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Assessment of Mothers' Knowledge and Practice Regarding Care of Their Children Suffering from Phenylketonuria

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Abstract

Background Phenylketonuria is an inherited metabolic disease caused by phenylalanine hydroxylase deficiency. **Aim** This study aimed to assess mothers' knowledge and practice regarding care of their children suffering from Phenylketonuria. **Design** A descriptive design was utilized in this study. **Sampling** A purposive sample comprised of 91 mothers having children with phenylketonuria was selected to conduct this study. **Setting** This study was conducted at specialized genetic outpatient clinic in children's hospital affiliated to Ain Shams University Hospital. **Tools** Two tools were used in this study **Tool (1)** A structured questionnaire to assess mothers' knowledge regarding Phenylketonuria. **Tool (2)** Observational checklists to assess mothers' reported practice regarding care of their children with phenylketonuria. **Results** The total knowledge of mothers was unsatisfactory, the total reported practice regarding care of their children with phenylketonuria was incomplete and there was a highly statistically significant difference between total knowledge and reported practice of mothers and their characteristics particularly educational level and occupation. **Conclusion** The studied mothers had unsatisfactory level of knowledge and incomplete practice regarding care of their children with Phenylketonuria, there was a statistically significant difference between studied mothers' total level of knowledge and reported practice and their characteristics. **Recommendation** Continuous educational programs for mothers about phenylketonuria are required to increase their awareness and performance regarding care of their children with phenylketonuria.

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

Introduction

Phenylketonuria (PKU) is a rare inherited disorder caused by a defect in the gene that creates the phenylalanine hydroxylase enzyme which is required for breakdown of phenylalanine. Absence of enzyme which is necessary to process phenylalanine results in dangerous buildup of phenylalanine especially with high dietary protein intake. This metabolic defect raise blood phenylalanine level to toxic levels and can cause serious problems particularly mental retardation ***(Manta-Vogli et al,2020).***

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Phenylketonuria caused by variants in the phenylalanine Hydroxylase (PAH) gene and it is considered as the most common autosomal-recessive mendelian phenotype of amino acid metabolism. The incidence of PKU varies among ethnic groups and geographic regions worldwide. Globally, there is 45.0 million child affected by PKU, with global prevalence 1:23,930 live births ***(Hillert &Alicia, 2020).***

Untreated PKU generally results in developmental delay or severe irreversible intellectual disability, as well as growth failure, hypopigmentation of skin and hair, motor deficits, ataxia, and seizures. The children affected with PKU are heterogeneous in terms of disease severity and dietary therapy. Early diagnosis and treatment with a low-Phenylalanine diet has enabled an almost normal life for the majority of PKU affected children ***(Van spronsen et al., 2021).***

Phenylketonuria diagnosed through performing serum Phenylalanine test within seven days after birth. Blood sample is obtained by pricking the heel of the newborn and analyzing it for phenylalanine concentration. Children with PKU need to keep Phenylalanine levels between 120-360 µmol/L (2-6 mg/DL). High blood Phe levels are toxic to the brain and can lead to lower intelligence, decreased concentration, decreased ability to remember, delayed speech and decreased thinking ***(Ashe et al., 2019).***

Dietary therapy remains the mainstay of therapy for PKU, requiring a decrease in the intake of natural protein
and replacing it with a protein source devoid of Phenylalanine. Dietary therapy must be managed by experienced metabolic physician and nutritionist team. Infants newly diagnosed with phenylalanine Phenylketonuria should be followed in clinic and have blood phenylalanine levels monitored frequently until stabilized. Because foods normally consumed as protein sources contain other essential nutrients, it is important that a diet modified for PAH deficiency provides source for all other nutrients necessary for normal growth and health maintenance ***(Ford et al., 2018).***

Mothers of children suffering from phenylketonuria play a crucial role in the physical and emotional well-being of the affected children but they face multiple difficulties throughout their lives including high parental responsibilities due to disease-specific requirements, limited access to high quality health care, and lack of experienced health care professionals to provide appropriate consultation and treatment ***(Boettcher et al, 2021).***

