

## Upgrading Mothers' Knowledge and Practice Regarding the Care of their Children Suffering from Phenylketonuria

Baraka Fam Mery<sup>1</sup>, Heba AbdAllah AbdElnabi <sup>2</sup>, and Shereen Said Gouda<sup>3</sup>

<sup>(1)</sup>Lecturer of Pediatric Nursing, Faculty of Nursing, Beni-Suef University, Egypt.

<sup>(2)</sup>Lecturer of Pediatric Nursing, Faculty of Nursing, Zagazig University, Egypt.

<sup>(3)</sup>Lecturer of Pediatric Nursing, Faculty of Nursing, Beni-Suef University, Egypt.

### Abstract

**Background:** Phenylketonuria (PKU) is a hereditary metabolic disorder caused by phenylalanine hydroxylase deficiency. **Aim:** This research aimed to assess the influence of an educational program on upgrading knowledge and practice of mothers regarding the care of their children suffering from phenylketonuria. **Design:** A quasi-experiment. **Sampling:** A convenience sample was used. **Setting:** At the Genetic & Metabolic Outpatient Clinic at Pediatric Zagazig University Hospital. **Tools:** Two tools; **tool I:** A structured interview survey, **tool II:** Reported practice checklist. **Results:** revealed that nearly one quarter of studied mothers had satisfactory total knowledge score at pretest compared to most of them post intervention. As well as, nearly three quarters of them had incompetent total practices toward caring of their children with PKU before intervention program that reduced to less than one fifth of them post of program implementation. Moreover, A statistically significant correlation detected between total mothers' knowledge score and their total reported practice at post intervention ( $p < 0.001$ ). **Conclusion:** The mothers had a low knowledge and practice levels regarding the care of their children with PKU, as well as the educational program was effective in upgrading mothers' knowledge and practice toward caring of their children. **Recommendation:** Continuous educational programs for mothers about phenylketonuria are required to increase their awareness and performance toward the care of their children with phenylketonuria.

**Keywords:** Children, Knowledge, Mothers, Phenylketonuria, Practice.

### Introduction

Phenylketonuria is a rare genetic condition that results in rising level of phenylalanine in the body. When an affected child consumes high protein diet, a dangerous buildup of phenylalanine can occur.

Phenylketonuria is caused by a gene defect that helps to produce the enzyme required for the break down phenylalanine (Phe). Hence, phenylalanine elevates to dangerous levels in the body and can lead to a variety of complications especially

mental retardation (*Manta-Vogli et al., 2020*).

Phenylketonuria is the most common autosomal-recessive mendelian trait of amino acid metabolism and is caused by variations in the phenylalanine Hydroxylase (PAH) gene. PKU is more common in some ethnic groups than others and in different parts of the world. PKU affects 45.0 million children worldwide, with an incidence of 1:23,930 live births (*Hillert & Alicia, 2020*).

Untreated phenylketonuria typically causes growth failure, severe irreversible intellectual disability, hypopigmentation of skin and hair, developmental delay, motor impairment, ataxia and seizures. In terms of disease severity and dietary therapy, children with PKU are heterogeneous. The majority of PKU-affected children can lead nearly normal life as a result of early diagnosis and treatment with a low-Phe diet (*Van spronsen et al., 2017*).

Within seven days of delivery, a serum phenylalanine test was used to diagnose phenylketonuria. Blood sample is obtained by pricking the newborn's heel and tested for phenylalanine concentration. Phe levels in PKU children must be maintained at 120-360  $\mu\text{mol/L}$  (2-6 mg/DL). Elevated Phe blood levels are harmful to the brain and potentially result in reduced cognitive function, attention,

memory, and delayed speech (*Ashe et al., 2019*).

Dietary therapy involves reducing the intake of natural protein and replacing it with a protein free Phe, is the backbone of treatment for PKU. Dietary therapy involves a skilled team of metabolic physician and nutritionist. Infants who recently diagnosed with PKU, blood phenylalanine levels should be evaluated in clinic and checked frequently until they stabilize. A diet tailored for PAH deficiency must provide sources of all other essential elements required for proper growth and health maintenance, as foods typically ingested as protein sources contain these other key nutrients (*Manta-Vogli et al., 2020*).

Mothers who have children with PKU are important to the afflicted child's physical and mental health. But these mothers have faced many obstacles throughout their lives, such as heavy parental responsibilities brought on by requirements specific to their diseases, restricted access to high-quality healthcare, and a shortage of qualified medical personnel to offer the right advice and treatment (*Boettcher et al., 2020*).

