Assessment of Mothers Care toward their Children having Phenylketonuria

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Abstract

Phenylketoneuria is an autosomal recessive disorder and cause irreversible brain damage if untreated. **Aim:** This study aimed to assess mothers care for children having Phenylketoneuria through assessing the mothers' knowledge, self-reported practice and coping patterns toward their children with PKU. **Design:** A descriptive study. **Sampling:** A purposive sample comprised of 94 mothers accompying children with PKU. **Setting:** This study was conducted at Specialized Genetic Outpatient Clinic, Children's Hospital affiliated to Ain Shams University Hospital. **Data collection tool: 1st tool**: Interviewing Questionnaire Format to assess socio- demographic data of the study children and their mothers, medical history of children with PKU, mothers' knowledge about PKU, assessment of children' health needs and problems, **2nd tool:** Mothers Reported Practice Format related to care of their children with PKU, as diet management and follow up for growth and development as weight, height. **3rd tool**: Psychometric Assessment for mothers as Coping Pattern Scale, Beck Depression Inventory and State-Trait Anxiety Inventory. **Results:** This study revealed that above half of studied children's were females, it ranged from birth to more than 9 years with a mean age of 4.45 ± 2.07 years, the mean age of studied mothers was (32.26 ± 4.2 years), **Conclusion:** about more than half of mothers had poor knowledge about PKU. As well more than one tenth of the mothers had poor total reported practice. There is a significant correlation between mothers' knowledge, practice and their caring roles, anxiety and depression. **Recommendation:** Continuous health teaching to children and their caregivers regarding PKU.

Keywords: Phenylketoneuria, Health Needs, Problems, Dietary Management, Psychometric Assessment

Introduction

Phenylketoneuria is an inborn error of amino acid metabolism caused by mutation in the Phenylalanine Hydroxylase *(PAH)* gene. Mutations in the PAH gene result in decreased catalytic activity affecting the catabolic pathway of Phenylalanine (Phe). Phenylalanine Hydroxylase (PAH) is a hepatic enzyme that requires the cofactor tetrahydrobiopterin (BH_4) to convert Phe to Tyrosine (Tyr). A deficiency in PAH or its cofactor BH_4, results in the accumulation of excess phenylalanine in the blood and can cause severe and irreversible intellectual disability if untreated ***(Saudubray and Garcia-Cazorla, 2018).***

Untreated, PKU is characterized by irreversible intellectual disability,

microcephaly, motor deficits, eczematous rash, autism, seizures, developmental problems, aberrant behaviour and psychiatric symptoms. The precise pathogenesis of brain dysfunction is still unclear. The prevalence of PKU varies worldwide. In Europe, the mean prevalence is approximately 1:10,000 newborns with a higher rate in some countries such as Ireland and Turkey, and a very low rate in Finland ***(Hagedorn et al., 2013).***

The cornerstone of PKU treatment is a low Phe diet in combination with Phe-free L-amino acid supplements. Some PKU centers use casein glycomacropeptide (GMP) or Large Neutral Amino Acids (LNAA) as alternative dietary supplements. Certain patients are responsive to and are treated with BH4, acting as a pharmaceutical chaperone (prescribed as sapropterin dihydrochloride). Possible future treatments include enzyme substitution and gene therapy ***(Trujillano et al., 2014).***

Pediatric nurses can help parents to gain confidence to care for their children by giving appropriate guidance and positive reinforcement. All nurses adapted to children suffering from PKU should be aware of the dietary requirements and restrictions; some medication contains phenylalanine ***(Manta-Vogli, and Schulpis, 2017)***.

Mothers play a pivotal role in the management of childhood phenylketoneuria which needs daily effort to deal with the children disabilities and to maintain a special rehabilitation and diet program ***(Kaakinen et al., 2018).***

Significance of the Study

Phenylketoneuria still has a burden on our Egyptian children especially in Upper Egypt as spread of consanguineous or relative marriage between family members leading to adverse effects related to developmental problems, mental retardation, and behavioral abnormalities. The problem rose in Egypt from late discovery and inefficient Newborn Screening (NBS) program and the mothers not oriented about methods of dealing with daily care related to their children illness. So that the current study is giving the light on the mothers care for their children suffering from Phenylketoneuria and the mothers need to make a firm commitment to this lifestyle change because it's the only way to prevent the serious health problems that children with PKU can develop.

