Mother's coping of Children Suffering from Phenylketonuria

Thesis

Submitted for Partial Fulfillment of Master Degree

In Community Health Nursing

By

Nema Rezk Abd El-Said Ibraheem

(B.Sc. Nursing, 1999)

Ain Shams University

Faculty of Nursing
Ain Shams University
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Supervised by

Dr. Magda Abd El-Sattar Ahmad

Assistant Professor of Community Health Nursing

Faculty of Nursing - Ain Shams University

Dr. Osama Kamal Zaki

Consultant of Medical Genetics

Faculty of Medicine - Ain Shams University

Dr. Ferial Fouad Melika

Lecturer of Community Health Nursing

Faculty of Nursing - Ain Shams University

Faculty of Nursing
Ain Shams University
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In the name of Allah the Most Gracious and most Merciful, for granting me the power accomplish this work.

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Contents

List of Abbreviations

ADHD : Attention-deficit/hyperactivity disorder

ASD : Autism spectrum disorder

BH4 : Cofactor tetrahydrobiopterin

CNS : Central nervous system

EEG : Electroencephalography

HPA : Hyperphenylalaninaemia

IQ : Intelligence quotient

mg/dl : Milligrams per deciliter

MS : Mass spectrometry

PAH : Phenylalanine hydroxylase

PHE : Phenylalanine

PKU : Phenylketonuria

THB : Tetrahydrobiopterin

Tyr : Tyrosine

µM/L : Micromoles per liter

NSPKU : The National Society for Phenylketonuria

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Abstract

Mother's coping of Children Suffering from Phenylketonuria
By
Nema Rezk Abd El-Said Ibraheem
Master Degree thesis
Faculty of Nursing - Ain Shams University

Phenylketonuria (PKU) is an inherited disorder in which the body lacks the enzyme needed to convert phenylalanine to tyrosine. It can damage the brain and nervous system if left untreated. ***Aim: this study*** aims to assess the mother's coping of children suffering from PKU. ***Study Design:*** *A* descriptive design was utilized in this study. ***Setting:*** the study was conducted in genetic clinic affiliated to pediatric hospital of Ain Shams University. ***Sample:*** A purposive sample composed of 125 children medically diagnosed with phenylketonuria and their mothers. ***Tools*****: 1st tool** an interviewing questionnaire to assess Socio-demographic data of the study children and their mothers, Current health needs and problems of children, mother's knowledge about phenylketonuria and Mother's reported practices related to care of their children with PKU as diet management and medical management. **2nd tool** coping scale modified to assess mothers coping toward their children suffering from PKU. **3rd tool** The medical record to assess health status of children**.** ***Results:*** the study revealed that the studied children suffering from PKU were having the highest scores of coping patterns of emotional adjustment in coping pattern scale 55.2% of mothers had satisfactory total knowledge about PKU, 60.0% of the mothers had acceptable practices about PKU. ***Conclusion:*** the present study concluded that 68.0% of the mothers had good coping of children suffering from PKU. There is a statistically highly significant positive relation between knowledge and practice. There is a statistically highly significant positive relation between knowledge and coping pattern. ***Recommendation****:* the study recommended that the community health nurse should identify mothers coping toward their children suffering from phenylketonuria and improved services to work with government to improve services to families of children with special needs.

Keywords: phenylketonuria, health need, problems, nutrition nursing care, prevention, coping scale

Introduction

Phenylketonuria (also known as PKU) is a rare inherited disorder that causes an amino acid called phenylalanine to build up in the body. PKU is caused by a defect in the gene that helps create the enzyme needed to break down phenylalanine. Without the enzyme necessary to process phenylalanine, a dangerous buildup can develop when a person with PKU eats foods that are high in protein. Because of this metabolic defect, phenylalanine rises to toxic levels inside the body and can cause mental retardation. **(Marcason, 2014).**

The incidence of PKU varies widely in different human populations. The prevalence in the general US population is approximately 4 cases per 100,000 individuals, and the incidence is 350 cases per million live births. Approximately 0.04-1% of the residents in intellectual disability clinics are affected by PKU. Turkey has the highest documented rate in the world, with approximately 1 case in 2600 births, while countries such as Finland and Japan have extremely low rates with less than one case of PKU in 100,000 births. These disorders are equally frequent in males and females. The incidence of PKU in Egypt is unknown but all cases about 8000 cases follow up in genetic clinic in Egypt **(Georgianne, 2014).**

phenylketonuria symptoms can be mild or severe and may include: Mental retardation, Behavioral or social problems, Seizures, tremors or jerking movements in the arms and legs, Hyperactivity, Stunted growth, Skin rashes (eczema) and Small head size, EEG abnormalities, and

severe learning disabilities. All of these symptoms can be avoided though when proper treatment is put into place and continues throughout life! **(Camp, 2014).**

