

# International Database of BH4-Responsive HPA/PKU "BIOPKU"



ID #:

For internal use only

## Prof. Dr. Nenad Blau

Division of Clinical Chem. & Biochemistry  
University Children's Hospital  
Steinwiesstrasse 75  
CH-8032 Zürich / Switzerland  
Tel.: +411 266 7544  
Fax: +411 266 7169  
E-mail: [blau@kispi.unizh.ch](mailto:blau@kispi.unizh.ch)  
URL: <http://www.bh4.org>

## PATIENT'S IDENTIFICATION

Last name:  male  
First name:  female  
Birth date (dd/mm/yy):  
Mother: Age: Ethnic origin:  
Father: Age: Ethnic origin:  
Are parents consanguineous?  yes  no  
Total number of life births (including patient):

### Type of HPA:

MPH  Mild PKU  Classical PKU

Has this case been reported in the literature? If yes, please cite the reference:

## BIRTH INFORMATION

Pregnancy:  full term (38-40 wg)  premature at \_\_\_\_\_ weeks

Birth weight: \_\_\_\_\_ Birth height: \_\_\_\_\_ Head circumference: \_\_\_\_\_

## HYPERPHENYLALANINEMIA

not screened at neonatal period  
 screened at age: \_\_\_\_\_ blood Phe: \_\_\_\_\_ (mg/dl or  $\mu\text{mol/l}$ ) Mode of feeding:  breast  
 controlled at age: \_\_\_\_\_ blood Phe: \_\_\_\_\_ (mg/dl or  $\mu\text{mol/l}$ )  formula  
 at current age: \_\_\_\_\_ blood Phe: \_\_\_\_\_ (mg/dl or  $\mu\text{mol/l}$ )

Phenylalanine load with: \_\_\_\_\_ (mg/kg or mg/d), at age :  
Results:  
Time/date:  
Phe :  
Tyr :

PKU diet: tolerance: \_\_\_\_\_ (mg/d o. mg/kg/d), at age:

**TETRAHYDROBIOPTERIN TEST**

screened for BH<sub>4</sub> deficiency at age:

**Tetrahydrobiopterin loading test**

BH<sub>4</sub> loading test with mg/kg  oral  i.v.

Hours:	0	2	4	6	8	12	24	
<b>Phe (P)</b>								
<b>Tyr (P)</b>								
<b>Biopterin (P)</b>								

**Combined loading test**

combined phenylalanine mg/kg + BH<sub>4</sub> mg/kg loading test

Hours:	-3	0	2	4	6	8	12	24	
<b>Phe (P)</b>									
<b>Tyr (P)</b>									
<b>Biopterin (P)</b>									

**DNA ANALYSIS**

1. allele: AA aberration:  
 Nucleotide aberration:  
 Exon/Intron: Comments:
2. allele: AA aberration:  
 Nucleotide aberration:  
 Exon/Intron: Comments:

**TREATMENT**

Please give values in mg/kg/d and doses/day

Enter Age:	Age:	Age:	Age:	Age:	Age:
<b>Medication (mg/kg/d)</b>					
<b>Tetrahydrobiopterin (BH<sub>4</sub>)</b>					
<b>Folinic acid</b>					
<b>Restricted diet (Phe)</b>					
<b>Others</b>					
<b>Clinical examinations</b>					
<b>Comments</b>					
<b>Weight</b>					
<b>Length</b>					
<b>Head circumference</b>					
<b>IQ / DQ</b>					
<b>Phenylalanine in blood</b>					

The patient is no longer in our care. Please contact:

The questionnaire has been completed by (name, address, Tel/fax/email):

Anonymous patient's data will be included in the BIOPKU database, which is by definition public.

Date:

Signature: