Consent for genetic testing using panel diagnostics

Patient details (use label):

Surname _____ Given name _____

Date of birth _____ ☐ male ☐ female

Street _____

Postcode _____ 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 putative **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.		☐ yes
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 disease. 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 use of my genetic material 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 being stored and used for internal quality control in the laboratory. Before such use, my sample will be anonymised.		☐ yes ☐ no
Genetic material from patients is also important for studying biological mechanisms which contribute to the development of hereditary diseases. I consent to my DNA being stored and used for research studies in the laboratory. Before such use, my sample will be anonymised.		☐ yes ☐ no
The German gene testing act requires genetic data 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 data 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. All data will 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 testing results and to terminate the testing procedure at any time. I can request my genetic material and my genetic data to be destroyed at any time 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

Order form for hearing loss panel sequencing

Patient details (use label):

☐ M ☐ F

Name

Surname Date of birth

Cost coverage declaration (mandatory to fill in):

☐ **A signed agreement to cost coverage 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 Büdingen e.V.

Relevant clinical patient and/or family data:

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

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

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 **35 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. pseudogene-containing regions), large deletion and duplications, as well as methylation deficiencies cannot be analyzed.

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

Contacts:

Dr. Gerhard Meng, Tel: +49-931-31-84064 (pricing information)
Dr. Jörg Schröder, Tel: +49-931-31-81455 (clinical 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*

☐ ***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*, *KITLG*)
- ☐ Branchio-oto-renal Syndrome (*EYA1*, *SIX5*, *SIX1*)

☐ ***Gene combinations:***

Dominante/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) ☐ *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 overleaf)