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UNTREATED GERIATRIC PHENYLKETONURIA WITH DELAYED DIAGNOSIS: AN ICF-BASED CASE REPORT

ORIGINAL ARTICLE

ABSTRACT

Purpose: Due to the nature of the phenylketonuria (PKU), it is difficult to categorize these patients' needs in a treatment management. The aim of this study was to evaluate an untreated geriatric PKU patient based on International Classification of Functioning, Disability and Health (ICF).

Method: The 72-year-old patient was presented with late-diagnosed and untreated PKU during his lifetime. The body structure and functions of the patient were determined by the Tinetti Scale, Sit-up test for 30-second, The Upper Extremity Index, Barthel Daily Life Activity Scale, Short Form-36 Quality of Life scales were used to determine activities and limitations. Functional Ambulation Scale and Barthel Index were used for assessing participation.

Results: In this case, physical disabilities caused problems in his self-care and participation in life accompanying with cognitive problems, environmental and personal factors are negatively affecting his quality of life.

Conclusion: Many aspects of the geriatric PKU's participation in life and the parameters that need to be evaluated remain uncertain. For this reason, we suggest that healthcare professionals working with individuals with geriatric PKU patients might follow the ICF qualifiers and assessment methods as in this case report.

Keywords: Case Base Studies, Disability and Health, International Classification of Functioning, Phenylketonuria

GECİKMİŞ TANI İLE TEDAVİ EDİLMEYEN GERİATRİK FENİLKETONÜRİ: ICF TEMELLİ BİR VAKA RAPORU

ARAŞTIRMA MAKALESİ

ÖZ

Amaç: Fenilketonüri (PKU) hastalığının doğası gereği, bu hastaların tedavi ve ölçüm seçeneklerini belirli bir şekilde kategorize etmek zordur. Bu çalışmanın amacı, tedavi edilmemiş geriatrik fenilketonüri hastasını Uluslararası İşlevsellik, Yetiyitimi ve Sağlık Sınıflandırmasına (ICF) göre değerlendirmektir.

Yöntem: Yaşamı boyunca spesifik tedavi görmemiş yetmiş iki yaşındaki bir fenilketonüri hastası sunulmaktadır. Hastanın vücut yapısı ve fonksiyonları Tinetti Skalası ve 30 saniyelik otur-kalk testi ile belirlendi. Aktiviteleri ve kısıtlamaları belirlemek için Üst Ekstremite İndeksi, Barthel Günlük Yaşam Aktivite Ölçeği, Kısa Form-36 Yaşam Kalitesi ölçekleri kullanıldı. Katılım için Fonksiyonel Ambulasyon Skalası ve Barthel İndeksi kullanıldı.

Sonuçlar: Bu vakada fiziksel engellilik öz bakımında sorunlara neden olmakta ve bilişsel sorunlar eşliğinde hayata katılımı, çevresel ve kişisel faktörler yaşam kalitesini olumsuz etkilemektedir.

Tartışma: Geriatrik fenilketonürinin hayata katılımının birçok yönü ve değerlendirilmesi gereken parametreler belirsizliğini korumaktadır. Bu nedenle, literature fenilketonüri hastası olan bireylerle çalışan sağlık çalışanlarının bu vaka raporunda olduğu gibi ICF niteleyicileri ve değerlendirme yöntemlerini takip etmelerini öneriyoruz.

Anahtar kelimeler: Vaka Temelli Çalışmalar, Engellilik Ve Sağlık, Uluslararası İşlevsellik Sınıflandırılması, Fenilketonüri

INTRODUCTION

Phenylketonuria (PKU) is a hereditary disease caused by a high level of phenylalanine in the blood due to the lack of phenylalanine hydroxylase enzyme, which converts the amino acid of phenylalanine to the tyrosine amino acid (1).

Classification of the problems existing in adult PKU patients may show different physical and mental characteristics due to the nature of the disease and low-phenylalanine diet treatment processes taken or not (2). The geriatric phenylketonuria case at this age according to the International Classification of Functionality, Disability and Health (ICF) is the first report presented in the national and international PKU literature, with a delayed diagnosis (at 30 years old) and any received dietary supplement treatment for his whole life.

Case presentation

A 72-year-old male patient with phenylketonuria first diagnosed at the age of 30 years old. The patient did not receive any diet therapy suitable for his disease, had reading and writing difficulties due to his mild perception/mental problem. According to his medical records, cognitive measures like working memory, attention and response inhibi-tion were reported to range between mild to severe. His intelligence quotient (IQ) was additionally noted as "54" which is comparatively lower than healthy peers. He is a retired person and married with healthy two sons. The patient tries to do his self-care and activities of daily living (ADL), but he is dependent on his caregiver (wife) and children.

METHODS

The study was carried out at Hacettepe University, Faculty of Physical Therapy and Rehabilitation. The research design was designed as a cross-sectional case study. All assesments were made after being informed in detail and the patient and his caregiver's approval were obtained. Measurement and evaluation tools were introduced to patient and his caregiver (his wife). The Turkish version of the scales was used. The assessments classifications were schematized according to the ICF Guidelines established by the World Health Organization (2001) (Figure 1).

Description of assessment tools

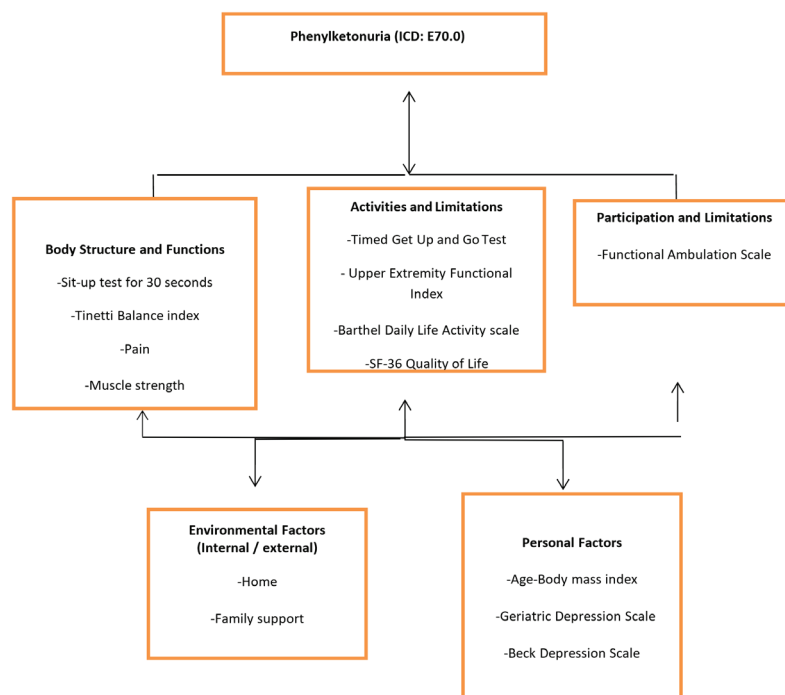Data sets were organized suitable methods in the ICF (Table 1).

Figure 1. ICF Based Classification of Assessments in PKU

The Upper Extremities Function Index-15 (UEFI-15) was used to evaluate the upper extremity function (3).

The Barthel Index was used to measure the functional independence of the case in ADL (4).

The Functional Ambulation Scale was used to assess the basic motor skill level required for the case to walk in everyday life (5).

The Tinetti Scale was used to evaluate the balance of the case in daily life (6).

The Short Form – 36 Scale was used to evaluate the health-related quality of life (7).

The Beck Depression Scale is a 21-item scale that measures the degree of anxiety in adults (8).

The Geriatric Depression Scale was used to evaluate the mental health and depression level (9).

30-Second Chair Stand Test and the Timed Up and Go, were used to measure the functional level (10, 11).

RESULTS

Body Functions and Structures

He scored 7 points in the 30 Second Chair Stand Test test, which indicates lower limb muscle weakness. Tinetti Balance and Gait Test score 16 was found to be high in our patient.

Activities

The Barthel ADL score was 75 in total, indicating that it was moderately dependent. The Upper Extremity Functional Index score was 47.6 out of 100. This score indicates that our patient had difficulty in upper limb activities. He had difficulties in carrying out body pain and energy deprivation activities in health-related quality of life assessment; scores other than emotional role were lower than men in Turkish society (Figure 2) (12).

Participation

According to the Functional Ambulation Classification, (0-5) got 3 points; this result shows that having someone during walking gives him confidence and is dependent on surveillance.