Pediatric nurse has a prominent role in providing the mothers of PKU children with appropriate guidance and positive reinforcement. Also, pediatric nurse can improve the ability of PKU children's mothers to care for their children, as well as acting as a family advocates to coordinate information and resources. All nurses who work with children suffering from PKU should provide the parents with instructions about dietary requirements, restrictions and contraindicated medications which contain the artificial sweetener aspartame. Moreover, the pediatric nurse should cooperate with other health care providers, the child and family caregivers to manage PKU successfully **(Patney, 2017).**

Significance of the study

Phenylketonuria is a rare condition among children that has numerous complications on child wellbeing and affects the child health in all domains of growth and development. While PKU disease is less common in ethnic groups descending from Africa and South Asia and Finland, the incidence of PKU in Europe is 1:10,000. In Egypt, the incidence of PKU is

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unknown but there is around 40000 cases follow up in genetic clinics in Egypt ***(Patrikios & Ioannis, 2019)*** **.**

High consanguinity rate in Egypt as well as decreased awareness of mothers regarding the importance of newborn screening programs in early detection and management of PKU favored the prevalence of phenylketonuria especially in rural areas of Egypt and the children's mental and psychosocial development is seriously affected. Therefore, from the researcher point of view, the current study is conducted to shed the light on the mothers' knowledge and reported practice regarding care of their children suffering from PKU, because mothers are the primary caregivers for their children and have a significant role in providing appropriate care and prevention of PKU complications

The aim of the study

This study aimed to assess mothers' knowledge and reported practice regarding care of their children suffering from phenylketonuria.

Subjects &methods

Design

A descriptive research design was used for conducting this study.

Setting

This study was conducted at the Specialized Genetic Outpatient Clinic in Children's Hospital affiliated to Ain Shams University Hospital.

Subject

A purposive sample that included 91mothers accompying their children who suffer from PKU and attended to the previously mentioned setting.

Inclusion criteria

Mothers having children confirmed diagnosis with PKU regardless their ages and gender and attended to the previously mentioned setting.

Exclusion criteria

Mothers having children suffering from other chronic illnesses either physical or mental.

Tools for data collection

Two tools were used to conduct this study

First tool

A structured questionnaire which was adapted from **Elsayed et al., (2020)** and modified by the researcher after reviewing the related literature and it consisted of 4 parts:

Part (1) concerned with characteristics of the studied mothers namely age, educational level, occupation and place of residence.

Part (2) Concerned with the characteristics of the studied children (gender, age, birth order, number of siblings, and educational level).

Part (3) Assess the medical history of children suffering from PKU (duration of the illness, date of discovery, disease progress, laboratory investigations, nutrition, follow up, sleep, exercise, physical and psychological problems).

Part (4) Concerned with the studied mothers' knowledge about PKU, the questionnaire consists of 20 closed end questions in relation to definition, causes, signs and symptoms, diagnosis, follow up examinations, factors that alter Phe level, treatment, diet, complications of PKU and prevention of complications.

Scoring system for knowledge

According to the answers obtained from studied mothers, a scoring system was followed. The total score was 40 and converted to 100%. The studied mothers' answers were compared with a model key answer, where (2) scores was given for correct answer, (1) score for incomplete answer, and (zero) for incorrect answer.

Second tool

Observational checklists were adapted and modified by the researcher after reviewing the related literature from ***Bowden, (2016)*** **.**It

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includes 62 items with done\not done answer format to assess mothers' reported practice regarding care of their children with PKU namely collecting sample for checking Phenylalanine level, measurement of diet balance, preparation of medical formula, weight measurement, height measurement, hand washing, oral care, nail trimming and bathing.

Scoring system for practice

The total scores of mothers' practice was 62 scores and converted to one hundred percentages (100%), Mothers' practice was evaluated by giving (1) score for done answer and (0) score for not done answer, then the scores are converted to percentage and total score categorized as the following:

- ≥ 75% = Complete practice
- < 75%= Incomplete practice.

Pilot study

A pilot study has been conducted to test the clarity and applicability of the tools. It has been conducted on 9 participants (10%) of the sample, no modification done, so, the participants in the pilot study were included in sample size.