The aim of the study

This study aimed to assess mothers' care for their children having Phenylketoneuria through assessing the mothers' knowledge, self-reported practice and coping patterns toward their children with PKU.

Subject & methods

Design:

A descriptive design was used.

Setting:

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

Subject:

A purposive sample comprised of 94 of mothers accompying children with PKU who attended the previously mentioned setting according to their following criteria:

1- Children suffering from classical PKU and their accompying mothers.

2- Children aged from birth until 18 years of age.

Exclusion criteria: All children having other chronic illness either physical or mental illness.

Tools of data collection:

The data was collected using the following Tools:

Tool I: Interviewing Questionnaire format: It was developed by the researcher after reviewing the recent literature to collect the required data. It included the following parts:

Part one: Socio-demographic data of the children such as: Gender, age, birth order, number of siblings, and educational level.

Part two: Socio-demographic data of the mothers such as: Age, degree of education, work position, place of residence and monthly income.

Part three: Medical history of children with PKU such as: Duration of the disease, date of discovery the disease, follow up frequency per month. In addition to review of the children medical records to obtain information about disease's progress and laboratory investigations.

Part four: Assessment of Mothers' knowledge about PKU as: Definition, causes, signs and symptoms, diagnosis, treatment, diet, complications and implications of the disease.

Part five: It included assessment of children's health needs as: Adequate nutrition, medical care and follow up, sleep, exercise, safety and psychosocial needs.

Part Six: It included assessment of the mothers' knowledge about their role in caring with children with PKU as: Prevention of complications, adequate nutrition, prevention of growth retardation, management of emergencies like fever, prevention of infection, follow up, practicing exercise and maintenance of the child's safety.

Tool II: Mothers Reported Practices format: It was developed by the researcher after reviewing the recent literature to collect the required data as measurement of diet balance, allowed and forbidden food, physical activity, dental care, check of phenylalanine level, follow up of weight at home, follow up of height at home and calculation of body mass index.

Tool III: Psychometric assessment of the mothers; three scales were utilized such as:-

1- Coping Pattern Scale: It is a psychometric scale developed by ***(Jalowic, 1991)*** to assess mothers' coping toward their children suffering from PKU.

2- Beck Depression Inventory (BDI): It is one of the most widely used psychometric tests for measuring the severity of depression, which was developed by (***Beck, et al., 1961)*.**

3- State Trait Anxiety Inventory (STAI): It is a psychological inventory developed by ***(Spielberger et al., 1983)*** used in clinical settings to diagnose state trait anxiety and used as indicator of caregiver distress.

Operational design:

The operational design includes preparatory phase, content validity, reliability, and Pilot study.

Preparatory:

- The study tool was developed by the researcher after through reviewing of recent literatures and reviewed by supervisor.

Content validity and reliability:

It was be done based on result of pilot study and ascertained by a jury of three expertise from Pediatric Nursing and medical to review the tools for clarity, relevance, comprehensiveness, understandable and applicability. For reliability test-retest was done (0.86).

Exploratory Phase:

A pilot study carried out on 10% (9 mothers) accompying their children with PKU during July, 2017 to test applicability, feasibility and clarity of the tool. No radical modification for the tool, so children in the pilot study were included in the study sample.

Field of work: The mothers were asked for an oral consent for participating in the study they were informed that their participation is completely voluntary. Each mother was interviewed individually to collect data.The data was collected individually after a brief explanation of the purpose and the nature of the research .

Questions were clearly explained. Each interview took from 30 - 45 minutes. This study was done two days / weeks (Mondays and Wednesdays) from 9 am to 1 pm. Data was collected over a period of 6 months started (September 2017 and ended in February 2018). Anonymity of mothers' responses was guaranteed and confidentiality of data was maintained.

Administrative Design:

An official approval was obtained to carry out the study that issued from the Dean of the Faculty of Nursing, Ain Shams University to the Directors of the Children's Hospital of Ain Shams University, and to the Director of Genetic Unit.