The pediatric nurse has a critical role in assisting parents of pediatrics with PKU in improving their capacity to provide for their children by offering clear instructions and constructive

criticism. When working with children who have PKU, nurses should be knowledgeable about the dietary requirements and restrictions, as well as medications that should be avoided due to their artificial sweetener content. Effective PKU management necessitates collaboration between medical professionals, families, and children (Patney, 2017).

### Significance of the study

Phenylketonuria is an uncommon condition that impairs a child's ability to grow and develop physically, mentally, and socially, among other domains. The prevalence of PKU in Europe is 1:10,000, while it is less common in ethnic groups descended from Africa and South Asia. Although the prevalence of PKU in Egypt is unclear, there are about 40,000 cases that are monitored at genetic clinics there (Patrikios & Ioannis, 2019).

The prevalence of phenylketonuria increased, particularly in rural areas of Egypt, because of a lack of awareness among mothers toward the importance of newborn screening programs, a high consanguinity rate, and inadequate management of PKU. As a result, the children's mental and psychosocial development was seriously affected (Hillert & Alicia, 2020). So, the present study is conducted to shed light on the knowledge and practice of mothers

toward caring of their pediatrics with PKU because the mothers are the children's primary caretakers and they have a crucial role in providing adequate care and preventing PKU problems.

### Aim:

To evaluate the influence of an educational program on upgrading mothers' knowledge and practice regarding the care of their children suffering from PKU through:

- Evaluating mothers' knowledge and practice about the care of their children with PKU.
- Designing and implementing an educational program to upgrade mothers' knowledge and practice.
- Estimating the effect of an educational program on upgrading mothers' knowledge and practice.

### Research Hypothesis

- 1- Mothers will have satisfactory knowledge and practice following execution of the program.
- 2- The program will improve knowledge and practice of mothers of children suffering from phenylketonuria.

### Subjects and Method

#### 1-Technical design

#### Research Design

A quasi- experimental design (pre & post) was established.

### **Setting**

The study was conducted at Genetic and Metabolic Outpatient Clinic at Pediatric Zagazig University Hospital.

### **Subjects:**

A convenience sample consisted of (100) mothers accompanied their children with phenylketonuria from different ages and both sexes at the previously mentioned setting.

### **Tools for data collection**

Two tools were involved:

#### **1. A Structured Interview Survey:**

It was created by the researchers as guided by **Elsayed et al. (2020)**. It included the following four sections and was written in Arabic.

**Part 1:** Socio-demographics of the mothers.

**Part II:** Characteristics of the children under study.

**Part III:** The medical history of children including family history, illness duration, how to discover the disease, follow up, number of other children with PKU in the family, manifestation, sleep pattern, and reported problems.

**Part IV:** Studied mothers' knowledge about PKU: it consisted of 20 closed-ended questions clustered into definition, causes, symptoms, laboratory investigation, factors that alter PKU, treatment, diet, complications, and prevention

of complications. The researcher designed it in Arabic form.

### **Scoring system:**

A scoring system was followed based on the obtained answers. The total score was 40, converted to 100%. The mothers' responses were compared with a model key answer; the correct answer was given 2 scores, whereas incomplete one obtained one score and incorrect one obtained zero.

Based on the mothers' answers, their overall score of knowledge was dichotomized into:

-Satisfactory  $\geq 65\%$

-Unsatisfactory  $< 65\%$

### **II. Reported Practice Checklist**

The researcher reviewed the relevant literature (**Bowden et al., 2016**) and made modifications to it. To estimate mothers' stated practices; it involves 62 items with done/not done response formats such as gathering samples to check the levels of Phe (10 items), measurement of diet balance (7 items), preparation of medical formula (7 items), weight (5 items), height measurement (7 items), hand washing (5 items), oral care (5 items), nail trimming (6 items) and bathing (10 items).

### **Scoring system:**

The overall score of practice was 62 and converted to one hundred percentages (100%). Mothers' practice was evaluated by giving a (1) and (0) score for a done and a not done answer, respectively. The

scores were then converted to percentages, and the overall score was categorized into:

- $\geq 75\%$  = Complete practice
- $< 75\%$  = Incomplete practice

### **Intervention program:**

The researchers created this program to teach and prepare mothers to care for PKU-affected children.