Operational design

The operational design included preparatory phase, tool validity and reliability and pilot study.

Preparatory phase

It included reviewing the past and current national and international related literature of various aspects of the study using books, articles, periodicals and magazines to develop tools for data collection.

Content validity and reliability

The tools validity was tested via three expertise in pediatrics health nursing and community health nursing affiliated to Faculty of Nursing, Helwan University to review the clarity, relevance, comprehensiveness and applicability of tools. Regarding reliability, the tools were tested by cronbach's Alpha, it was 0.826 for Knowledge questionnaire and 0.931 for reported practice checklist.

Pilot study

A pilot study has been conducted to test the clarity and applicability of the tools. It has been conducted on 9 participants (10%) of the sample, no modification done, so, the participants in the pilot study were included in sample size.

Work field

The actual field of work was carried out over six months started from March (2021) to the end of August (2021). For data collection, each mother was interviewed and assessed individually using the study tools. The researcher was available at the study setting two days weekly from 9am to 2 pm and started by introducing herself to the mothers then informing them about the aim of the study. The time consumed for completion of the questionnaire format was 10-15 minutes. As regards the mothers' reported practice, time consumed for the checklists was 10-20 minutes.

III- Administrative Design

An official permission was obtained from the Dean of Faculty of Nursing, Helwan University to the director of Ain Shams Pediatrics Hospital to conduct this study, the permission letter included the necessary data, the purpose and nature of the study.

Ethical considerations

An official permission to conduct the proposed study was obtained from the Scientific Research Ethics Committee/ Faculty of Nursing/ Helwan University. Participation in the study was voluntary and subjects were given complete full information about the study and their role before signing the informed consent and that they had the right to refuse to participate. The ethical considerations included explaining the purpose and nature of the study,

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stating the possibility to withdraw at any time, confidentiality of the information where it was not accessed by any other party without taking permission of the participants for research purpose only. Ethics, values, culture and beliefs was respected.

IV-Statistical Design

Data were coded, scored, tabulated, and analyzed by computer using the "statistical package for the social science" (SPSS windows) version11.5. Numerical data were expressed as mean±SD. Qualitative data were expressed as frequency and percentage. Relations between different numerical variables were tested using Pearson correlation. Probability (*p*-value) less than 0.05 was considered significant and less than 0.001 was considered as highly significant

Results

Table (1): Number and percentage distribution of the studied children according to their characteristics (n=91)

Items	No	%
Gender		
Male	56	**61.5**
Female	35	38.5
Age \ years		
Birth to < 6	70	**76.9**
6 < 12	13	14.3
12 ≤18	8	8.8
Mean ±SD	**4.19±3.47**	
Educational level		
Not yet enrolled in school	60	**65.9**
Nursery level	10	11.0
primary level	17	18.7
Secondary level	4	4.4
Birth order		
First	38	**41.8**
Second	20	22.0
Third	13	14.3
Fourth and more	20	22.0
Number of sibling		
No sibling	23	25.3
1:3	58	**63.7**
4:6	10	11.0
Weight/kg compared with normal peers		
Over weight	18	19.8
Normal weight	48	**52.7**
Under weight	25	27.5
Height\ cm compared with normal peers		
Normal	60	**65.9**
Abnormal	40	34.1

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Table (1) showed that, less than two thirds (61.5%) of the children were males and more than three quarters (76.9%) of them aged < 6 years with mean and SD (4.19±3.47). Regarding children's weight and height, this table showed that, more than half (52.7%) of them had normal weight and less than two thirds (65.9%) of them had length <100 cm. As regards sibling number, less than two thirds (63.7%) of them had 1:3 sibling.

Table (2): Number and percentage distribution of studied mothers according to their characteristics (n= 91).