Ethical Considerations:

Ethical approval was obtained from the Scientific Ethical Committee of Faculty of Nursing, Ain Shams University. In addition, written or oral informed consent was obtained from each participant prior to data collection. They were assured that anonymity and confidentiality that being guaranteed and the right to withdraw from the study at any time. Ethics, values, culture and beliefs were respected.

Statistical Design:

The data obtained was categorized, analyzed using the Statistical Package for Social Science SPSS (version 20). The following statistical analysis measures were used:

Descriptive statistical measures, which included: numbers, percentages, and averages (Minimum, Maximum, Mean ($\overline{X}$), Standard Deviation (SD).

Statistical analysis tests, which included Chi square, Pearson correlation and regression analysis. **Graphical presentation included**: Bar graphs were done for data visualization.

Results

Table (1): Number and percentage distribution of the studied children according to their socio demographic characteristics: N= 94.

children's' characteristics	No	%
Gender		
- Male	32	34.0
- Female	62	66.0
Age (years)		
- Birth < 1 year	10	10.6
- 1 to < 3 years	27	28.7
- 3 to < 6 years	12	12.8
- 6 to < 9 years	41	43.6
- ≥ 9 years	4	4.3
Mean ± SD	4.45 ± 2.07	
Educational level		
- Not attending school	30	31.9
- Preschool	23	24.5
- Primary	41	43.6
Birth order		
- First	40	42.6
- Second	38	40.4
- Third and more	16	17.0
Number of Siblings		
- No Siblings	45	47.9
- One	34	36.2
- Two and more	15	16.0

Table (1) as regards characteristics of studied children, this table showed that (66.0%) of them were females, it ranged from birth to more than 9 years with a mean age of 4.45 ± 2.07 years, their level of education (31.9%) of the children did not attend school yet. Whiles, (42.6%) of them were the first child within their families, regarding to number of siblings (36.2%) of them had one sibling.

Table (2): Number and percentage distribution of the studied mothers according to their socio demographic characteristics: N= 94.

Mothers characteristics	No	%
Age (years)		
- < 20	15	16.0
- 20 to < 30	60	63.8
- 30 to < 40	15	16.0
- ≥ 40	4	4.3
Mean ± SD 32.26 ± 4.2		
Marital status		
- Married	90	95.7
- Divorced / widowed	4	4.3
Educational qualifications		
- Illiterate	5	5.3
- Read & write	20	21.3
- Basic education	2	2.1
- Intermediate education	46	48.9
- Higher education	21	22.4
Occupation		
- Not working (housewife)	61	64.9
- Working	33	35.1
Place of residence		
- Rural	56	59.6
- Urban	38	40.4
Type of dwelling		
- Rent	34	36.2
- Owned	60	63.8
Type of family		
- Nuclear	56	59.6
- Extended	38	40.4
Monthly income		
- Adequate	34	36.2
- Inadequate	60	63.8

Table (2) as regards characteristics of studied mothers, their age were ranged from 20 to ≥ 40 years with a mean of 32.26 ± 4.2 years, (95.7%) of them were married, only 5.3% of them were illiterate, compared to (22.4 %) of them had university education and 59.6% of them lived in rural areas.

Figure (1): Distribution of the studied mothers according to their total knowledge level about PKU: N=94.

