



شبكة المعلومات الجامعية  
التوثيق الإلكتروني والميكروفيلم

# بسم الله الرحمن الرحيم



**MONA MAGHRABY**



شبكة المعلومات الجامعية  
التوثيق الإلكتروني والميكروفيلم



# شبكة المعلومات الجامعية التوثيق الإلكتروني والميكروفيلم



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التوثيق الإلكتروني 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 تغييرات



## يجب أن

تحفظ هذه الأقراص المدمجة بعيدا عن الغبار



**MONA MAGHRABY**

# **Assessment of Lifestyle for Children with Phenylketonuria**

**Thesis**

*Submitted for Partial Fulfillment of Master  
Degree in Pediatric Nursing*

**By**

**Sohair Roby Abd Alghafar**

*(BSc. in Nursing, 2014)*

**Faculty of Nursing  
Ain-Shams University  
2020**

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## **List of Abbreviations (Abb.)**

<i>Abb.</i>	<i>Full term</i>
<b>AAV</b>	Adeno Associated Virus
<b>ADD</b>	Attention Deficit Disorder
<b>BH4</b>	Tetrahydrobiopterin
<b>CVS</b>	Chorionic Villus Sampling
<b>CPK</b>	Creatinine phosphor- kinase
<b>DTR</b>	Deep tendon reflex
<b>DM</b>	Diabetes Mellitus
<b>GMP</b>	Glycomacropetides
<b>HRQOL</b>	Health-related quality of life
<b>HPLC</b>	High Performance Liquid Chromatography
<b>HPA</b>	Hyperphenylalaninemia
<b>IQ</b>	Intelligence quotient
<b>LNAA</b>	Large Neutral Amino Acid
<b>MRI</b>	Magnetic Resonance Imaging
<b>MLD</b>	Metachromatic Leukodystrophy
<b>MOH</b>	Ministry of Health
<b>NCS</b>	Nerve conduction study
<b>PQOL</b>	Parental Quality of Life
<b>Phe</b>	phenylalanine
<b>PAL</b>	Phenylalanine Ammonia Lyase
<b>PAH</b>	Phenylalanine Hydroxylase
<b>PAHCDNA</b>	Phenylalanine Hydroxylase complementary deoxyribo nucleic acid

<i>Abb.</i>	<i>Full term</i>
<b>PKU</b>	Phenylketonuria
<b>PEG</b>	Poly Ethylene Glycol
<b>QOL</b>	Quality of Life
<b>rAAV</b>	Recombinant Adeno Associated Virus
<b>UK</b>	United Kingdom

## Abstract

Phenylketonuria (PKU) is an autosomal recessive disorder characterized by accumulation of phenylalanine (Phe) in blood and body fluids that is caused by defective Phe hydroxylase activity. **Aim** of the present study is to assess lifestyle for children with phenylketonuria. **Design:** A descriptive design was used in this study. **Setting:** conducted at genetic counseling clinic in basic health care unit in Fayoum affiliated to Ministry of Health (MOH). **Subject:** A purposive sample of included all children have PKU composed of 79 child accompanied by their mothers. **Tools:** 1) structural interviewing questionnaire sheet to assess demographic characteristics, and medical history 2) lifestyle assessment Questionnaire for children with PKU to identify current strengths of child's health, any risk factors. **Results:** (74.7%) of families were discover the disease by MOH screening programs. (92.4%) of children were made regular follow up.(78.5%) of children had follow up visits once monthly. 89.9% of children were not taking breast feeding. 62% of mothers had inadequate knowledge about the relation between breast feeding and the disease. **Conclusion:** There is a statistically significant correlation between compliance with the prescribed diet and growth problems. There is no statistically significant correlation between compliance with the prescribed diet and sleep problems. **Recommendations:** The current study recommended Periodic assessment of Children commitment to prescribed diet. Increase mother awareness about the importance of prescribed diet for children with Phenylketonuria

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**Key words:** life style, children, Phenylketonuria

## Introduction

Phenylketonuria (PKU) is an autosomal recessive disorder characterized by accumulation of phenylalanine (Phe) in blood and body fluids that is caused by defective Phe hydroxylase activity. PAH deficiency is classified as classical PKU (Phe  $\geq$  1200  $\mu\text{mol/L}$ ), mild PKU (Phe 600– $\leq$ 1200  $\mu\text{mol/L}$ ) or mild hyperphenylalaninaemia (Phe  $>$  600  $\mu\text{mol/L}$ ). The incidence of PKU varies according to ethnic background. In the UK, PKU affects about 1 in every 10,000 newborns of white European ancestry with around 70 babies born with PKU annually, suggesting that over 6000 people in the UK have PKU (*Williams, Mamotte, Burnett, 2018*).

Provisional diagnosis to be Metachromatic Leukodystrophy (MLD)/ Aminoaciduria/ Leigh syndrome, Advising for urine and serum amino acid test, thyroid hormone stimulating test, creatinine phosphor- kinase (CPK), nerve conduction study (NCS), serum lactate pyruvate tests. Physiotherapy and on anti-epileptic medications. Later when the child visited for the outpatient department with conditions of global developmental delay, right focal seizures, poor visual perception and not yet