



PATIENT/IN (use sticker if available)

Last name: _____

First name(s): _____

Date of birth: _____ gender: m f

Address: _____

Ethnic background: _____
(may be important for recessive conditions)

urgent

Pregnancy? yes no

Pregnent person: _____

Week of gestation: _____

Please ship samples to:

Institute of Human Genetics
University Hospital of Cologne
Kerpener Str. 34
50931 Cologne, Germany

Phone +49-221-478-86811
Fax +49-221-478-86812

Billing

Test will be paid by Referring facility Patient

Please note that international requests must be accompanied by a confirmation of payment. Please contact us for details.

Informed consent form

(according to German Genetic Diagnostics Act)

The planned genetic test, its limitations and possible interpretations of results have been explained to me in detail by the physician stated below. I have had the opportunity to discuss the details and ask questions about this information. By signing this form, I consent that genetic testing will be performed for the following disease/condition/diagnosis (to be entered by physician):

I consent that the required sample (e.g. blood, tissue, amniotic fluid) will be taken.

The sample and the results of the testing may be used as follows:

I consent that **remaining sample material** will be **stored** for verification of results and quality management purposes.

I consent that **remaining sample material** will be **stored** to be available for new diagnostic options in the future.

I consent that the **test results and records** will not be destroyed after 10 years — as laid down in German statutory provisions — but will be **stored**.

I consent that the request for testing and all personal details required for the testing are **forwarded** to a specialized cooperating laboratory if necessary .

I wish to be informed on **incidental findings** that may be discovered by the genetic testing, even if they do not directly relate to the above mentioned disease/condition/diagnosis in question.

I consent that **remaining sample material** will be used for **research on the causes** and improved treatment of genetic diseases.

I consent that **data/results** that may be generated on the condition in question will be used in de-identified (pseudonymized) form for **academic or medical teaching** and for **scientific purposes** and will be published in anonymized form in **scientific journals**.

I consent that the results of the testing may be sent to the following physicians:

- Please delete as appropriate -

I am free to withdraw any of the above statements at any time. I have had enough time to consider my decision.

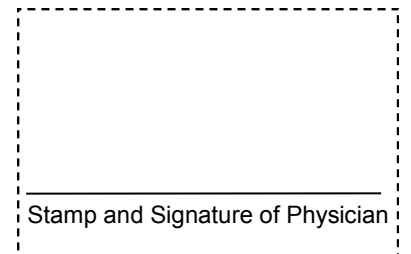
I obtained an informed consent form signed by the patient.

Place, Date

Signature Patient/
Parent/Legal Guardian

Name of Physician in
printed characters

Stamp and Signature of Physician



Request for molecular genetic testing — Neuroendocrine tumors

Clinical Findings / Diagnosis / Indication / Reason for Testing

Has any previous genetic test been performed in the patient relating to the above reason for testing?

- no
- yes, please specify previous findings / genetic tests:

Is there an Index patient known?

- no
- yes (please specify mutation, disorder and relationship)
- yes; but there are no or incomplete information on the index patient. In this case a specific reason has to be given for the genetic mutation analysis on your patient. The statement **must** include the probability of predisposition of your patient or the remaining lifetime risk of developing the disease.

Type of testing

- diagnostic testing
- prenatal testing
- diagnostic testing - segregation analysis in the parents/further family members if the test result of the child/index patient requires clarification
- predictive testing
- heterozygosity/carrier testing

Type of sample

- blood
- saliva
- amniotic fluid
- DNA
- fibroblasts
- other:
- buccal swab
- chorion villi

Date of sample collection

Sample was collected on:

Family history / Pedigree

- significant family history (see above information on index patient)
- mother clearly affected
- father clearly affected
- family history not available
- no significant family history
- both parents clearly not affected
- mother clearly not affected,
no information on father available
- father clearly not affected,
no information on mother available

Parental consanguinity

- no
- yes (please specify):

Pedigree (optional)

Request for molecular genetic testing — Neuroendocrine tumors

You can choose between standard panels, complete panels or single gene diagnostics. Standard panels combine the genes most frequently mutated in patients with the respective phenotype and contain up to 25 kb of coding sequence (only relevant for patients with the E112 [S2] health insurance document). Complete panels cover all genes associated with the phenotype. If mutations of a single gene are known to be causative for a large proportion of the phenotype, the gene may be analyzed by conventional Sanger sequencing prior to an NGS-analysis.

Contact person for clinical questions: Dr. med. F. Marbach (+49 221-478-89298, felix.marbach@uk-koeln.de) or Prof. Dr. med. C. Netzer (+49 221-478-89586, christian.netzer@uk-koeln.de). For all other questions related to genetic diagnostics, sample material and billing, please contact: +49 221-478-86811 and +49 221-478-86193 or mvz-humangenetik@uk-koeln.de.

Multi-gene panel diagnostics:

„**Core-Gene**“ of panels are printed in **bold**; the coding sequence of these genes has to be analysed to 100% in the highest quality.

*: for these genes a quantitative analysis by MLPA is also available.

Neuroendocrine tumors

- Multiple endocrine neoplasia type I, II, IV and hereditary paraganglioma/pheochromocytoma according to NCBI GeneReviews and Endocrine Society Clinical**

*CDKN1B, KIF1B, MAX, **MEN1***, **RET**, SDHA, SDHAF2*, **SDHB***, SDHC*, **SDHD***, TMEM127, **VHL**** (10 genes, 11.23 kb)

- Single gene diagnostic**

<input type="checkbox"/> <i>CDKN1B</i>	<input type="checkbox"/> <i>RET</i>	<input type="checkbox"/> <i>SDHB*</i>	<input type="checkbox"/> <i>TMEM127</i>
<input type="checkbox"/> <i>MAX</i>	<input type="checkbox"/> <i>SDHA</i>	<input type="checkbox"/> <i>SDHC*</i>	<input type="checkbox"/> <i>VHL*</i>
<input type="checkbox"/> <i>MEN1*</i>	<input type="checkbox"/> <i>SDHAF2*</i>	<input type="checkbox"/> <i>SDHD*</i>	

Request for molecular genetic testing — Neuroendocrine tumors

You can choose between the complete panel *Neuroendocrine tumors* or single gene diagnostic.

Contact person for clinical questions: Dr. med. F. Marbach (+49 221-478-89298, felix.marbach@uk-koeln.de) or Prof. Dr. med. C. Netzer (+49 221-478-89586, christian.netzer@uk-koeln.de). All other questions related to genetic diagnostics, sample material and billing (+49 221-478-86811 and +49 221-478-86193 or mvz-humangenetik@uk-koeln.de).

Multi-gene panel diagnostics:

*: for these genes a quantitative analysis by MLPA is also available.

Complete panel *multiple Endocrine Neoplasia and Paraganglioma / Pheochromocytoma syndrome*

Single gene diagnostic by Sanger-sequencing technology

Genes	kb	Single gene diagnostic
CDKN1B	0.60	<input type="checkbox"/>
KIF1B	5.31	not available
MAX	0.48	<input type="checkbox"/>
MEN1*	1.83	<input type="checkbox"/>
RET	3.35	<input type="checkbox"/>
SDHA	2.00	<input type="checkbox"/>
SDHAF2*	0.50	<input type="checkbox"/>
SDHB*	0.84	<input type="checkbox"/>
SDHC*	0.51	<input type="checkbox"/>
SDHD*	0.48	<input type="checkbox"/>
TMEM127	0.71	<input type="checkbox"/>
VHL*	0.64	<input type="checkbox"/>