Items	No	%
Age /year		
15 to <20	4	4.4
20 to<30	47	**51.6**
30 to <40	33	36.3
≥40	7	7.7
Mean ±SD	**28.09 ± 6.64**	
Educational level		
Illiterate	27	29.7
Read and write	22	24.2
Intermediate education	29	**31.9**
Higher education	13	14.3
Occupation		
Not working	78	**85.7**
Working	13	14.3
Monthly income		
Sufficient	53	**58.2**
Insufficient	38	41.8
Place of residence		
Rural	73	**80.2**
Urban	18	19.8
Mothers source of knowledge regarding the PKU		
Media	31	34.1
Physicians or nurses	48	**52.7**
Neighbors or friends	12	13.2

As clear from table (2), more than half (51.6%) of the studied mothers aged 20 to less than 30 years with mean age and SD 28.09 ± 6.64. Concerning the mothers' educational level, less than one third (31.9%) of them had intermediate level of education. Also, this table revealed that more than two thirds (85.7 %) of them were not working. Additionally, it was observed that more than half (52.7%) of mothers got their knowledge about PKU from physician or nurses.

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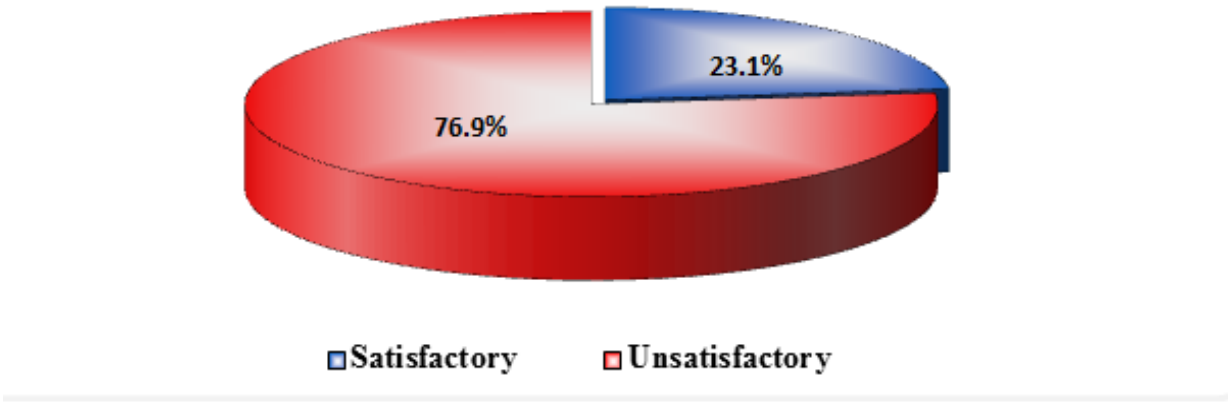

Figure 1: Percentage distribution of the studied mothers' total knowledge regarding care of their children suffering from phenylketonuria (n=91)

Fig (1) showed that more than three quarters (76.9%) of the studied mothers had unsatisfactory level of total knowledge regarding care of their children suffering from phenylketonuria.

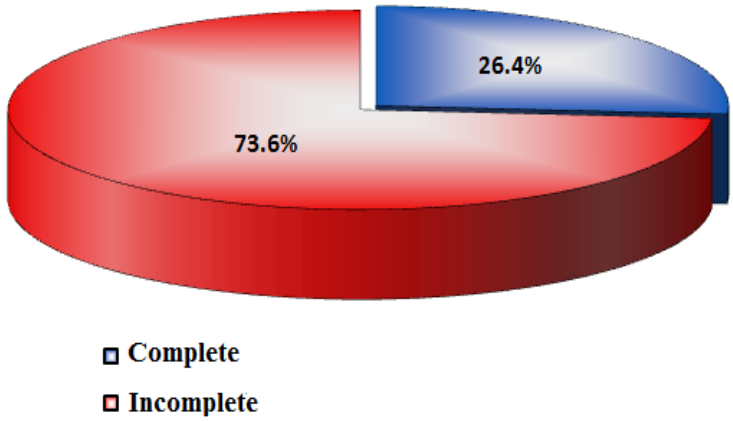

Figure 2: Percentage distribution of the studied mothers' total reported practice regarding phenylketonuria (n=91)

Fig (2) revealed that, less than three quarters (73.6%) of the studied mothers had incomplete level of total practice regarding care of their children suffering from Phenylketonuria.

