بسم الله الرحمن الرحيم

MONA MAGHRABY

شبكة المعلومات الجامعية

التوثيق الالكتروني والميكروفيلم

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شبكة المعلومات الجامعية

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جامعة عين شمس

التوثيق الإلكتروني والميكروفيلم

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MONA MAGHRABY

Assessment of Lifestyle for Children with Phenylketonuria

Thesis

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

By

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Sohair Roby Abd Alghafa

List of Content

List of Tables

List of Figures

List of Abbreviations (Abb.)

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

Abb.	Full term
PKU	Phenylketonuria
PEG	Poly Ethylene Glycol
QOL	Quality of Life
rAAV	Recombinant Adeno Associated Virus
UK	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

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 N 1200 μmol/L), mild PKU (Phe 600–≤1200 μmol/L) or mild hyperphenylalaninaemia (Phe b 600 μmol/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