



**Physician:**

Dr. M. Jaksch  
Freiburg Medical Lab

**Laboratory Report**  
**Special Tests**

Report Date: 13.03.2014

**Patient Name: NBS abnormal Hb**

Gender: Female  
Date of Birth: 04.03.2014  
Nationality:  
Your ID:

Test Request Code: 941  
Sample ID:  
Patient IDNo: 379712

Sampling Date / Time: 13.03.2014 / 15:52  
Entry Date / Time: 13.03.2014 / 15:52

**Remarks:**

**Insurance:**

Analysis	Result	Flag	Units	Reference Range
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**Exclusion of Metabolic Diseases (Filtercard)**

**Newborn screening \*** see text

No sign of the following disorders:  
Amino Acid related disorders such as: Phenylketonuria (PKU),  
Hyperphenylalaninaemia, Maple syrup disease etc.  
Fatty acid oxidation disorders and other disorders recognized in the  
acylcarnitine spectrum: e.g. MCAD, LCAD, VLCAD  
Other screened disorders: e.g. Hypothyreosis, AGS, Biotinidase deficiency,  
Galactosaemia, G6PDH Deficiency, Cystic Fibrosis, Hemoglobinopathies  
(HbS, HbC, HbD, HbE, HbO, HbLepore, HbG, beta-Thalassaemia)

Methods: MS-MS, FIA, Photometry, Capillary Electrophoresis

<b>Amino Acids (PKU etc.) *</b>	<b>normal</b>		quantitative	normal
<b>Acylcarnitines (Fatty acids) *</b>	<b>normal</b>		quantitative	normal
<b>TSH (Hypothyreodism) *</b>	<b>normal</b>		quantitative	normal
<b>17a-OH-Progesterone (AGS) *</b>	<b>normal</b>		quantitative	normal
<b>Galactose (Galactosemia) *</b>	<b>normal</b>		quantitative	normal
<b>Biotinidase (Biot.deficiency)*</b>	<b>normal</b>		quantitative	normal
<b>Glucose-6-PDH *</b>	<b>normal</b>		quantitative	normal
<b>Immunoreactive Trypsin *</b>	<b>normal</b>		quantitative	normal
<b>Hemoglobin Screening *</b>	<b>HbH/HbBarts</b>		quantitative	normal

The Hb electrophoresis detected an abnormal hemoglobin in the HbH/Hb Barts-range (<1.0%).  
This finding of a hemoglobin-Barts, a tetramer of 4 gamma-chains, indirectly indicates an  
alpha-thalassaemia (-a/aa). Clinical symptoms like microcytic hypochrome anemia can progress over years.  
Usually, there is no indication for therapy. For a further differentiation, we recommend an additional molecular  
genetic DNA analysis.

Note:  
Our reference values are adjusted to age and gender.  
Daily internal Quality Control within the required range  
(according to ISO 15189).  
External Quality Control available on request.  
^ non-accredited parameter  
\* This investigation was performed in a collaborating  
accredited laboratory (Germany).

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