

International Database of Tetrahydrobiopterin Deficiencies



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URL: <http://www.bh4.org>

ID #:

For internal use only

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):

Number of affected siblings:

First name: Age:
Died: yes no Age:

First name: Age:
Died: yes no Age:

Type of BH4 deficiency:

GTPCH PTPS SR PCD DHPR _____

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: _____
Clinical status: normal abnormal (poor sucking, irritability, etc.)
describe: _____

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 AND NEUROTRANSMITTERS DEFICIENCY

- screened for BH₄ deficiency at age:
 initial diagnosis at age: because case already known in the family
 atypical neurological signs

diagnosed by (laboratory):

Pterins in urine (units:):

Date	Neopterin	Biopterin	%Biopterin	%BH ₄	Pterin	Primapterin	Others
Controls:							

Pterins and folates in serum (units:):

Date	Neopterin	Biopterin	%Biopterin	%BH ₄	Pterin	Folate (rbc)
Controls:						

Pterins in CSF (units:):

Date	Neopterin	Biopterin	%Biopterin	%BH ₄	Pterin	Others
Controls:						

Neurotransmitters and folates in CSF (units:):

Date	5HIAA	HVA	MHPG	E	NE	5MTHF
Controls:						

Tetrahydrobiopterin loading test

- BH₄ loading test with mg/kg oral i.v.
 combined phenylalanine mg/kg + BH₄ mg/kg loading test

Hours:	-3	0	2	4	6	8
Phe (P)						
Tyr (P)						
Neo (U)						
Bio (U)						

Enzymes

	Phenylalanine hydroxylase	DHPR	PTPS	GTPCH
Material (Guthrie card, rbc, liver, fibroblasts, etc.)				
Units				
Patient				
Mother				
Father				
Controls				

CLINICAL SYMPTOMS

Onset of symptoms at age:

Was the patient already on a phenylalanine restricted diet: yes no

Signs	Age at which symptoms			
	appeared		disappeared	
	before treatment	on treatment	spontaneously	on treatment
Convulsions				
Myoclonic seizures				
Hypotonia				
Hypertonia				
Flexor spasms				
Dystonic posture				
Areflexia				
Athetosis				
Ataxia				
Hyperkinesia				
Extrapyramidal signs				
Irritability				
Ocular deviation				
Hyperreflexia				
Hypersalivation				
Excessive drooling				
Drowsiness				
Hyperventilation				
Swallowing difficulties				
Failure to thrive				
Retardation				
Microcephaly				
others				

EEG / CT / MRI

Age and findings:

DNA analysis

1. allele: AA aberration:

Nucleotide aberration:

Exon:

2. allele: AA aberration:

Nucleotide aberration:

Exon:

FOLLOW-UP

The patient is alive, present age:

died at age:

Cause of death:

TREATMENT AND CSF STATUS

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

	Enter Age: (mg/kg/d)	Age:	Age:	Age:	Age:	Age:
Medication						
L-Dopa						
Carbidopa						
5-OH-Tryptophan						
Tetrahydrobiopterin (BH ₄)						
Folinic acid						
Deprenyl (Selegilin)						
Anticonvulsants						
Restricted diet						
Others						
Clinical examinations						
Comments						
Improvement						
Weight						
Length						
Head circumference						
IQ / DQ						
Convulsions						
Myoclonic seizures						
Hypotonia						
Hypertonia						
Retardation						
Dystonic posture						
Areflexia						
Athetosis						
Ataxia						
Hyperkinesia						
Extrapyramidal signs						
Irritability						
Ocular deviation						
Hyperreflexia						
Hyperventilation						
Swallowing difficulties						
CSF status						
Neopterin						
Biopterin						
5HIAA						
HVA						
MHPG						
3-O-Methyl-Dopa						
5MTHF						
Phe						
Phenylalanine in blood						

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 BIODDEF database, which is by definition public.

Date:

Signature: