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(RESEARCH ARTICLE)

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Pediatrics 2 experience, Neuropediatrics and neurometabolic diseases unit on phenylcetonuria: About 36 patients

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Abstract

Phenylketonuria (PKU) is a hereditary metabolic disease of phenylalanine metabolism, linked to a deficiency of phenylalanine hydroxylase or its cofactor, an enzyme allowing the transformation of phenylalanine into tyrosine, resulting in an increase in blood and brain concentration. of PHA, comprising an intellectual disability with cognitive and behavioral disorders if it is not treated. We report 36 cases of phenylketonuria followed in the pediatric department 2, born to consanguineous parents in 58.4% of cases and non-consanguineous in 41.6%, the male sex represents the majority with 22 cases and female with 14 cases, The age of diagnosis minimum 3 days of life and maximum 16 years and the current age of patients minimum 4 years and maximum 37 years with comorbidities like epilepsy, motor disorders, behavioral disorders, Autism Spectrum Disorder and mental retardation and about 70% have a regular monitoring. Through this series we will support the epidemiological, clinical, paraclinical and therapeutic particularities of patients with phenylketonuria. Early diagnosis of the disease makes it possible to initiate treatment early, which is mainly based on a diet low in phenylalanine for life in order to improve the progressive prognosis and avoid irreversible after-effects.

Keywords: Phenylketonuria; Phenylalanine; Phenylalanine Hydroxylase; Tetrahydrobiopterin

1. Introduction

Phenylketonuria (PKU) is a hereditary metabolic disease of phenylalanine metabolism, linked to a deficiency of phenylalanine hydroxylase or its cofactor, an enzyme allowing the transformation of phenylalanine into tyrosine, resulting in an increase in blood and brain concentration of PHA, comprising an intellectual disability with cognitive and behavioral disorders if it is not treated. Neonatal biological screening for the disease and rapid treatment prevent irreversible brain damage and allow an almost normal life. (1, 8, 12)

2. Materials and methods

We report 36 cases of phenylketonuria followed in the pediatric department 2, born to consanguineous parents in 58.4% of cases and non-consanguineous in 41.6%, the male sex represents the majority with 22 cases and female with 14 cases(table 1), The age of diagnosis minimum 3 days of life and maximum 16 years (table 2) and the current age of patients minimum 4 years and maximum 37 years (table 3) with comorbidities like epilepsy, motor disorders, behavioral disorders, Autism Spectrum Disorder and mental retardation (table 4) and about 70% have a regular monitoring.

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2.1. Représentative graphs

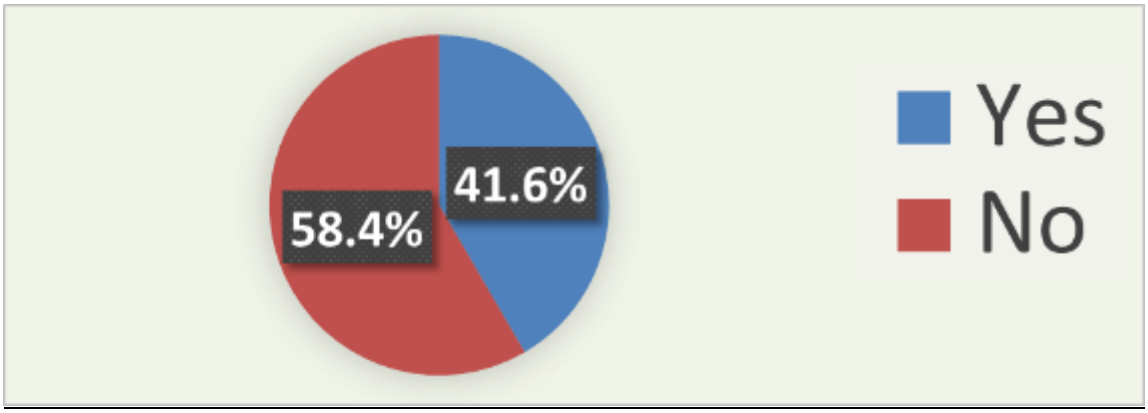

Figure 1 Consanguinity

Table 1 Distribution according to sex

Sex	Number	percentage
Feminine	14	38.9%
Masculine	22	61.1%

Table 2 Age of Diagnosis

Age	
Minimum	3 days of life
Maximum	16 years
Medium	3.5 years

Table 3 Age current patients

Age	
Minimum	4 years
Maximum	37 years
Medium	13 years

Table 4 Comorbidities

COMORBIDITIES	NUMBER
Good psychomotor development (GPD)	2
Epilepsy	7
Motor disorders	9
Autism Spectrum Disorder (ASD)	11
Behavioral Disorders	32
Mental retardation	34