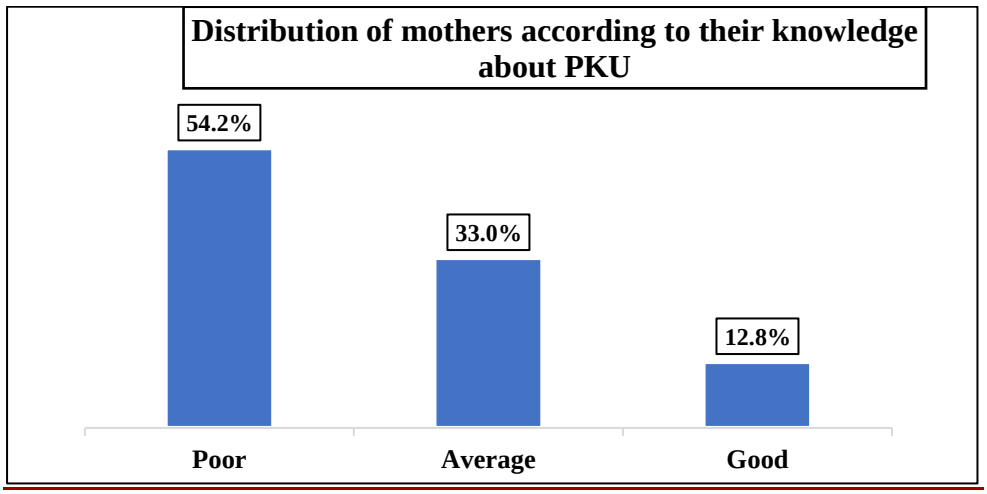
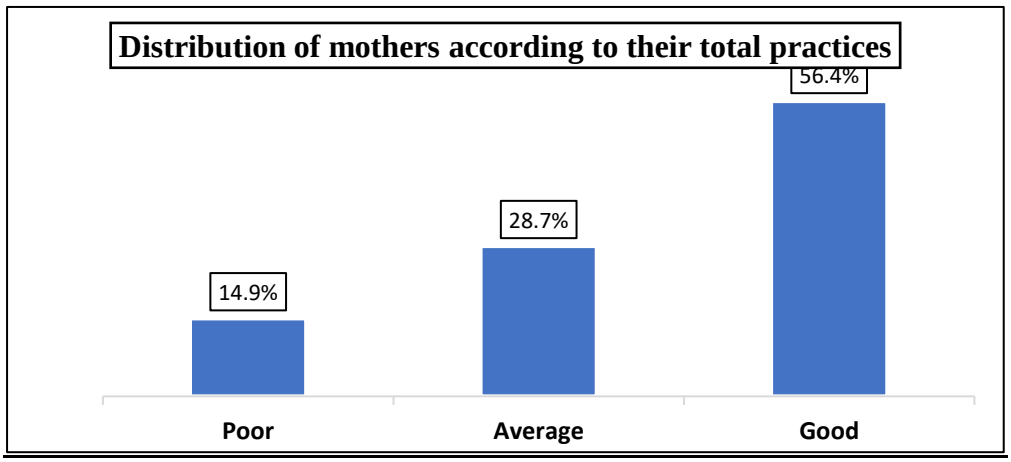


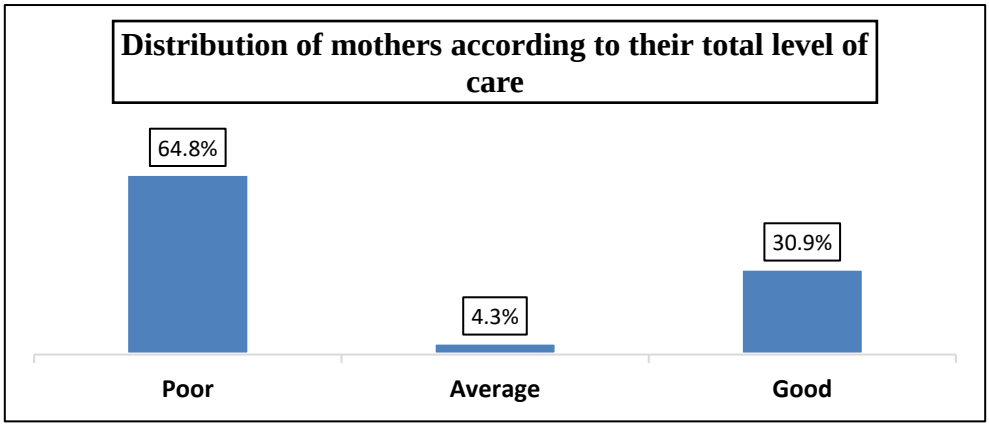
Figure (1) showed that, (54.2%) of the mothers had poor knowledge level, compared to 12.8% of them had good knowledge level. While, those with average knowledge constituted (33.0%) of them.

Figure (2): Distribution of the studied mothers according to their total practice level: N=94.