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Table (3): Relation between the studied mothers' characteristics and their total knowledge about the PKU disease n=91

Items	Total knowledge						
	Satisfactory		Unsatisfactory		Total	Test of significance	
	N	%	N	%		X^2	P-value
Age/year							
15 to<20	1	25.0	3	75.0	4	5.058	0.168
20 to<30	9	19.1	38	80.9	47		
30 to <40	7	21.2	26	78.8	33		
>40	4	57.1	3	42.9	7		
Educational level							
Illiterate	1	3.7	26	96.3	27	26.145	<0.001*
Basic	8	36.4	14	63.6	22		
Intermediate	3	10.3	26	89.7	29		
Higher	9	69.2	4	30.8	13		
Occupation							
Not working	12	15.4	66	84.6	78	18.200	<0.001*
Working	9	69.2	4	30.8	13		
Monthly income							
Adequate	17	32.1	36	67.9	53	5.790	0.016*
Inadequate	4	10.5	34	89.5	38		
Place of residence							
Rural	15	20.5	58	79.5	73	1.330	0.249
Urban	6	33.3	12	66.7	18		

*significant at P≤0.05

As shown from table (3), there was a highly statistically significant difference between studied mothers total level of knowledge, educational level and occupation where p-value <0.001* .Moreover, there was statistically significant relation between total knowledge and monthly income where p-value <0.05*

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Table (4): Relation between the studied mothers' characteristics and their total reported practice regarding PKU n=91

Items	Total reported practice						
	Complete		Incomplete		Total	Test of significance	
	N	%	N	%		X^2	P-value
Age/year							
<20	2	50.0	2	50.0	4	9.822	0.020*
20 to<30	11	23.4	36	76.6	47		
30 to <40	6	18.2	27	81.8	33		
≥40	5	71.4	2	28.6	7		
Educational level							
Illiterate	2	7.4	25	92.6	27	36.479	<0.001*
Basic education	6	27.3	16	72.7	22		
Intermediate education	4	13.8	25	86.2	29		
Higher education	12	92.3	1	7.7	13		
Occupation							
Not working(house wife)	13	16.7	65	83.3	78	26.495	<0.001*
Working	11	84.6	2	15.4	13		
Monthly income							
Adequate	18	34.0	35	66.0	53	3.764	0.052
Inadequate	6	15.8	32	84.2	38		
Place of residence							
Rural	17	23.3	56	76.7	73	1.810	0.179
Urban	7	38.9	11	61.1	18		

*significant at P≤0.05

Table (4) revealed a highly statistically significant difference between mothers' total reported practice and their educational level and occupation where p-value <0.001*. Additionally, there was a statistically significant difference between mothers' total practice and their age where p-value <0.05*

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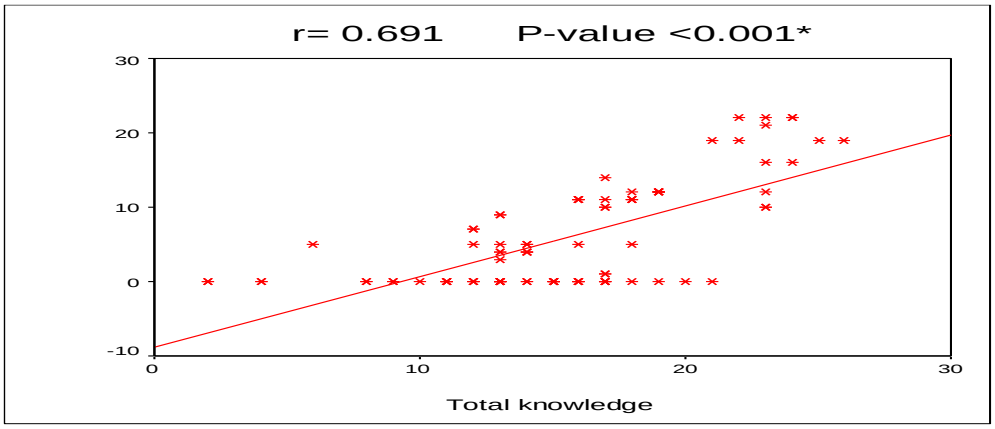

Figure (3): Correlation between studied mothers' total level of knowledge and their total level of reported practice regarding care of their children suffering from Phenylketonuria (n=91)

Fig (3) illustrated that, there was a highly statistically significant correlation between total knowledge of the mothers and their total reported practice where p-value <0.001*, r=0.691.