### **General goals of the program:**

It aimed to upgrade mothers' knowledge and practice toward the care of pediatric suffering from phenylketonuria.

### **Specific goals:**

Following this program, mothers would have the following abilities:

1. Define phenylketonuria and its causes.
2. Enumerate signs and symptoms.
3. Describe the dangers of phenylketonuria and how to avoid it.
4. Explain diagnosis and management of phenylketonuria.
5. Show how to provide their PKU children with proper care.
6. Investigate the effect of the intervention program on mothers' understanding and behavior in relation to caring for their PKU-affected children.

**The intervention program was created in the following four stages:**

#### **(I) Phase of assessment**

A portion of the program was designed with the purpose of evaluating mothers' practices and knowledge. Before the intervention

program was put into action, each mother was interviewed one-on-one to gauge her general knowledge and practice (pre-test) using tools I and II. This was done after informing them of the study's purpose and obtaining their consent to take part in it.

#### **(II) Phase of planning**

The researchers prepared the intervention program in agreement with the findings from the structured interview survey and checklist (the pilot study and evaluation phase) and a review of the relevant literature. The goals of the program, which took the form of a booklet, were derived from the needs, requirements, and shortcomings that were identified.

The assessed needs guided the selection of the intervention program's contents. Different teaching strategies, such as lectures, group discussions, role plays, demonstrations, storytelling, and reinforcement, were chosen to fit the needs of one-on-one instruction. Teaching resources included instructional movies that addressed both theoretical and practical material, colorful posters, and handouts (books).

#### **(III) Phase of implementation**

- To aid in the learning process, the intervention program for this study was carried out over the course of three sessions, during which time each mother was questioned independently. Each session had a

different duration based on the mothers' reactions and the subject matter. It was between thirty and forty-five minutes. The explanations of the sessions were given in plain Arabic, appropriate for the comprehension level of the studied mothers. To increase mothers' active engagement and promote learning, each session included motivation and reinforcement.

- The intervention program took the form of showing instructional films and meeting in person. The mothers were initially given a face-to-face explanation of the main elements, followed by the showing of instructional videos that included both practical and theoretical content.

#### **Session I:**

Mothers and researchers met and discussed the goal of the intervention, the number of sessions, the time of the meeting, the rules of the program, the expected results, and the advantages of participating in the program.

#### **Session II:**

Information on the definition, causes, symptoms, complications, diagnosis, and management for their PKU children were covered in the second session.

#### **Session III:**

During the third session, studied mothers were trained and educated by the researchers on how to care for their children who had PKU.

#### **(IV) Phase of evaluation**

- During this phase, each mother underwent a follow-up interview one month later, during which they were reassessed (post-test) by re-answering the questions (using tool I) and evaluating their practice (using tool II). The pamphlet or handout was given to the mothers after the conclusion of the training sessions (post-test).

#### **II- Operational design**

Fieldwork, a pilot research, and the validity and reliability were all incorporated in the operational design.

#### **Validity and reliability**

The validity was measured by a jury group consisting of five professors (three professors of pediatric nursing and two professors of pediatrics & genetics). The group judged the tools for accuracy, comprehensiveness, and language clarity. According to their suggestions that certain elements be corrected, added, or omitted, The Cronbach's alpha test was used to evaluate the tools' reliability. For knowledge and practice (Cronbach's  $\alpha = 0.812$  and  $0.832$ , respectively), the reliability coefficient was good.

#### **b- Ethical consideration**

Subjects' consent was obtained following an explanation of the study's purpose. They were informed that they could get out of the study at any point and that their information would be kept private and used exclusively for research.

They were also given the option to decline participation.

#### **c- Pilot study**

Ten mothers (10%) participated in a pilot research to assess the tools' applicability and clarity and to estimate how long it would require for completing the data. This pilot study indicates that the necessary adjustments were done. The time of pilot study collected in November 2020. Simple modifications in the tools were done so the mothers who took part in the pilot trial were not involved in the current work.

#### **d- Field work**

Following the identification of the participants who met the study's requirements, the researchers gave a brief explanation of the study's purpose and methodology and got each mother's informed consent. The meeting location and schedule were also decided upon by the researchers. The researchers greeted them at the morning shift, which lasted from 10:00 a.m. to 1:00 p.m. For the purpose of gathering data and implementing a post-test after a month, the researchers visited the study settings three days a week on (Monday, Tuesday, and Wednesday) at the Pediatric Zagazig University Hospital's Genetic & Metabolic Outpatient Clinic. The study was established over six months, from the start of January 2021 to the end of June 2021.