This figure showed that (14.9%) of the mothers had poor total practice in caring children with PKU and 28.7% of them had average practices. On the other hand, 56.4% of the mothers had good practices.

Figure (3): Distribution of the studied mothers according to their total level of care: N=94.



It was noticed from figure (3) that less than two thirds (64.8%) of the mothers had poor total role in caring children with PKU and 4.3% of them had average caring role. On the other hand, 30.9% of them had good role.

Figure (4): Distribution of the studied mothers according to their total level of coping: N=94.

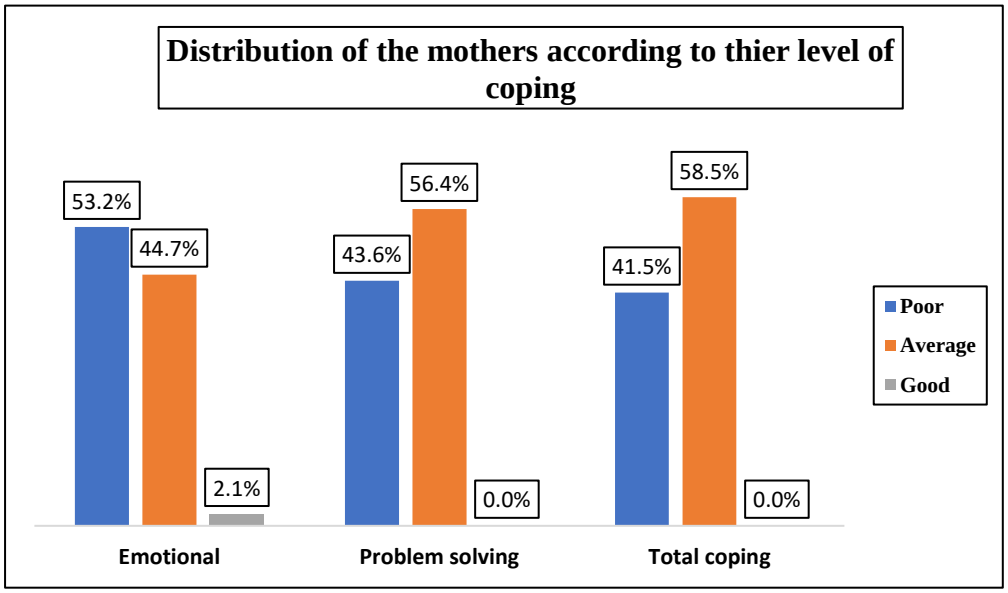


Figure (4) illustrated that (41.5%) of the mothers had poor total coping skills and the rest 58.5% had average total coping skills. While none (0.0%) of them had good total coping skills.

Table (3): Predictors of coping among the mothers of children with PKU using binary logistic regression analysis (Enter method): N=94.

Characteristics	B	S.E.	Wald	Sig.
Child's gender (male/ female)	1.440	2.151	0.448	0.503
Child's age (<1 year, >1 year)	-2.504	1.016	6.081	**0.014***
Child BMI (Normal/ abnormal)	0.761	.859	0.785	0.376
Child's educational level (not enrolled, at school)	3.762	1.500	6.288	**0.012***
Number of siblings(no sibling/one and more siblings)	-1.994	1.448	1.896	0.169
Mother age(<20 years, >20 years)	-4.655	1.739	7.160	**0.007***
Mother education (university education/less education)	1.865	1.450	3.699	**0.054***
Mother job (working/not working)	2.505	1.087	5.312	**0.021***
Marital status (married/widowed or divorced)	33.178	12.99	0.000	0.998
Place of residence (rural/urban)	2.233	1.389	2.586	0.108
Type of family (nuclear/ extended)	3.020	2.194	1.895	0.169
Income (sufficient/ insufficient)	3.583	1.313	7.447	**0.006***
Diagnosis time (<1 year, >1 year)	0.006	1.122	0.000	0.996
Follow up (regular/irregular)	10.61	3.337	10.117	**0.001***
Presence of disease complications (yes/no)	-1.481	1.662	4.084	**0.027***
Child's needs (low/high)	-9.050	3.196	8.018	**0.005***
Mother's knowledge (satisfactory/not satisfactory)	4.909	1.709	8.251	**0.004***
Mother's practice (satisfactory/not satisfactory)	3.209	1.578	3.131	**0.017***
Mother's caring roles (poor/good)	2.478	1.100	5.076	**0.024***
Mother's depression (low/high)	-2.039	1.289	2.502	**0.011***
Mother's anxiety (low/high)	-2.179	1.425	3.016	**0.009***
Constant	-18.21	152.95	0.000	0.999
Model $\chi^2 = 49.193$, $P < 0.0001$ Cox & Snell $R^2 = .566$ Significant at $P \leq 0.05$				

