobalamin Associated D.
MMSA	Methylmalonic Semialdehyde Acid.
MMSDH	Methylmalonate Semialdehyde Dehydrogenase.
MMUT	Methylmalonyl-CoA Mutase.
MRI	Magnetic Resonance Imaging.

Abbreviations	Full Term
MS	Mass Spectrometry.
MS/MS	Mass Spectrometry/ Mass Spectrometry.
MTHFR	Methylenete Trahydro Folate Reducta.
MUT	Methylmalonyl-CoA Mutase.
NBS	Newborn Screening.
Na+	Sodium.
NCG	N-Carbamyl Glutamate.
NHGRI	National Human Genome Research Institute.
NIH	National Institute of Health.
OAs	Organic Acid Disorders.
OH	Hydroxo.
OH-Cbl	Hydroxo- Cobalamin.
PA	Propionic Acidemia.
PCC	Propionyl-CoA Carboxylase.
PGD	Preimplantation Genetic Diagnosis.
Q10	Coenzyme Q10.
QT	Qualification Test usually use for "QT segment in ECG".
QTc	QT orrected for heart rate.
SAM	S-Adenosyl Methionine.
SCD	Stearoyl-CoA Desaturase.
SCS-A	Succinyl-CoA Succinate.
SUCL	Succinyl-CoA Ligase.
SUCLA2	Succinate-CoA Ligase ADP-Forming Subunit Beta.
SUCLG1	Succinate-CoA Ligase GDP/ADP-Forming Subunit Alpha.
TMS	Transcranial Magnetic Scan.
TPN	Total Parenteral Nutrition.
US	United State.
UNU	United Nations University.
UT	Urea Transporter.
WHO	World Health Organization.

Abstract

Methylmalonic acidemia comprises a series of autosomal recessive inherited disorders of organic acid metabolism. Primary defect lies in methymalonyl-CoA mutase (MCM) or its cofactor, adenosyl-cobalamin. **Aim:** This study aimed to assess mothers' knowledge towards care of their children suffering from Methylmalonic acidemia. **Design:** A descriptive study was used. **Setting:** This study was conducted at the Specialized Genetic Outpatient Clinic at Pediatrics' Hospital affiliated with Ain Shams University Hospitals. **Subject:** A purposive sample which included 50 mothers accompanying their children with MMA. **Tools:** two tools were used**; I:** Interview questionnaire format to assess the characteristics of the study subjects, mothers' knowledge about methylmalonic acidemia, medical history of children, physical and psychological children's health needs and problems. **II:** Mothers' total reported practices related to care of their children suffering from Methylmalonic acidemia. **Results**: This study revealed that the mean age of studied mothers was 25 ± 6.6 years and the mean age of studied children's was 4.8 ± 2.49 years. Less than two thirds of studied mothers had unsatisfactory knowledge about Methylmalonic acidemia; as well more than two thirds of them had inadequate total reported practice. **Conclusion:** This result concluded that, less than two thirds of studied mothers had unsatisfactory total knowledge and more than two thirds of them had inadequate total reported practices about methylmalonic acidemia. There were statistical significant differences between all mothers' characteristics and their total knowledge and total reported practice regarding care of their children suffering from Methylmalonic acidemia. **Recommendation:** Teaching programs for families having children suffering from Methylmalonic acidemia is important to raise their knowledge and practices about the disease, its management and prevention of complications. Future research should be focused on the risk health problems among children suffering from Methylmalonic acidemia.

Keywords: Methylmalonic Academia, Knowledge, Metabolism, Dietary Restrictions, Problems, Mothers, Children.