Phenylketonuria is diagnosed through performing between one and seven days after birth. Blood is obtained by pricking the heel of the newborn and analyzing it for phenylalanine concentration. Children with PKU need to keep PHE levels low. So Keeping PHE levels between 120-360 µmol/L (2-6 mg/dL) for life. High blood PHE levels are toxic to the brain and can lead to: Lower intelligence, slowed reaction time, decreased ability to focus, decreased ability to remember, Delayed speech and decreased thinking. **(Acosta, 2010).**

The best treatment for PKU is a diet of low-protein food. There are special formulas for newborns. For older children and adults, the diet includes many fruits and vegetables. It also includes some low-protein breads, pastas and cereals. Nutritional formulas provide the vitamins and minerals they can't get from their food. Untreated PKU can lead to: Irreversible brain damage and marked mental retardation within the first few months of life Behavioral problems and seizures in older children Epilepsy, Severe behavioral, disturbance, Congenital anomaly and Skin lesions. **(Macdonald et al, 2012).**

Family coping is defined as a process of family adaptation that involves coping strategies within the family and community **(Kelly, 2010).** Coping with the care of a child with a chronic health condition occurs as parents must cognitively and behaviorally manage the stress of

comprehending the medical condition, adjusting to the diagnosis and providing appropriate care to meet the needs of the child and family **(Hobdell et al., 2010).**

All nurses who work with children which suffering from phenylketonuria should be aware of the dietary requirements and restrictions. Some medicines contain phenylalanine as a sweetener and should be avoided. - Living with dietary restrictions can be very difficult for children. They may wish to share concerns or feelings about being 'different' from peers. - If there is insufficient intake of phenylalanine, which is an essential amino acid, levels may to be too low for growth and body functions, so routine screening is especially important. **(Patney, 2011).**

Significance of the study:

Phenylketonuria is a genetic disorder. The incidence of PKU in Egypt is unknown but all cases about 8000 cases follow up in genetic clinic in Egypt. The prevalence in the population is approximately 4 cases per 100,000 individuals. It is diagnosed through performing between one and seven days after birth. Through pricking the heel of the newborn and test the blood obtained for phenylalanine concentration. PKU unfortunately in Egypt this screening test is not routine consequently it is discovered late and the parents of child have to cope with the burden of daily care related to the illness. Therefore it is necessary to assess their coping of their children in terms of stress management, adjusting to diagnosis and providing appropriate care to meet the needs of the child. Living with PKU can be difficult. The combination of a limited diet,

expensive groceries, regular blood tests, detailed food records and frequent visits to the doctor can make for trying and frustrating times. If you have a child with PKU, it can be difficult to explain why he or she can't eat "normal" foods. And, mealtimes and snack times can be a battle. The parents of child with a chronic condition have to cope with the burden of daily care related to the illness. Babies who get on this special diet soon after they are born develop normally. Many have no symptoms of PKU. It is important that they stay on the diet for the rest of their lives. Development of effective coping patterns allows mothers of PKU children to release stress and improve family functioning **(Rodenburg, 2010).**

http://www.rightdiagnosis.com/p/phenylketonuria/stats-country.htm [Accessed in 13th August, 2015 (1:50)]

Aim of the Study

The aim of this study is to assess the mothers' coping of children suffering from phenylketonuria through:-

1- Assessing the health needs and problems of children with phenylketonuria.

2- Identifying the mothers' knowledge about phenylketonuria.

3- Determining the mothers' practice related to care of their children with phenylketonuria.

4- Determining the mothers' coping pattern toward their children with phenylketonuria.

Research question:-

1- Is there relation between the mothers' knowledge and practices and their coping patterns related to care children with phenylketonuria?

2- Is there relation between mothers' socio-demographic data and their coping of children with phenylketonuria?

Out line:

Part I:

Introduction

Overview of the Phenylketonuria Disease:

1-Definitions

2- Pathophysiology

3- Epidemiology The incidence of Phenylketonuria.

4- Classification of Phenylketonuria.

5- Causes of Phenylketonuria

6- Risk factors of Phenylketonuria

7- Clinical manifestation of phenylketonuria

8- Complication of phenylketonuria.

9- Laboratory Investigations of phenylketonuria.

10- Management: of phenylketonuria

*Indication KUVAN® (sapropterin dihydrochloride)

*Diet of PKU

*A PKU Formula

*The Importance of Phenylalanine Control

*Follow up of phenlketonuria

*Screening and presentation

11- Prognosis of phenylketonuria

12- Prevention of phenylketonuria