

**Division of Clinical Chemistry and Biochemistry**

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**Differential diagnosis of BH<sub>4</sub> deficiencies and inherited neurometabolic disorders**

Patient's data (Name, Date of birth, Address)

Sender (Address/Stamp)

Date in: Internal no.:

Sample infectious

Name of the physician: \_\_\_\_\_

Report by phone:

Please bill:

Sender  Insurance  Patient  research collaboration\*

Send a copy to:

Without specification the sender will be billed

Diagnosis / Comments

Neurologically abnormal  yes  no if yes:

Medications (please specify):

Body weight (kg):

BH<sub>4</sub>-dosage (mg):

Side effects during the test:

- Seizures (therapy resistant)
- Ataxia
- Dys- and hyperkinesias
- Hypotonia / hypertonia
- Dystonia
- Hyperreflexia
- Hypersalivation
- Opisthotonus
- Retardation (psychomotor)
- Behavioral abnormalities
- MRI / CT:
- 

URINE	PLASMA	CSF	GUTHRIE CARD															
2-5ml (random sample)	ca. 1ml (Heparin or EDTA)	ca. 1ml (no additives)	min. 3-4 blood spots per test															
Sample date:	Sample date:	Sample date:	Sample date:															
<input type="checkbox"/> Pterins <input type="checkbox"/> Vanillinlactic acid <input type="checkbox"/> <input type="checkbox"/> before BH <sub>4</sub> <input type="checkbox"/> 4-8 h after BH <sub>4</sub>	<input type="checkbox"/> Pterins <input type="checkbox"/> Amino acids <input type="checkbox"/> AADC activity <input type="checkbox"/> Folates <table border="1"> <thead> <tr> <th>µmol/L**</th> <th>Phe</th> <th>Tyr</th> </tr> </thead> <tbody> <tr> <td><input type="checkbox"/> 0 h before BH<sub>4</sub></td> <td></td> <td></td> </tr> <tr> <td><input type="checkbox"/> 4 h after BH<sub>4</sub></td> <td></td> <td></td> </tr> <tr> <td><input type="checkbox"/> 8 h after BH<sub>4</sub></td> <td></td> <td></td> </tr> <tr> <td><input type="checkbox"/> 24 h after BH<sub>4</sub></td> <td></td> <td></td> </tr> </tbody> </table> <p>** provide if analyzed in another laboratory</p>	µmol/L**	Phe	Tyr	<input type="checkbox"/> 0 h before BH <sub>4</sub>			<input type="checkbox"/> 4 h after BH <sub>4</sub>			<input type="checkbox"/> 8 h after BH <sub>4</sub>			<input type="checkbox"/> 24 h after BH <sub>4</sub>			<input type="checkbox"/> Pterins <input type="checkbox"/> Amino acids <input type="checkbox"/> Neurotransmitters <input type="checkbox"/> Folates Lumbar puncture (Time): <input type="checkbox"/> morning <input type="checkbox"/> Fraction: <input type="checkbox"/> 0-1ml <input type="checkbox"/> 1-2ml <input type="checkbox"/> >2ml Constitution: <input type="checkbox"/> blood <input type="checkbox"/> centrifuged <input type="checkbox"/> xanthochrom	<input type="checkbox"/> DHPR activity <input type="checkbox"/> Pterins <input type="checkbox"/> Amino acids <input type="checkbox"/> before BH <sub>4</sub> <input type="checkbox"/> 4 h after BH <sub>4</sub> <input type="checkbox"/> 8 h after BH <sub>4</sub> <input type="checkbox"/> 24 h after BH <sub>4</sub> <input type="checkbox"/> 32 h after BH <sub>4</sub> <input type="checkbox"/> 48 h after BH <sub>4</sub>
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**General Information**

**Pterins in urine:** Protect from light. Store at -20°C or oxidize with manganese dioxide (MnO<sub>2</sub>). One random portion only (no 24h collection).

**Pterins in plasma:** Protect from light. Store at -80°C. For BH<sub>4</sub> analysis use special sample tubes (EDTA + DTE).

**Pterins in CSF:** Protect from light. Store at -80°C.

**Neurotransmitters and folates in CSF:** Protect from light. Store at -80°C. Discard first 0,5 ml. Send the second fraction (0,5-1.5 ml). No additives.

**Amino acids in plasma and CSF:** Store at -20°C.

**AADC in plasma:** Store at -80°C.

**SEND WITH SUFFICIENT DRY ICE** (oxidized urines can be transported at room temperature)

For more information please contact the BH<sub>4</sub> Home Page [www.bh4.org](http://www.bh4.org)

Laboratory for Selective Screening, University Children's Hospital, Switzerland (Accredited according to ISO/IEC 17025)  
"Tetrahydrobiopterin Research Group"

\*You are not allowed to publish any results done on research basis without prior permission of the laboratory in Zürich.