### **III. Administrative Design.**

The directors of the previously mentioned locations officially gave permission for the study to be conducted in the settings that were chosen.

#### **IV- Statistical Analysis**

The data was coded and transferred into IBM SPSS for Windows, version 24 (IBM Corp., Armonk, NY, USA). The data was revised to find out any errors. The data was analyzed to obtain frequency tables with proportions. Qualitative variables were expressed in frequency and proportions, and quantitative ones were represented in mean and standard deviation. Chi-square test compared the categorical variables were compared to examine the differences between categorical data. The inter-correlation between quantitative data was evaluated using Spearman rank correlation. **The results were deemed statistically significant at  $p \leq 0.05$  and high significant if  $p < 0.001^{**}$ .**

**Table 1** displays that 59% of the studied pediatrics were males. Regarding their age, 70% were <6 years old, with a mean and SD of  $4.19 \pm 3.47$  years. Additionally, 63% of the studied children did not attend school, 38% were the first child within their families' birth order, and 64% had one to three siblings. Also, 54% of the children had a normal weight and 66% had a length <100 cm.

As is clear from **Table 2**, it was found that 52% of mothers aged 20 to less than 30 years, with a mean age of  $28.09 \pm 6.64$  years. Concerning the mothers' educational level, 15% had a

higher level of education. Also, this table reveals that 86% were housewives. Additionally, it is observed that 81% lived in rural areas and 51% got their knowledge about PKU from physicians or nurses.

**Table 3** clarifies that 51% of the children had no family history for PKU, and 73 % discovered the disease since birth and within the first year of life. Moreover, 75% discovered the disease by the Guthrie test. Concerning the frequency of follow-up, 61% reported that they follow up once a month, and 69% had only one child diagnosed with PKU.

**Table 4** shows that 75% of the pediatrics had eczema as a manifestation of PKU, and 77% had a normal sleep pattern. In addition, 60% compliance with the prescribed diet for PKU, and 56% experienced delays in mental and social skills.

The overall score of knowledge was presented in **Fig 1**. It was noticed that 77% of the studied mothers had an unsatisfactory knowledge in pre-intervention. After the intervention, 82% had a satisfactory level of total knowledge.

**Fig 2** shows that 74% of mothers had an incompetent total practice at

pretest. On the other hand, in post-intervention, 89% had a competent level of overall reported practice.

As shown in **Table 5**, there was a marked improvement in total knowledge level among the studied mothers at posttest compared to pretest with a highly statistically significant difference ( $p < 0.001$ ). As evidence, there was statistical significant relation between the total mothers' knowledge scores and their educational level & occupation ( $p < 0.001$ ), but there was no statistical relationship between total knowledge and age & residence.

**Table 6** clarifies that there were marked statistically significant improvements in each item of mothers' reported practice after the intervention program compared to before the intervention. As observed, there was a highly significant relation between total reported practice scores and educational level and occupation ( $p < 0.001$ ). Additionally, there was a statistically remarkable relationship between the practice and age ( $p < 0.05$ ).

It was evident from **Table 7** that there was a highly statistically positive significant correlation between the total mothers' knowledge score and the total reported practice at post intervention ( $p < 0.001$ ).

**Table (1): Socio- demographic characteristics of studied children suffering from PKU**

<b>Socio- demographics</b>	<b>No</b>	<b>%</b>
<b>Sex</b>		
Male	59	<b>59</b>
Female	41	41
<b>Age (years)</b>		
< 6	70	<b>70</b>
6 < 12	17	17
12 ≤ 18	13	13
<b>Mean ±SD</b>	<b>4.19±3.47</b>	
<b>Educational level</b>		
Not attending school	63	<b>63</b>
Nursery level	12	12
Primary level	19	19
Secondary level	6	6
<b>Birth order</b>		
First	38	<b>38</b>
Second	22	22
Third	17	17
Fourth and more	23	23
<b>Number of sibling</b>		
No sibling	29	29
1:3	64	<b>64</b>
4:6	7	7
<b>Weight</b>		
Over weight	23	23
Normal weight	54	<b>54</b>
Under weight	23	23
<b>Height\ cm</b>		
<100	66	<b>66</b>
100<150	24	24
≥150	10	10