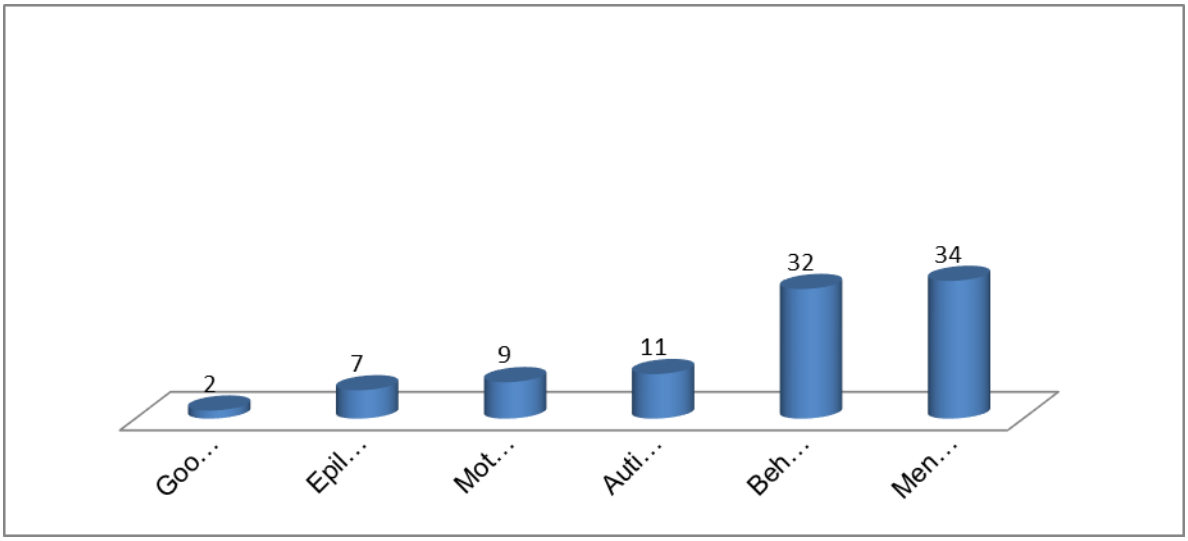

Figure 2 The photos are of a girl who has progressed badly due to poor compliance with the dietary regime.

Figure 3 The photos show a boy who has progressed well under a well-monitored diet**.**

3. Discussion

Through this series we will support the epidemiological, clinical, paraclinical and therapeutic particularities of patients with phenylketonuria.

Phenylketonuria (PKU) is due to a deficiency of a liver enzyme: phenylalanine hydroxylase (PAL) which allows the transformation of phenylalanine into tyrosine. (1) Tetrahydrobiopterin (BH4) is the essential cofactor for this hydroxylation reaction .(8)

Untreated phenylketonuria causes serious neurological disorders such as mental retardation, behavioral disorders, psychoses, flexion spasms, epilepsy and is associated with skin appendage disorders with global hypopigmentation . Approximately 25% of cases develop “grand mal” epilepsy, other neurological signs (extrapyramidal, global hypertonia, pyramidal syndrome, tremors, parkinsonian syndrome). (2 ;9)

phenylketonuric children are taken care of from birth. (3)

PKU is a hereditary condition transmitted in an autosomal recessive manner. The PAH gene is located on chromosome 12 at 12q24.1. Around 500 different mutations have been described (4), with an imperfect genotype/ phenotype correlation. (5)

The diagnosis is based on the detection of a high phenylalanine level and a normal or low tyrosine level. Routine newborn screening (3,7)

The treatment consists of a diet low in phenylalanine and makes it possible to maintain a good nutritional balance and to obtain almost normal physical and intellectual growth and must be continued for life. (6,10,11,12)

4. Conclusion

Early diagnosis of the disease makes it possible to initiate treatment early, which is mainly based on a diet low in phenylalanine for life,in order to improve the progressive prognosis and avoid irreversible after-effects.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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