Personal Factors

Our case had a physical and cognitive deficiency, which we thought started earlier than the age, due to the negativity of not being able to receive diet therapy. Apart from this, as a result of the Beck Depression Scale (33 points), he had a severe anxiety. The definitive depression in the Geriatric Depression Scale (22 points) supported this.

Table 1. Data Sets Selected for the Case With Phenylketonuria

Domains	Codes	Qualifiers	Assessment Methods
	b2800.1	Sensation of pain	SF-36
	b730.2	Muscle power functions	Sit-up test for 30 seconds
Body functions (b)	b780.3	Sensations related to muscles and movement functions	Tinetti Balance Test
	b7356.0	Tone of all body muscles	Ashworth Scale
Body structures (s)	s7600.1	Structure of trunk	Observational
	d599.2	Selfcare	Barthel ADL Index
	d550.1	Eating	Barthel ADL Index
	d5101.2	Washing oneself	Barthel ADL Index
	d2400.2	Handling stress and other psychological demands	SF-36
Activities and Participation (d)	d4500.2	Walking	Barthel ADL Index
	d5400.1	Dressing	Barthel ADL Index
	d7200.1	Forming Relationship	SF-36
	d910.2	Community life	SF-36
EnvironmentalFactors (e)	e410.2	Individual attitudes of immediate familymembers	Caregiving OutcomesScale

Abbreviations: SF-36: Short Form 36, ADL: Activities of Daily Living

Table 2. Scale Results of the Case and Cut Off Values

	Measurements	Case results	Cut off values
Body Functions and Structures	30 Second Chair Stand Test Tinetti Balance and Gait Test	7 second 16 points	<11: below average <18: High risk of falling
Activities	Barthel ADL score The Upper Extremity Functional Index score	75 points 47.6 points	61-90: moderate dependency 0: lowest 80: highest
Participation	Functional Ambulation Classification	3 points	0: Nonfunctional ambulator 5: Independent
Personal Factors	Beck Depression Scale Geriatric Depression Scale	33 points 22 points	29-63: severe depression >14: definite depression
Environmental Factors	Observation		

Environmental Factors

The ergonomic arrangement at home was not designed for the patient. Her caregiver stated that she had difficulties in patient care and was sometimes psychologically uncomfortable with this situation.

The scale results and cut-off values of the case are shown in Table 2.

DISCUSSION

In this case report, a geriatric untreated late diagnosed Phenylketonuria patient has been evaluated according to the ICF guidelines. In addition to his physical deficiencies due to the disease, we also examined limitations with the environmental and personal factors required for participation in cognitive, social and daily life activities. Basically, in our case, physical disabilities caused problems in his self-care and participation in life accompanying with cognitive problems, environmental and personal factors are negatively affecting his quality of life. Mazur et al. evaluated the degree of independence with the Barthel ADL Scale and the Tinetti Balance Scale in 11 adults untreated PKU patients (13). They stated that 8 of these individuals had moderate insufficiency and 3 had mild insufficiency, and 6 people had a high risk of falling and 5 people had a moderate risk of falling. The average age of adults in this study is 47.5. Compared to this study, the fact that our case had a higher age supports high risk of falling and high level of disorder. However, the fact that our patient is geriatric should also be considered.This is not the first study that investigate PKU from the ICF perspective. Bingöl et al. classified a young PKU patient according to

ICF criteria (14). However, our case is unique with being an untreated late diagnosed geriatric PKU, who was classified according to ICF criteria.

In our case, moderate to severe intellectual disability reported in his previous medical reports which might be a risk factor for functional limitations in daily activities. It should be underlined that the presence of physical and functional deficiencies of our case compared to healthy peers does not exclude the coexistence of neurocognitive deficits caused by PKU. Despite being an untreated and late-diagnosed geriatric case, he could find opportunity to have his retirement from a job, married and had children that could be a significant indicator to be participated in real life.

Considering the limited comparable literature, our case is unsurprisingly lower than its peers in terms of body structure and functions (premature aging), considering its age and its lack of diagnosis and treatment processes, mostly dependent on the caregiver in daily life activities and self-care. When his personal and environmental factors are combined with the processes caused by his disease, it was seen that his mental health deteriorated in a similar way by negatively affecting the quality of life. It is known that PKU patients who receive treatment at an early age experience less mental and neurological disorders (15). The absence of any treatment in our case may have triggered more of these problems. Prolonged diagnosis may be a limitation of the study.

