



RUHR-UNIVERSITY BOCHUM

Human Genetics

Prof. Dr. med. Jörg Thomas Epplen

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With my signature overleaf I agree that the blood (tissue) that was taken from me / my child / the person under my guardianship

.....

(name and date of birth in block letters)

shall be analysed for genetic changes in order to clarify the respective disease(s) / genetic risk(s) as specified overleaf. In addition, I declare that I was informed on the possibilities and the limits of the requested analyses and about the consequences, which result from the diagnostic findings. I was informed about the possibility of genetic counselling.

I agree that the specimen may be stored to verify the findings and for eventual additional analyses (scientific as well) without temporary limitation.

My data and the diagnostic findings underlie the legal requirements in Germany concerning confidential medical communication. My data may be referred to other persons (family members or further attending physicians) only with my explicit agreement and on my demand.

This informed consent can be revoked any time. Individual paragraphs of this consent form may be deleted as intended.

This information is also available online (www.rub.de/mhg)

Molecular Genetic Diagnostics - Order form

Patient:

Name:	Send to: Ruhr-Universität Bochum Humangenetik z.H. Frau G. Rodepeter D-44780 BOCHUM Germany Fax: +49-234-32-14196	Sender:
Born:		
Addr.:		
.....		

Sample: DNA (preferred): volume ____ µl, concentration: ____ ng/µl
 Blood, taken on: ____-____-____ (sample requirements: **EDTA blood** (lavender tube, 5-10 ml for adults / 2 ml for neonates); send via express mail on Mondays or Tuesdays!)

Patient information: clinical diagnosis:

Gender: female male family history:

Symptoms:

Genetic test(s) to be performed:

- | | | |
|--|---|--|
| <input type="checkbox"/> Huntington's disease[#]
<input type="checkbox"/> SCA [1] [2] [3] [6] [7] [17]
<input type="checkbox"/> SCA13 (<i>KCNC3</i>)
<input type="checkbox"/> Hallervorden-Spatz syndrome
<input type="checkbox"/> SCA14[#] (<i>PRKCG</i>)
<input type="checkbox"/> NBIA 1 (<i>PANK2</i>)
<input type="checkbox"/> NBIA 2 (<i>PLA2G6</i>)

<input type="checkbox"/> malignant hyperthermia[#] (<i>MH1</i>)
<input type="checkbox"/> sarcoglycanopathies[#] (<i>SCG</i>)** [α] [β] [γ] [δ] [ε]
<input type="checkbox"/> glucocorticoid-remediable aldosteronism[#] (<i>GSH</i>)
<input type="checkbox"/> Canavan disease[#] (<i>ASPA</i>)
<input type="checkbox"/> congenital merosin-deficient muscular dystrophy[#] (<i>LAMA2</i>)
<input type="checkbox"/> LGMD2J[#] (<i>TTN</i> exons 312 & 313)
<input type="checkbox"/> Sandhoff disease[#] (<i>HEXB</i>)
<input type="checkbox"/> Nemaline Myopathy 1 (<i>TPM2, TPM3</i>)

<input type="checkbox"/> HMSN type I + III[#]
<input type="checkbox"/> HMSN type II[#] [X] [1B] [2A2]**
<input type="checkbox"/> HNPP[#]
<input type="checkbox"/> HMSN[#] [1A] [1B] [1C] [1D] [1E] [2A2] [2I/J] [4E] [X]**
<input type="checkbox"/> deafness/amblyocousia[#] (<i>GJB2, GJB6</i>)

<input type="checkbox"/> HNPCC (Hered. Non-Polyposis Colon-Carcinoma) ^{#,*} (<i>MLH1, MSH2, MSH6</i>)
<input type="checkbox"/> microsatellite instability[#] (from carcinoma tissue)
<input type="checkbox"/> Immunohistochemistry (in cooperation with Prof. Tannapfel, Pathology; from carcinoma tissue)

<input type="checkbox"/> spastic paraplegia (HSP/SPG)**
<input type="checkbox"/> autosomal dominant: type [3A] [#] [4] [#] [31] ([10] [13]) [*]
<input type="checkbox"/> autosomal recessive ^{**} : type [7] [11] [#]

<input type="checkbox"/> ataxia with oculo-mot. apraxia[#] (AOA)** [1] [2]
<input type="checkbox"/> juvenile amyotrophic lateral sclerosis[#] (ALS4)
<input type="checkbox"/> neurofibromatosis[#] type [1] [2] | <input type="checkbox"/> Friedreich's ataxia[#] (FA)
<input type="checkbox"/> DRPLA[#] (<i>Atrophin1</i>)
<input type="checkbox"/> SCA27[#] (<i>FGF14</i>)
<input type="checkbox"/> HSAN[#] type [1] [4/CIPA]

<input type="checkbox"/> central core disease[#] (CC)
<input type="checkbox"/> fragile-X syndrome / FXTAS[#] (FRAX-A) ^{***}
<input type="checkbox"/> hypoadosteronism[#] (<i>CYP11B2</i>)
<input type="checkbox"/> immunosseous dysplasia, Schimke type[#] (<i>SMARCAL1</i>)
<input type="checkbox"/> caveolin 3 related myopathy[#] (<i>CAV3</i>)
<input type="checkbox"/> myofibrillar myopathy[#] (MYOT)
<input type="checkbox"/> Tay-Sachs disease[#] (GM2-AB variant, <i>HEXA</i>)
<input type="checkbox"/> IBMPFD myopathy (VCP)

<input type="checkbox"/> Alexander disease[#] (<i>GFAP</i>)
<input type="checkbox"/> Rett syndrome[#] (<i>MeCP2</i>)

<input type="checkbox"/> deafness (mitochondrial) | <p>Phone: +49-234-32 + extension</p> <p>Dr. med. S. Wieczorek +23831
stefan.wieczorek@rub.de</p> <p>Dr. rer.nat. G. Dekomien +25764
gabriele.dekomien@rub.de</p> <p>Dr. W. M. Gerding +23831
wanda.gerding@rub.de</p> <p>Dr. rer. nat. M. Schlinghoff +25764
micaela.schlinghoff@rub.de</p> <p>Dr. med. S. Stemmler +25762
susanne.stemmler@rub.de</p> <p>Dr. rer.nat. L. Arning +23831
larissa.arning@rub.de</p> |
|--|---|--|

Other :

[#] accredited analysis according to DIN EN ISO 15189:2007
^{*} please enquire ^{**} please specify requested subtype ^{***} ≥ 10ml EDTA blood



Informed consent: I have read and acknowledged the explanations on the front side and agree with the analyses. Sections I do not agree with are crossed out.

.....
 Location Date Signature

Invoice to transferring physician patient

.....
 Date Signature of physician Name in block letters Email address