Relationship between socioeconomic status and quality of life of children with phenylketonuria

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Phenylketonuria

- Phenylketonuria is a complex autosomal recessive metabolic disorder caused by mutations of the gene encoding for phenylalanine hydroxylase enzyme. Phenylalanine hydroxylase deficiency leads to an accumulation of phenylalanine in blood and brain that gradually impairs metabolic functioning and cognitive development
- An early dietary intervention based on a phenylalanin restricted diet and the supplementation of an amino acids mixture is able to keep phenylalanine blood levels low, avoiding mental retardation or neurological abnormalities that have been described in untreated patients

Phenylketonuria and quality of life

- Diet should be maintained lifelong and this requires a lot of effort, especially in adulthood, owing to the growing instances of social relationships.
- PKU patients must go through frequent biochemical controls, several dietary assessments and adjustments that represent stressors for both adolescent and adult patients. Furthermore, in young adults, the strict adherence to diet may lead to some difficulties in building up good social relationships, often resulting in poor adherence to dietary regimen with risks of cognitive, neuropsychological and behavioral impairment.
- QoL scores of PKU patients and their parents measured by current inventories are reported to be generally similar to healthy controls.

Aim of the study

- to investigate the relationship between sociodemographic status and quality of life among patients under 12 years old who were diagnosed with PKU and treated early by a single multidisciplinary team, in order to contribute to understanding of the effects these factors have on PKU patients treated early.
- Objectives: - to describe the sociodemographic status of PKU and their parents
- - to assess the quality of life in PKU children
- - to identify the perceived health status in PKU children

Methods

- This **descriptive study** was conducted from March 2014 till January 2015, as a result of the collaboration of the University of Medicine and Pharmacy Carol Davila Bucharest with the Institute for Mother and Child Protection Bucharest (enrolling pediatric patients)
- **Patients**: 44 under 12-year-old phenylketonuria patients treated at IOMC were invited to take part in the study. (all the patients were included in the National Program for Rare Diseases)
- *Inclusion criteria* were: patients receiving treatment at IOMC, who were free from other diseases that compromise cognitive and physical development; and whose parents or legal guardians signed free and informed consent forms.
- *Exclusion criteria* were no free and informed consent form; child unwilling to take part in the study

Technical instruments

- **EQ-5D-3L** (a standardised measure of health status developed by the EuroQol Group) consists of **the EQ-5D descriptive system** (comprises the following *5 dimensions*: mobility, self-care, usual activities, pain/discomfort and anxiety/depression), each dimension has *3 levels*: no problems, some problems, extreme problems and **the EQ visual analogue scale** (EQ VAS - a vertical, visual analogue scale where the endpoints are labelled 'Best imaginable health state' – 100 points and 'Worst imaginable health state' 0 points.

Sociodemografic status of the children

Variables	Strata	No.	Mean±SD	Sig.
Gender	Female	26		0,291
	Male	18		
Age (months)			48,36±44,05	0,021
Diagnosis age (months)			9,75±24,84	<0,001
Education level	No (under age)	24		<0,001
	Yes, normal school	10		
	Yes, special school	4		
	No, cause of disease	6		
Residence area	Rural	28		0,097
	Urban	16		

Sociodemographic status of the parents

Variables	Strata	No.	Mean±SD	Sig.
Gender	Female	40		<0,001
	Male	4		
Age (years)			34,36±8,61	0,249
Education level	Primary school	4		0,201
	Secondary school	10		
	Tehnical school	6		
	Highschool	12		
	Community College	8		
	University	4		
Relantionship with the child	Parent	38		<0,001
	Grandparent	2		
	Others	4		

Frequency of levels 1, 2 and 3 by dimension and by gender

EQ-5D DIMENSION		GENDER		Exact sig. (2-sided)
		MALE	FEMALE	
MOBILITY	Level 1	14	20	0,706
	Level 2	5	5	
	Level 3	0	0	
SELF-CARE	Level 1	14	20	0,869
	Level 2	2	2	
	Level 3	2	4	
USUAL ACTIVITIES	Level 1	16	18	0,179
	Level 2	0	4	
	Level 3	2	4	
PAIN/ DISCOMFORT	Level 1	14	16	0,353
	Level 2	4	8	
	Level 3	0	2	
ANXIETY/ DEPRESSION	Level 1	14	18	0,474
	Level 2	4	6	
	Level 3	0	2	

Frequency of levels 1, 2 and 3 by dimension and by area of residence

EQ-5D DIMENSION		AREA OF RESIDENCE		Exact sig. (2-sided)
		URBAN	RURAL	
MOBILITY	Level 1	12	22	0,454
	Level 2	5	5	
	Level 3	0	0	
SELF-CARE	Level 1	12	22	0,834
	Level 2	2	2	
	Level 3	2	4	
USUAL ACTIVITIES	Level 1	14	20	0,266
	Level 2	0	4	
	Level 3	2	4	
PAIN/ DISCOMFORT	Level 1	10	20	0,329
	Level 2	6	6	
	Level 3	0	2	
ANXIETY/ DEPRESSION	Level 1	10	22	0,141
	Level 2	6	4	
	Level 3	0	2	

Correlation between sociodemographic status and EQ VAS

		EQ VAS	Child age (months)	Diagnosis age (months)	Age (parent)	Education level
EQ VAS	Pearson Correlation		-,564**	-,409**	,084	,094
	Sig. (2-tailed)		,000	,009	,588	,543
	N		44	40	44	44
Child age (months)	Pearson Correlation	-,564**		,541**	,119	-,293
	Sig. (2-tailed)	,000		,000	,440	,053
	N	44		40	44	44
Diagnosis age (months)	Pearson Correlation	-,409**	,541**		-,140	-,052
	Sig. (2-tailed)	,009	,000		,388	,748
	N	40	40		40	40
Parent age (years)	Pearson Correlation	,084	,119	-,140		,316*
	Sig. (2-tailed)	,588	,440	,388		,037
	N	44	44	40		44
Education level	Pearson Correlation	,094	-,293	-,052	,316*	
	Sig. (2-tailed)	,543	,053	,748	,037	
	N	44	44	40	44	

**. Correlation is significant at the 0.01 level (2-tailed).

*. Correlation is significant at the 0.05 level (2-tailed).

Conclusions (I)

- It was observed a positive correlation between parents' schooling and child's motor performance, usual activities, self-care and negative correlation betweeen pain/discomfort and anxiety/ depression. PKU children whose parents have more schooling tend to show better quality of life scores. The program for PKU should continue to focus on orienting and supporting parents who have less schooling.

- The four children with low quality of life scores showed a critical condition in terms of the study variables: they are institutionalized, their diet is not maintained all the time and the perception about their quality of life is measured differently (the nurses completed the questionnaire).

Conclusions (II)

- In 75% of the cases studied, treatment started within the first month of life. The correlation between quality of life and start of treatment clearly shows that an efficient PKU program requires an active case search and a focus on immediate treatment adherence. It is crucial to have an integrated, complex and multidisciplinary system in place in order to handle cases in timely fashion.

- There was no significant relationship between quality of life and family income in the cases studied. This reveals the efficiency of the support provided to low-income families served by the program, a basic condition for prevention and health promotion measures to be developed successfully due to the high cost of diet therapy.

THANK YOU!