Discussion

Phenylketonuria is a lifelong condition, which requires strict dietary intervention to control Phenylalanine levels. Children with PKU rely on mothers' knowledge regarding PKU management to maintain their blood Phe levels within the acceptable range **(Van Spronsen et al., 2021).** Therefore, mothers caring for children with PKU should be given adequate guidance and information regarding PKU management. This study aimed to assess mothers' knowledge and reported practice regarding care of their children suffering from Phenylketonuria.

As regards children characteristics, the present study revealed that less than two thirds of the children were males. This result agreed with Fouad & Abd Elmoneem (2016), who conducted a study about "a nursing intervention program for family caregivers of children with Phenylketonuria", and found that more than half of the children were males. From a researcher's perspective, this male predominance in Phenylketonuria cases may be attributed to the X-linked inheritance pattern of the disorder, as the gene responsible for PKU is located on the X chromosome.

The present study revealed that more than three quarter of the children were aged < 6years old with mean and SD about 4.19±3.47. This result agreed with **Yuskiv & Nataliya (2020)** who conducted a comprehensive analysis for current PKU management practices and associated patient and parent reported outcomes in Canada and found that, around three quarters of children were aged 2 to <6 years.

As regards the educational level, the present study revealed that, around two thirds of children were not attending school yet. From the researcher point of view, this may be due to mental and psychosocial problems. This result disagreed with **Abd Alghafar et al., *(2021)*** who conducted a study about "assessment of lifestyle in children with Phenylketonuria" and mentioned that, less than one third of the studied children were not attending school yet. Additionally, this result was congruent with **Williams et al., (2018)** who studied "School performance in early and continuously treated

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Phenylketonuria" and found that, around one third of children did not enroll in school yet.

Regarding the child's birth order, the current study revealed that more than two-fifths of the children were the first baby in the family. From the researcher's point of view, this finding may be related to heightened parental attention and vigilance typically observed with firstborn children, leading to earlier detection and diagnosis of health conditions. This result was congruent with **Pugliese et al., (2020)** who conducted study about "outcomes in pediatric studies of medium- chain acyl-coA dehydrogenase (MCAD) deficiency and PKU" and mentioned that, around one fifth of children were in the first rank in the family birth order.

As regards mothers' characteristics, the present study revealed that one-half of the mothers were aged 20 to >30 years. From the researcher's point of view, this could be due to the fact that women in this age group are more likely to be in their childbearing years and actively engaged in child-rearing responsibilities, which may influence their awareness and involvement in managing their children's health conditions. This result agreed with **Evans et al., (2019)** who studied the "influence of parental food preference and neophobia on children with PKU" and found that, one half of mothers were in age ranged between 20 and 30 years old.

As regards the educational level, the present study revealed that less than one-fifth of mothers had a higher level of education. From the researcher's point of view, this may be due to socioeconomic factors or limited access to educational opportunities in certain regions, which can impact mothers' ability to pursue higher education. This result disagreed with **Carpenter et al., (2018)** who conducted a study about "Parenting a child with PKU" and stated that more than one fifth of the studied mothers' were had higher education.

As regards mothers' occupation, the current study showed that the majority of the studied mothers were not working. From the researcher's point of view, this may reflect societal norms or economic conditions that limit women's participation in the workforce, especially in areas where caregiving roles are predominantly assumed by mothers. This result was consistent to **Shaji et al., *(2021)*** who conducted a study about "health related quality of life of caregivers of children and adolescent with PKU" and mentioned that, the majority of studied mothers were not working. On the other hand, the current study result was congruent with **Fidika et al., (2019)** who found that, around three quarters of parents were employed.

