

Order form for hearing loss panel sequencing



Patient details (use label):

Male Female

Name _____

Surname _____ Date of birth _____

Cost coverage declaration (mandatory to fill in):

A signed agreement for payment is enclosed

Please send us a cost quote for an:

- Invoice to patient (give details and address on reverse page)
- Invoice to referring institution
- Invoice by special agreement with Zentrum Medizinische Genetik Würzburg (to be obtained prior to testing)

The invoicing process will be handled on our behalf by Ärztliche Verrechnungsstelle Büdigen e.V.

Relevant clinical patient and/or family data:

Information to anamnesis and other remarks: (please enclose report, if available)

Information about the hearing loss panel:

"Next generation sequencing" (NGS) technology allows the parallel analysis of numerous genes in a single approach permitting panel diagnostics.

We have selected **39 genes** which are known to be responsible for common hereditary forms of hearing loss.

Unless otherwise ordered, we will analyze all genes on the panel. However, if you would like the analysis to be restricted to a sub-group of 'core genes', please tick the boxes to specify the subpanel.

The analysis will be performed by Illumina TruSeq Rapid Exome enrichment and NextSeq 500 technologies. Together with the report, you will receive an overview of the analyzed genes and the achieved coverage.

The NGS technology covers only mutations which are detectable by DNA sequencing. Other types of mutations residing in complex genomic regions (e.g. gene-pseudogene homologous regions), large deletion and duplications, as well as methylation deficiencies cannot be analyzed.

For further information, please contact us at the telephone numbers below.

MATERIAL: 5-10 ml EDTA blood. Please label tubes clearly, package in protective packaging, and ship at ambient temperature as soon as possible within one week

Contacts:

Dr. Gerhard Meng, Tel: +49-931-31-84064 (pricing information)
Dr. Jörg Schröder, Tel: +49-931-31-81455 (clinical and general questions)
Dr. Barbara Vona, Tel: +49-931-31-84244 (technical questions)
Michaela Hofrichter, M. Sc., Tel: +49-931-31-81573 (technical questions)
Paulina L. Bahena C., Tel: +49-931-31-83397 (clinical and technical questions)

Subpanels (please tick boxes)

> Please note our order form for single gene analysis (e.g. *GJB2*, *STRC*, *SLC26A4*, etc.) on our homepage:
<http://www.humgen.biozentrum.uni-wuerzburg.de/patientenversorgung/formulare/>

Non-syndromic dominant hearing loss

TECTA, *MYO7A*, *TMC1*, *WFS1*, *MYO6*, *KCNQ4*, *ACTG1*

Non-syndromic recessive hearing loss

SLC26A4, *OTOF*, *MYO15A*, *TMPRSS3*, *POU3F4*, *USH1G*, *USH1C*
(*STRC* excluded in advance)

Non-syndromic middle-frequency hearing loss

CCDC50, *CEACAM16*, *COL11A2*, *EYA4*, *TECTA*, *SLC44A4*

Auditory neuropathy

AIFM1, *ATP1A3*, *DFNB59*, *DIAPH3*, *OTOF*

Syndromic hearing loss

- Usher Syndrome Type 1 (*MYO7A*, *CDH23*, *PCDH15*, *USH1C*)
- Usher Syndrome Type 2 (*USH2A*, *DFNB31*)
- Pendred Syndrome (*SLC26A4*, *FOXI1*, *KCNJ10*)
- Waardenburg Syndrome (*MITF*, *PAX3*, *SNAI2*, *EDN3*, *SOX10*, *KIT*,
- Branchio-oto-renal Syndrome (*EYA1*, *SIX5*, *SIX1*)

Gene combinations:

Dominant/recessive Genes:

TECTA (6.5 kb) *TMC1* (2.3 kb) *MYO6* (3.9 kb)

Recessive Genes

OTOF (6 kb) *MYO15A* (10.6 kb) *TMPRSS3* (1.4 kb)

STRC (5.3 kb)

Dominant Genes

KCNQ4 (2.1 kb) *MYH14* (6.1 kb) *COCH* (1.7 kb)

ACTG1 (1.1 kb)

X-chromosomal Gene

POU3F4 (1.1 kb)