New problems arise with the aging of PKU patients. Information about holistic problems in a geriatric individual at PKU will guide clinicians. Many aspects of the Geriatric PKU's participation in life and

the parameters that need to be evaluated remain uncertain. For this reason, we recommend that healthcare professionals working with individuals with adult PKU follow the assessment processes of patients according to the ICF guidelines, as in this case report.

REFERENCES

1. ten Hoedt AE, de Sonneville LM, Francois B, ter Horst NM, Janssen MC, Rubio-Gozalbo ME, Wijburg FA, Hollak CE, Bosch AM. High phenylalanine levels directly affect mood and sustained attention in adults with phenylketonuria: a randomised, double-blind, placebo-controlled, crossover trial. J Inherit Metab Dis. 2011; 34(1):165-71.

2. Trefz F, Maillot F, Motzfeldt K, Schwarz M. Adult phenylketonuria outcome and management. Mol Genet Metab. 2011;104:S26-30.

3. Hamilton CB, Chesworth BM. A Rasch-validated version of the upper extremity functional index for interval-level measurement of upper extremity function. Phys Ther. 2013;93(11):1507-19.

4. Bouwstra H, Smit EB, Wattel EM, van der Wouden JC, Hertogh CMPM, Terluin B, Terwee CB. Measurement Properties of the Barthel Index in Geriatric Rehabilitation. J Am Med Dir Assoc. 2019;20(4):420-25.e1.

5. Silva ADM, Furtado G, Dos Santos IP, da Silva CB, Caldas LR, Bernardes KO, Ferraz DD. Functional capacity of elderly with lower-limb amputation after prosthesis rehabilitation: a longitudinal study. Disabil Rehabil Assist Technol. 2021;16(5):556-60.

6. Gümüş E, Arslan İ, Tekin O, Fidancı İ, Eren ŞÜ et al. Kendi evi ve huzurevinde yaşayan yaşlılarda, denge ve yürüme skorları ile düşme riskinin karşılaştırılması. Ankara Med J. 2017; 2:102-10.

7. Lærum-Onsager E, Brovold T, Bergland A, Pripp AH, Bye A. Associations between health-related quality of life, body mass index, health status and sociodemographic variables in geriatric patients and non-hospitalized older people: A comparative cross-sectional study. Nutr Health. 2020;26(2):141-150.

8. v von Glischinski M, von Brachel R, Hirschfeld G. How depressed is "depressed"? A systematic review and diagnostic meta-analysis of optimal cut points for the Beck Depression Inventory revised (BDI-II). Qual Life Res. 2019;28(5):1111-8.

9. Parlar Kılıç S, Karadağ G, Koçak HS, Korhan EA. Investigation of the old age perceptions with the loneliness and depression levels of the elderly living at home. Turk Geriatri Derg. 2014;17(1):70-6.

10. Püllüm E, Sökmen ÜN, Çevik Akyıl R, Ongun G, Özdemir Cihangir H. Aydın Huzurevinde düşme riski taşıyan yaşlıların belirlenmesi: tanımlayıcı çalışma. İzmir Kâtip Çelebi Üniversitesi Sağlık Bilimleri Fakültesi Derg. 2018; 3(2):9-14.

11. Ibrahim A, Singh DKA, Shahar S. 'Timed Up and Go' test: Age, gender and cognitive impairment stratified normative values of older adults. PLoS One. 2017;12(10):e0185641.

12. Demiral Y, Ergor G, Unal B, Semin S, Akvardar Y, Kivircik B, Alptekin K. Normative data and discriminative properties of short form 36 (SF-36) in Turkish urban population. BMC Public Health. 2006;6:247.

13. Mazur A, Jarochowicz S, Ołtarzewski M, Sykut-Cegielska J, Kwolek A, O'Malley G. Measurement of functional independence level and falls-risk in individuals with undiagnosed phenylketonuria. Acta Biochim Pol. 2009;56(4):613-8.

14. Bingöl H, Günel MK. Functional Status in a Teenager with Phenylketonuria from the ICF Perspective. J Child Sci. 2017;7:14-8.

15. Romani C, Palermo L, MacDonald A, Limback E, Hall SK, Geberhiwot T. The impact of phenylalanine levels on cognitive outcomes in adults with phenylketonuria: Effects across tasks and developmental stages. Neuropsychology. 2017;31(3):242-54.