**Table (2): Socio- demographics of the mothers of affected children (n = 100)**

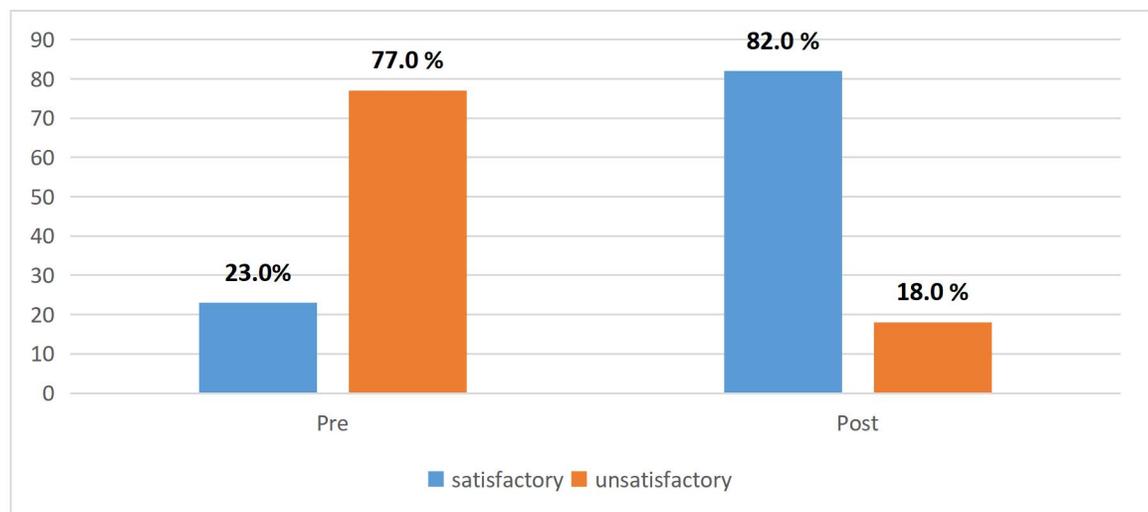
<b>Characteristics</b>	<b>No</b>	<b>%</b>
<b>Age</b>		
<20 year	5	5
20 to <30 year	52	<b>52</b>
30 to <40 year	35	35
≥40 year	8	8
<b>Mean ±SD</b>	<b>28.09 ± 6.64</b>	
<b>Education</b>		
Illiterate	29	29
Read and write	24	24
Intermediate	32	32
Higher	15	<b>15</b>
<b>Occupation</b>		
Housewife	86	<b>86</b>
Working	14	14
<b>Residence</b>		
Rural	81	<b>81</b>
Urban	19	19
<b>Mothers' source of knowledge regarding the PKU</b>		
Media	35	35
Physicians or nurses	51	<b>51</b>
Neighbors or friends	14	14

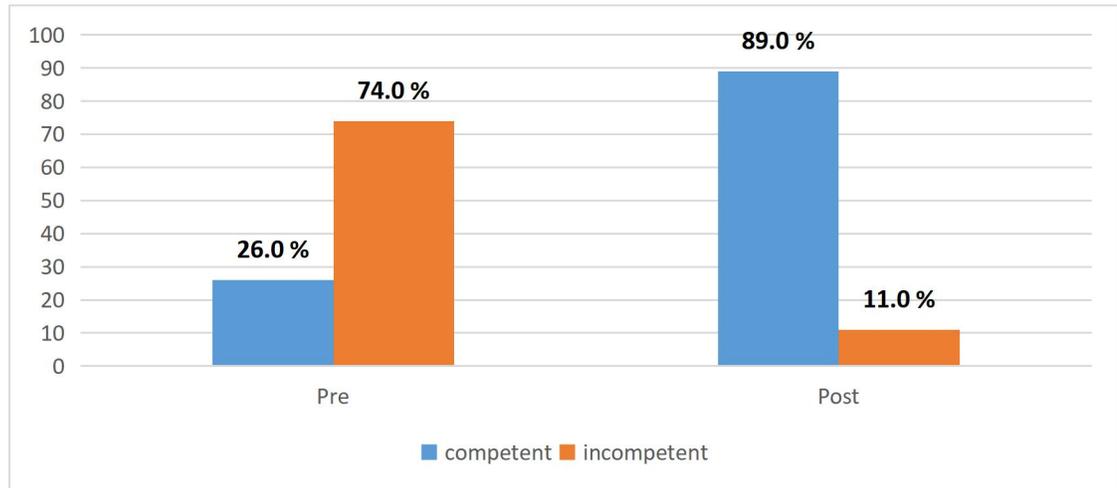
**Table (3): Medical history of children regarding the PKU (n = 100)**

