



شبكة المعلومات الجامعية  
التوثيق الإلكتروني والميكروفيلم

# بسم الله الرحمن الرحيم



**MONA MAGHRABY**



شبكة المعلومات الجامعية  
التوثيق الإلكتروني والميكرو فيلم



# شبكة المعلومات الجامعية التوثيق الإلكتروني والميكرو فيلم



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التوثيق الإلكتروني والميكروفيلم

# جامعة عين شمس

## التوثيق الإلكتروني والميكروفيلم

### قسم

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**MONA MAGHRABY**

# **Assessment of Mothers Care towards their Children Suffering from Branched Chain Keto-Aciduria (BCKA)**

Thesis

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

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## Abstract

Branched-chain Keto-aciduria (BCKA) known as Maple Syrup Urine Disease (MSUD) is an aminoacidopathy due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. This defect leads to particularly toxic to the central nervous system. **Aim:** This study aims to assess mothers care for children having BCKA, through assess the mother's knowledge and self-reported practices toward caring for their children with BCKA. **Design:** A descriptive study. **Sampling:** A purposive sample which included 100 mothers accompanying their children with BCKA. **Setting:** This study was conducted at Specialized Genetic Outpatient Clinic at Children's Hospital affiliated to Ain Shams University Hospital. **Tools:** **Tool I:** A structure interview questionnaire format to assess characteristics of the study children and their mothers, medical history of children with BCKA, physical and psychological children's health needs and problems, mother's knowledge about BCKA. **Tool II:** Mothers self-reported practices related to care of their children with BCKA. **Tool III:** Medical records. **Results:** This study revealed that the mean age of studied children's was  $3.82 \pm 2.97$  years and the mean age of studied mothers was  $33.8 \pm 1.35$  years. Meanwhile about three quarters of studied mothers had unsatisfactory knowledge about BCKA, as well more than two thirds of them had poor total reported practice. The highest total knowledge and total self reported practices mean score are for mother whose young age, high education and living in urban area. **Conclusion:** This result concluded that majority of mothers had unsatisfactory total knowledge and poor total self-reported practices about branched-chain keto-aciduria. There were highly significant relation between the mother's characteristics and their care for children suffering from BCKA. **Recommendation:** Teaching program for families having children with BCKA is important to raise their knowledge about the disease, its management and prevention of complications. Future research should be focused on measuring the prevalence of BCKA in Egypt and investigating the risk health problems among mothers of children with BCKA.

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**Keywords:** Branched Chain Keto-Aciduria, Maple syrup urine disease, Problems, Mothers, Children, Care, Amino acids.

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## List of Abbreviations

Abbreviations	Full terms.
<b>AA</b>	: Amino Acid
<b>ADHD</b>	: Attention-Deficit/Hyperactivity Disorder
<b><math>\alpha</math>kIC</b>	: Alpha-Ketoisocaproic
<b>ASD</b>	: Autism Spectrum Disorder
<b>ASL</b>	: Argininosuccinate Synthetase
<b>BCAAs</b>	: Branched Chain Amino-Acids
<b>BCHA</b>	: Branched-Chain Hydroxy Acids
<b>BCKA</b>	: Branched Chain Keto-Aciduria
<b>BCKAD</b>	: Branched-Chain $\alpha$ -Ketoacid Dehydrogenase
<b>BCKD</b>	: Branched-Chain $\alpha$ -Ketoacid Dehydrogenase
<b>BCKDC</b>	: Branched-Chain $\alpha$ -Ketoacid Dehydrogenase Complex
<b>BCKDHA</b>	: Branched-Chain $\alpha$ -Ketoacid Dehydrogenase E1 Alpha polypeptide
<b>BCKDHB</b>	: Branched-Chain $\alpha$ -Ketoacid Dehydrogenase E1 Beta polypeptide
<b>BMI</b>	: Body Mass Index
<b>BDI</b>	: Beck Depression Inventory Scale
<b>CoA</b>	: Co-enzyme Aminoacid
<b>CPS</b>	: Carbamyl Phosphate Synthetase
<b>CSF</b>	: Cerebrospinal Fluid
<b>DBT</b>	: Dihydrolipoamide Branched Chain Transacylase
<b>DLDD</b>	: Dihydrolipoamidodehydrogenase deficiency
<b>DNPH</b>	: Dinitrophenylhydrazine
<b>DRI</b>	: Daily Requirements Intake
<b>EEGs</b>	: Electroencephalograms
<b>ESMSUD</b>	: European Society of Maple Syrup Urine Disease

<b>GC-MS</b>	: Gas Chromatography-Mass Spectrometry
<b>GLU</b>	: Glutamate
<b>GMP</b>	: Glycomacropeptide
<b>GTP</b>	: Guanosine Triphosphate
<b>HCL</b>	: Hydrochloric acid
<b>Hrs</b>	: Hours
<b>IEM</b>	: Inborn Error of Metabolism
<b>IMDs</b>	: Inherited Metabolic Disorders
<b>IDSA</b>	: Infectious Diseases Society of America
<b>ILE</b>	: Isoleucine
<b>ISO-LEU</b>	: Isoleucine
<b>IQ</b>	: Intelligence quotient
<b>KIC</b>	: $\alpha$ -Ketoisocaproic acid
<b>KIV</b>	: $\alpha$ -Ketoisovaleric acid
<b>KMV</b>	: $\alpha$ -Keto- $\beta$ -methylvaleric acid
<b>LEU</b>	: Leucine
<b>LNAA</b>	: Large Neutral Amino Acids
<b>MCQ</b>	: Multiple Choice Questions
<b>MMA</b>	: Methyl Malonic Acidemia
<b>MMAA</b>	: Methyl Malonic Aciduria type A porotein
<b>MMAB</b>	: Methyl Malonic Aciduria type B porotein
<b>MMADHC</b>	: Methyl Malonic Aciduria and Homocystinuria type D porotein
<b>MCEE</b>	: Methylmalonyl CoA mutase
<b>Mmol</b>	: Millimoles
<b>MS</b>	: Mass Spectrometry
<b>MUT</b>	: Mutations
<b>MSUD</b>	: Maple Syrup Urine Disease
<b>MRI</b>	: Magnetic Resonance Imaging
<b>NAGS</b>	: N-acetyl-Glutamate Synthetase
<b>NBS</b>	: Newborn Screening
<b>NIH</b>	: National Institute of Health
<b>NORD</b>	: National Organization for Rare Disorders
<b>OTC</b>	: Ornithine Transcarbamylase

<b>PAA</b>	: Plasma Amino Acids
<b>PRO</b>	: Protein
<b>TPN</b>	: Total Parental Nutrition
<b>SD</b>	: Standard Deviation
<b>STDs</b>	: Sexually Transmitted Disease
<b>SPSS</b>	: Statistical Package for Social Science
<b>TPN</b>	: Total parental nutrition
<b>Tyr</b>	: Tyrosine
<b>VAL</b>	: Valine
<b>U-MOL</b>	: Unit Moles
<b>UOA</b>	: Urine Organic Acids
<b>USA</b>	: United State of America
<b>WHO</b>	: World Health Organization
<b>QOL</b>	: Quality of Life

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