Syndromic/non-syndromic Genes

MYO7A (6.6 kb) *SLC26A4* (2.3 kb) *WFS1* (2.7 kb)

CDH23 (10 kb) *PCDH15* (5.9 kb) *COL11A2* (5.2 kb)

USH1G (1.1 kb) *USH1C* (2.7 kb)

Syndromic Genes

CHD7 (9 kb) *EYA1* (1.8 kb) *COL4A5* (5.1 kb)

FOXI1 (0.9 kb) *MITF* (1.6 kb) *PAX3* (1.5 kb)

USH2A (15.6 kb) *GPR98* (18.9 kb)

Samples taken (date): _____ by: _____

Physician name (please print) _____

Date _____ Physician's signature _____

(Physician's stamp)

According to the German gene testing act written patient's consent is required for every genetic test (see page 2)

Consent for genetic testing using panel diagnostics

Patient details (use label):

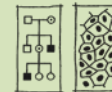
Surname _____ Given name _____
 male female
 Date of birth _____
 Street _____
 Postal code _____ City, Country _____

Please return to:

**Zentrum Med. Genetik Würzburg
 Biozentrum, Am Hubland
 97074 Würzburg**



**Praxis für Humangenetik
 PD Dr. med. Erdmute Kunstmann**
 Tel: 0931-3184435, Fax: 0931-45265859
 E-Mail: kunstmann@biozentrum.uni-wuerzburg.de



Institut für Humangenetik, DNA-Labor
 Tel: 0931-3184064, Fax: 0931-3184069
 E-Mail: gmeng@biozentrum.uni-wuerzburg.de



The German gene testing act (GenDG) requires written informed consent to be obtained from every patient prior to genetic testing.

Please read the following carefully and tick **each** box as appropriate.

I hereby consent to the analysis of my DNA or my child's DNA by panel diagnostics for clarification of a suspected **hereditary hearing loss**.

I have been informed about its genetic basis, options for prevention and treatment and about the scope and aims of the planned genetic test, its predictive value and its limits. I have been informed about the risk of the required blood / tissue sampling. All my questions have been answered to my satisfaction.

I consent to the reporting of genetic results to my primary care physician or the following physician:

Dr.:
 Address:
 City, Country:

yes
 no

The application of such testing can result in incidental findings, which are not associated with the above named disorder. I wish to be informed of any such incidental findings.

yes
 no

By German law, surplus genetic material (blood, DNA sample) must be destroyed after the completion of the genetic test. However, with my consent it may be stored and used for subsequent additional tests (if required) and/or as a control for subsequent testing of family members and relatives.

I consent to storage and subsequent use of my genetic material and/or the genetic material of my child for the above purposes.

yes
 no

Internal quality control is an important tool to guarantee the accuracy and reliability of genetic testing methods. For this purpose, genetic material from patients with rare genetic variants is an indispensable control material.

I consent to my DNA and/or the DNA of my child to be stored and used for internal quality control in the laboratory. Before such use, my sample and/or the sample of my child will be anonymised.

yes
 no

Genetic material from patients is also important for studying biological mechanisms which contribute to the development of hereditary disorders.

I consent to my DNA and/or the DNA of my child to be stored and used for research studies in the laboratory. I consent to be re-contacted before such use.

yes
 no

The German gene testing act requires genetic results to be stored for 10 years and then destroyed. With patient consent, they may be stored for longer. Often genetic data are required for counselling of children and relatives even after 10 years.

I consent to storage of my genetic results and/or the results of my child beyond the legal time-span and its use for my family only.

yes
 no

As required, the results may be used for the counselling / analysis of my relatives.

yes
 no

Genetic data will be deposited in a database at the Institute of Human Genetics at the University of Würzburg. Selected data will be anonymised and only used for the purpose of quality control and data comparison.

I have been informed that I can withdraw my consent at any time without giving reason and without incurring any penalty. I have further been informed that I have the right not to know about my genetic test results and to terminate the testing procedure at any time. I can request my genetic material and/or the genetic material of my child and my genetic results and reports and/or the genetic results and reports of my child to be destroyed before genetic counselling if I have changed my mind.

With my signature, I consent to the genetic test(s) indicated above and the sampling of blood or tissue for this purpose.

 City, date

 Signature of the patient or his/her legal representative