Also, the finding of the current study revealed that more than three-quarters of the mothers lived in rural areas. From the researcher's point of view, this may indicate that rural communities often have less access to healthcare services and educational resources, which could impact both the management of the child's condition and the mothers' awareness of available support systems. This finding was supported with **Abdelaziz et al., (2019)** who conducted a study about "reported Anxiety, depression and coping in parents of children with Phenylketonuria" and found that, more than three quarter of the sample lived in rural area. This result disagreed with **Witalis et al., (2017)** who conducted a study about "Phenylketonuria patients and their parent's knowledge and attitudes to the daily diet -multi-center" and stated that, around one third of parents lives in rural area.

The present study results revealed that, nearly three quarters of mothers had unsatisfactory knowledge about the PKU disease. This finding may be due to lack of

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training programs and instructions for mothers related to PKU disease, their rural residence, and negligence of mothers to gain knowledge. The previous result was in agreement with ***AbdElkodoos et al., (2018)*** who found that, about two thirds of family caregivers had poor knowledge regarding PKU. The current study results were disagree with **Fouad & Abd Elmoneem, (2016**) who found that, almost three fords of mothers had satisfactory knowledge regarding PKU.

As regards total reported practice of the studied mothers regarding care of their children with PKU, it was found that, nearly three quarters of them had incomplete level of practice about PKU. This might be due to faulty technique related to absence of training programs, low educational level, lack of specific PKU teams in rural areas and inadequate guidance by health professionals. This result was agreed with **Akkus et al., (2020)** who found that, the majority of family caregivers had unsatisfactory level of practice regarding PKU. On the other hand, this result was congruent with the study done by **Fouad & Abd Elmoneem, (2016**) who found that, three quarters of family caregivers have adequate total practice regarding PKU.

Regarding the relationship between characteristics of the studied mothers and their total knowledge about PKU, it was found that, there was a highly statistically significant relation between their total knowledge with educational level and occupation. Mothers who had a higher educational level and were working are accompanying with satisfactory level of knowledge. This result was supported by **Ingram & Lauren, (2017)** who conducted a study about "parental knowledge of Phenylketonuria and the effects of Phenylalanine levels on children with Phenylketonuria" and mentioned that, there was a highly statistically significant positive correlation between educational status of the family caregivers and total knowledge scores. This finding goes in the line with ***Asfour, (2013)*** who conducted a study about "the impact of dietary adherence on blood L-phenylalanine Levels among Phenylketonuria children aged 6-18 years in the Gaza Strip" and reported that, family caregiver's knowledge scores increased with their educational levels which may improve the care provided for their PKU children.

The current study showed that, there was a positive relation between total practice of the studied mothers and their age. This result was congruent with ***AbdElkodoos et al., (2018)*** who found that, there was a negative relation between the total practice scores of the family caregivers and their ages.In this regard, ***Witalis et al., (2017)*** reported that there was a negative correlation between the total practice scores of the family caregivers and their ages.

The findings of the current study illustrated that, there was a positive correlation with a highly statistically significant relation between total knowledge of the studied mothers and their total practice. This result indicates that, the mothers' knowledge had an effect on their practice meant while, the mothers have a satisfactory knowledge about their children condition, this will improve their practices regarding care of their children with PKU. This finding was in agreement with **Elsayed et al., (*2020)*** who stated that, good practice was more encountered among those mothers with good knowledge level. Also, these finding was supported by **Fouad & Abd Elmoneem, (2016**) who found that, there was a highly statistically significant relation between the caregivers' total knowledge and their total practices toward the care of their children suffering from PKU.

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Conclusion

The current study findings concluded that, about three quarters of the studied mothers had unsatisfactory knowledge and incomplete reported practice regarding care of their children suffering from PKU. Also, there was a statistically significant difference between studied mothers' total level of knowledge and reported practice and their characteristics.

Recommendations

In the light of the findings of this study, the following recommendations are suggested:

- Constantly training programs to increase awareness of mothers having children with phenylketonuria to ensure enough knowledge and decrease complications that may occur to their children.
- A simplified, comprehensive and clarified Arabic guided picture booklet about PKU must be available in all genetic clinics for each newly admitted child diagnosed with PKU.
- Replication of the study on a large sample in other different setting is highly recommended to assess the needs of mothers who have children suffering from PKU.

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