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Date in: \_\_\_\_\_ Internal no. \_\_\_\_\_  
 Sample: Day 16 8 4 2 1 Mt. 8 4 2 1  
 Patient's data (Family name, First name, Date of birth, Address)

**DIFFERENTIAL DIAGNOSIS OF BH<sub>4</sub> DEFICIENCIES AND INHERITED NEUROMETABOLIC DISORDERS**

Sender (Address/Stamp): \_\_\_\_\_

Please bill:  Sender  Insurance  Patient  research collaboration\*  
 Without specification the sender will be billed.

Sample:  infectious

Report by phone: \_\_\_\_\_  
 Send a copy to: \_\_\_\_\_

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															
2-5 ml (random sample)	ca. 1 ml (Heparin or EDTA)	ca. 1 ml (no additives)	min. 3-4 blood spots															
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 <table border="1"> <thead> <tr> <th>mmol/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> **provide if analyzed in another laboratory	mmol/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"/> 0-1 ml <input type="checkbox"/> 1-2 ml <input type="checkbox"/> >2 ml Constitution: <input type="checkbox"/> blood if yes <input type="checkbox"/> centrifuged? <input type="checkbox"/> xanthochrom	<input type="checkbox"/> DHPR activity <input type="checkbox"/> Pterins <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>
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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 -70°C. For BH<sub>4</sub> analysis use special sample tubes (EDTA+DTE).  
**Pterins in Guthrie card:** 3—4 blood spots. Store in dark.  
**Pterins in CSF:** Protect from light. Store at -70°C.  
**Neurotransmitters und folates in CSF:** Protect from light. Store at -70°C. Send the second fraction (0.5-1.5 ml). Discard first 0.5 ml. No additives.  
**Amino acids in plasma or CSF:** Protect from light. Store at -20°C.  
**AADC in plasma:** Store at -70°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/EC 17025)  
 »Tetrahydrobiopterin Research Group«

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