Table (3) illustrates that fourteen variables were found to predictors of good coping among mothers of children with PKU namely child age (P = 0.014), child's level of education (P = 0.012), mothers age (P= 0.007), mothers level of education (P= 0.054), mothers job (P= 0.021), income sufficiency (P = 0.006), regularity of follow up (P = 0.001), presence of complications (P = 0.027), child's needs (P = 0.005), mothers knowledge (P = 0.004), mothers practice level (P = 0.017), mothers caring roles level (P= 0.024), mothers depression level (P= 0.011) and mothers anxiety level (P = 0.009).

Table (4): Correlation between the mothers' level of knowledge, practice, caring roles, coping, anxiety and depression.

Items	Mothers knowledge	Mothers practices	Mothers caring roles	Mothers coping	Mothers anxiety	Mothers depression
Mothers knowledge						
Mothers practices	r = 0.020 P = 0.848					
Mothers caring roles	r = 0.352 P = 0.000*	r = 0.047 P =0.651				
Mothers coping	r = 0.147 P = 0.156	r = 0.112 P = 0.284	r = 0.056 P = 0.295			
Mothers anxiety	r = 0.300 P = 0.003*	r = 0.004 P = 0.968	r = 0.231 P = 0.025*	r = 0.215 P = 0.027*		
Mothers depression	r = 0.334 P = 0.001*	r = 0.025 P = 0.811	r = 0.144 P = 0.054*	r = 0.218 P = 0.035*	r = 0.307 P = 0.003*	

Table (4) portrays a significant correlation between mothers' knowledge and their caring roles, anxiety and depression (P= 0.000, P= 0.003, and P= 0.001 respectively). Furthermore, a statistically significant correlation between mothers' caring roles and their anxiety and depression level (P= 0.025, P= 0.054 respectively). Additionally, mothers coping was significantly correlated with their anxiety and depression levels (P=0.027, and P= 0.035 respectively). Finally, the same table shows a significant correlation between mothers anxiety and their depression level where (P= 0.003).

Discussion

Phenylketonuria disease is a common chronic genetic disorder which needs caregiver's daily effort to maintain a special diet program, rehabilitation and to deal with the child's disabilities.

Family caregivers play a pivotal role in the management of childhood Phenylketonuria. PKU is a chronic disorder which needs family caregivers' daily effort to deal with the child's condition and to maintain a special rehabilitation and diet program.

Concerning mothers' knowledge about PKU nearly more than half of them have poor knowledge about PKU. They got their knowledge mainly from physicians, followed by media and friends. These findings were similar to those of ***AbdElkodoos, Badr- Eldein, and Ismail, (2012) and Chakrapani, (2008)*** who found that most caregivers of PKU children had multiple sources of information about PKU, including health care providers, the internet, support groups, and organizations associated with this condition. These results reflect the need of those families for effective educational programs about proper management of PKU children.

Regarding the mothers' practices and caring role, the current study reveals that less than two thirds of the mothers had a poor caring role and less than one fifth of them had poor practice level especially in measurement of diet and follow of the child growth and development. These findings could be attributed to the nature of the disease PKU; it is among the demanding chronic

illnesses. It poses a big burden on individuals, families and societies. It challenges every fiber of a patient's body and spirit and demands a system of care that ministers to the biological, social and psychological aspects of the illness. It requires vigilant and sustained adherence to a complex and coordinated treatment regimen and daily management comprising multiple health behaviors to reduce patients risk of serious complications ***(Durham et al. 2008, Stanhope and Lancaster, 2008 and Ellis Hartley, 2012).***

Moreover, there were statistically significance relations between the total mothers knowledge and practice scores and the child's age and their age at detection of the disease, where poor knowledge was more encountered among those mothers with children aged less than one year and among those diagnosed at age less than one year. These results were contrary to those of ***Alaei et al. (2011) and MacDonald et al. (2016)*** who found that, family caregiver's knowledge level declines with increasing age of the child at diagnosis and their practice level increase with it.