<b>History</b>	<b>No</b>	<b>%</b>
<b>Family history for PKU</b>		
Yes	49	49
No	51	<b>51</b>
<b>Beginning of the disease (years)</b>		
Birth-<1	73	<b>73</b>
1 <3	8	8
3 <9	15	15
≥9	4	4
<b>Methods of the disease discovery</b>		
Suddenly	15	15
Via the manifestations of the disease	3	3
Via Guthrie test	75	<b>75</b>
Via complications	7	7
<b>Follow up frequency (month)</b>		
Two times	22	22
One time	61	<b>61</b>
Not following up	17	17
<b>Frequency of PKU peditrics in the family</b>		
One child	69	<b>69</b>
Two children	25	25
>Two children	6	6

**Cont. Table (4)**

<b>Medical History</b>	<b>No</b>	<b>%</b>
<b>Manifestations experienced by the child</b>		
Change in skin and hair color	13	13
Musty odor of skin or urine	2	2
Eczema	75	75
All of the above	10	10
<b>Sleep pattern</b>		
Normal	77	77
Abnormal	23	23
<b>PKU diet compliance</b>		
Yes	60	60
No	40	40
<b>Reported problems</b>		
Psychological and behavioral problems	15	15
Mental and social skills delay	56	56
Osteopenia ,osteoporosis and fractures	12	12
Delayed motor development	7	7
No complications	10	10

**Figure 1: Total knowledge score of studied mothers.**



**Figure 2: Overall practice score of studied mothers.**

**Table (5): Relationship between studied mothers' characteristics and their overall score of knowledge.**

Socio-demographic characteristics	Total Knowledge Score										
	Pre				Post				Total	Chi-square	
	Satisfactory		Unsatisfactory		Satisfactory		Unsatisfactory			X <sup>2</sup>	P-value
	N	%	N	%	N	%	N	%			
<b>Age</b>											
<20 year	2	40.0	3	60.0	4	80.0	1	20.0	5	5.058	0.168
20- <30year	11	21.6	40	78.4	35	68.7	16	31.3	51		
30- <40 year	6	18.2	27	81.8	23	69.7	10	30.3	33		
≥40 year	5	45.5	6	54.5	8	72.7	3	27.3	11		
<b>Educational level</b>											
Illiterate	3	10.3	26	89.7	25	86.2	4	13.8	29	26.14	<0.001**
Basic education	7	26.9	19	73.1	18	69.2	8	30.8	26		
Intermediate education	10	25.6	29	74.4	31	79.5	8	20.5	39		
Higher education	4	66.7	2	33.3	6	100.0	0	0.0	6		
<b>Occupation</b>											
Housewife	10	12.0	73	88.0	57	68.7	26	31.3	83	18.20	<0.001**
Working	11	64.7	6	35.3	16	94.1	1	5.9	17		
<b>Residence</b>											
Rural	17	21.8	61	78.2	67	85.9	11	14.1	78	1.330	0.249
Urban	7	31.8	15	68.2	18	81.8	4	18.2	22		

(\*\*) Highly significant at p< (0.001)

**Table (6): Relationship between mothers' characteristics and their overall reported practice score regarding PKU disease.**

Socio-demographic characteristics	Overall score											
	Pre					Post					Chi-square	
	Complete		Incomplete		Complete		Incomplete		Total	X <sup>2</sup>		
	N	%	N	%	N	%	N	%				
<b>Age (year)</b>												
<20	2	40.0	3	60.0	3	60.0	2	40.0	5	9.822	<0.020*	
20- <30	10	20.4	39	79.6	41	83.7	8	16.3	49			
30- <40	6	17.1	29	82.9	24	68.6	11	31.4	35			
≥40	7	63.6	4	36.4	9	81.8	2	18.2	11			
<b>Educational level</b>												
Illiterate	4	13.8	25	86.2	21	72.4	8	27.6	29	36.479	<0.001**	
Basic education	7	28.0	18	72.0	19	76.0	6	24.0	25			
Intermediate education	4	12.9	27	87.1	24	77.4	7	22.6	31			
Higher education	12	80.0	3	20.0	14	93.3	1	6.7	15			
<b>Occupation</b>												
Housewife	13	15.5	71	84.5	71	84.5	13	15.5	84	26.495	<0.001**	
Working	12	75.0	4	25.0	14	87.5	2	12.5	16			
<b>Residence</b>												
Rural	13	16.9	64	83.1	59	76.6	18	23.4	77	1.810	0.179	
Urban	6	26.1	17	73.9	18	78.3	5	21.7	23			

(\*\*) Highly significant at p&lt; (0.001)

**Table (7): Correlation between the total knowledge and practice score**

Variables		Knowledge	
		r	p-value
Reported practice	Pre	0.541	0.00**
	Post	0.782	0.00**

(\*\*) Highly significant at p&lt; (0.001)

## Discussion

Phenylketonuria is a common chronic genetic illness that requires daily effort from the mother to adhere to a specific diet plan and manage the child's limits. Moreover, the mother bears the majority of the responsibility for the child's care and the hospital's workload. As a result, mothers are extremely important in the care and treatment of their phenylketonuric children. Phenylketonuria is a lifelong disorder necessitates rigorous dietary management (**Mortazavi et al., 2020**).

The mothers caring for children with PKU should be provided with appropriate instructions and guidance about PKU management (**Van Spronsen et al., 2017**). So this work assessed the impact of an educational program on upgrading knowledge and practice of mothers regarding the care of children suffering from PKU. The same findings were reached by **Abd-Elkodoos et al. (2022)**, who found that the educational intervention for caregivers of pediatric with PKU had a significant positive influence as it improved knowledge and practice.

Regarding children's characteristics, the current research showed that more than two thirds of pediatrics aged < 6 years with a mean age of  $4.19 \pm 3.47$  years; less than two thirds were males and more than one third were the first baby in the

family. Moreover, around two thirds were not attending school yet, and around two thirds had one to three siblings diagnosed with PKU. These results are consistent with **Elsayed et al. (2020)**, who mentioned that the mean age of children was  $4.45 \pm 2.07$  years; males represented one third of them; more than two fifths were born first within their families, one third of children hadn't entered school and more than one third had one sibling with PKU.

Concerning the mothers, more than half of them aged 20 to <30 years in this study. The same result was reached by **Evans et al. (2019)**, who clarified that half of mothers were between 20 and 30 years old. Additionally, the present study found that less than one fifth of mothers had higher level of education; greater than three quarters were housewives. These results agree with **Carpenter et al. (2018)**; **Shaji et al. (2020)**, who demonstrated that greater than one fifth of the mothers had higher education and almost all of them, were not working.

As regard the source of knowledge about PKU, this study illustrated that more than half of mothers got knowledge from physicians and nurses. This result is in agreement with **Rahgoi et al. (2019)**, who stated that greater than two-thirds of mothers got their knowledge about PKU from nurses and physicians. Based on the explanation of the

researchers, such results reflect the mothers' need for effective education about proper PKU child management.

Concerning children's medical history, the current findings show that about greater than half of children do not have a family history of PKU. This may be attributed to the tradition of consanguineous marriage in many Egyptian families (**Hillert & Alicia, 2020**). This result is similar to the study established by **Abd-Elkodoos et al. (2022)**, who mentioned that around 50% of the studied children did not have a family history of PKU disease. Also, such finding agrees with **Sadek et al. (2018)**, who revealed that around half of pediatric didn't have history of PKU. As regards the beginning of the disease manifestations, this research clarified that nearly three-quarters of children are diagnosed with PKU in the first year of life. Those children were early diagnosed through the neonatal screening program for inborn errors of metabolism (IEM). This outcome is in line with **Carpenter et al. (2018)**, who found that the majority of children were detected during the first year of life. On the contrary, this finding disagrees with **Abd-Elkodoos et al. (2022)**, who found that less than one fifth of children were detected in the first year of life.

This work found that 75% of children discovered the disease through the Guthrie test. This result may be due to the routine heel sample that is given to all children in Egypt immediately after birth. This result contrasts with **Elsayed et al. (2020)**, who demonstrated that the majority of children were discovered via lab investigations.

As regards clinical signs experienced by the child, our study showed that three-quarters of children had eczema as a manifestation of PKU. This may be attributed to elevated Phe levels and the products of its breakdown, which cause permanent harm to body tissues (**Van spronsen et al., 2017**). This finding agrees with the results of **Gad et al. (2019)**, who displayed that less than one fifth of the children had eczema. In addition, more than half of children follow the prescribed diet for PKU. Similar findings were reported by **Medford et al. (2017)**, who reported that the majority of children followed the food plan, and got the recommended amount of diet.

Regarding the experienced complications, we found that more than half of the studied sample had delayed mental and social skills. The current study finding is congruent with **Sadek et al. (2018)**, who stated that the majority of pediatrics suffered from delayed social development. Also, these findings agree with **Gad et al.**

(2019), who discovered that the pediatrics had a history of motor progressive delay, seizures, and autism as a complication of PKU.

Concerning pre-test mothers' knowledge about the PKU, the present study displayed that more than three-quarters of women had unsatisfactory knowledge about the PKU disease. This finding may be due to a lack of educational programs specified for mothers about PKU disease and mothers' negligence to gain knowledge. The study result is in line with **Elsayed et al. (2020)**, who reported that more than half of women had inadequate knowledge regarding PKU. On the other hand, **Fouad & Abd-Elmoneem (2016)** reported that three-quarters of the females displayed satisfactory knowledge regarding PKU. This difference between the studies may be related to differences regarding the availability of training programs for mothers about PKU disease between the study subjects.

Our research displayed that the highest percentage of women had satisfactory knowledge about the PKU disease post-test compared to pre-test. Such finding may be due to the impact of the program on updating and enhancing mothers' knowledge. This finding is similar to the results of **Abd-Elkodoos et al. (2018)**, who declared that almost three-quarters of caregivers had a

good level of knowledge post of program implementation.

As observed from the current study, it was found that almost three-quarters of them had an incompetent level of practice about PKU. This may be related to a low educational level, a lack of specific PKU teams in rural areas, and inadequate guidance by health professionals. This goes in line with **Akkus et al. (2020)**, who discovered that the majority of family caregivers had an unsatisfactory practice regarding PKU. This result is incongruent with the research by **Fouad & Abd-Elmoneem (2016)**, who showed that family caregivers have adequate total practice regarding PKU.

The highest proportion of women had a competent practice about the PKU disease post-test. This outcome may reflect that the intervention had a remarkable favorable impact on mothers' practices regarding PKU disease. This finding is similar to the results of **Abd-Elkodoos et al. (2018)**, who stated that nearly three-quarters of caregivers had a good practice post-program.

This study displayed a highly potential relationship between mothers overall knowledge score and their educational level. This may be related to mothers who have a higher educational level having a satisfactory level of knowledge. This result is in line with **Akkus et al. (2020)**, who reported that family

caregivers' knowledge scores increased with their educational levels, which may improve the provided care for their PKU pediatrics.

The present study clarified a highly statistically significant relationship between the overall practice scores of mothers and their education. This finding may be attributed to the fact that mothers with higher education have updated and improved their level of practice. This finding matches that of **Fouad & Abd-Elmoneem (2016)**, who found that family caregivers' practice scores increased with their educational levels.

The results of this work showed that there was a statistically significant relation between the total reported practice of the mothers and their age. This may be related to increasing mother's age, which helps her gain more experience and practice. This is consistent with **Witalis et al., (2017) and Abd-Elkodoos et al. (2022)**, who found significant relation between the overall practice of the caregivers and their ages.

Our findings illustrated a highly significant correlation between the overall knowledge of the studied mothers and their overall practice. This indicates that mothers with adequate knowledge will help her to improve their care and practice. This finding is in accordance with **Elsayed et al. (2020)**, who stated that good practice was more

commonly encountered among mothers with a high level of knowledge. Also, this finding is supported by **Fouad & Abd-Elmoneem (2016)**, who found that there was a highly significant relationship between the overall knowledge and total practice.

#### **Conclusion:**

**Based on the findings, it can be deduced that:**

The educational intervention was efficient in upgrading mothers' knowledge and practice with respect to the care of their children suffering from phenylketonuria.

#### **Recommendations:**

- Dissemination of educational programs for mothers with pediatrics suffering from PKU.
- Presence of educational kitchen in all clinics for phenylketonuria children to educate mothers how to make healthy meals with low or free phenylalanine.
- A lot of places should be available for selling any product with low phenylalanine for PKU children at affordable prices.
- Establishing support groups and family therapy programs to overcome deficient knowledge and practice toward the children with PKU.
- Designing suitable booklets and written leaflets including basic knowledge about PKU

disease and its care management should be provided and distributed in outpatient clinics.

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