Additionally, the current study showed that good practice was more encountered among those mothers with good knowledge level and who have good caring role. These results mean that the mothers' knowledge had an effect on their practices, when the mothers have a satisfactory knowledge about their children condition; this will improve their practices regarding care of their children with PKU and enhance their caring roles. This explanation goes in the same line with ***Ozel et al. (2008)*** who mentioned that, when caregivers are provided with the basic knowledge about their children condition, developmental prognosis and various treatment approaches, this will assist them in practicing new and healthy behaviors or also can change unhealthy behavior. Also, these findings are congruent with ***Olsson and Montgomery (2007) and Sharman, Mulgrew and Katsikitis, (2013)*** who found that dietary knowledge are an essential factor on dietary compliance.

When the child is diagnosed with an Inherited Metabolic Disorder (IMD), parents are expected to adapt and cope, integrating generic parenting with specific tasks related to their child's disorder, whilst managing their own emotions and adjusting to their child's diagnosis and prognosis. Actual and perceived high burdens of care are associated with parental stress, and parents of children with chronic conditions experience a high burden of care together with elevated levels of emotional distress and poorer adjustment ***(Fidika, Salewski and Goldbeck, 2013 and Finan et al., 2015).***

This would explain the results of the current study where the majority of the mothers had moderate anxiety and less than half had moderate depression which were reflected on their coping strategies as less than half of them had poor coping with their disease. Similar findings were reported by ***Gentile, Hoedt and Bosch, (2010), Harding and Blau, (2010) and George et al. (2007)*** who found that most parents of PKU children report significant care demands, whilst some report similar or even lower levels of stress and poorer coping when compared to parents of healthy children and parents of other inherited metabolic disorders.

This picture of parental stress and improper coping could be complicated by lack of knowledge about the disease and reflected on the mothers practice. Knowledge of one's disease may increase successful management of that disease

and lessen the stress associated with it and enhance proper coping with it ***(Hoedt et al., 2011).*** This picture was portrayed in the current study finding where, poor mothers' knowledge was more encountered among those mothers with higher level of anxiety and depression, those with poor coping and those with poor practice and among those mothers who expressed high needs of PKU children. Similar finding were found by ***Zeltner et al. (2014), Fidika, Salewski and Goldbeck, (2013) and Fouad and Abd Elmoneem, (2016).***

Conclusion

Upon the findings of the current study, concluded that more than half (54.2%) of the mothers had unsatisfactory knowledge about phenylketonuria. As well more than one tenth (14.9%) of the mothers had poor total reported practice. Moreover there is a significant statistical relation between mother's characteristics as age, educational level, work, and residence and their total knowledge and total reported practice. Similar there is a significant correlation between mothers' knowledge, reported practice and their caring roles, anxiety and depression.

Recommendations

In view of the study findings, the following recommendations are suggested:

- Gene testing (molecular analysis) is very essential, prenatal testing to help for confirm diagnosis of suspected PKU cases and proper genetic counseling.
- Early detection and proper treatment of suspected PKU patients to guard against cognitive impairment.
- Appropriate intellectual and mental health assessments are an important component of care for children affected with PKU.
- Frequent growth monitoring to avoid obesity among PKU children as essential care component.
- Continuous health teaching to children with phenylketoneuria and their caregivers regarding phenylketoneuria, its complication, management, designing an education handout about phenylketoneuria & its management plan and allocated for affected children and their caregivers.
- Awareness of the nursing intervention program in all maternal and child health care centers for families having children with PKU to raise their knowledge about the disease, its management and prevention of complications.
- Further researches are needed for proper data analysis and collection and to identify the common genetic mutations among Egyptian patients.

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