UNIKLINIK KÖLN | Institut für Humangenetik

Certified by: TÜV SÜD ISO 9001

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 informend consent form signed by the patient.

Place, Date	Signature Patient/ Parent/Legal Guardian	Name of Physician in printed characters	Stamp and Signature of Physician
_____	_____	_____	_____

►Please give a copy of the completed and signed consent form to the patient◄

Request for molecular genetic testing-
Marfan syndrome and other connective tissue disorders

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 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

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

☐ prenatal testing

☐ predictive testing

☐ heterozygosity/carrier testing

Type of sample

☐ blood

☐ DNA

☐ buccal swab

☐ saliva

☐ fibroblasts

☐ chorion villi

☐ amniotic fluid

☐ other:

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-
Marfan syndrome and other connective tissue disorders

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: 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.

Marfan syndrome, Loeys-Dietz syndrome, Ehlers-Danlos syndrome, genes associated with aortic dilatation

☐ **Standard panel (#010): Marfan syndrome and type 1 fibrillinopathies**

FBN1*; TGFBR1*, TGFBR2 (3 genes, 11.83 kb)

☐ **Standard panel (#142): Thoracic aortic dilatation and dissection, genes according to German EBM**

ACTA2**, C1R, CBS, COL1A1*, COL1A2*, **COL3A1, COL4A5*, COL5A1, COL5A2, EFEMP2, ELN, **FBN1***, FBN2, FKBP14, FLNA, GATA5, MAT2A, MFAP5, **MYH11**, **MYLK**, NOTCH1, PLOD1*, PRKG1, SKI, SLC2A10, **SMAD3**, SMAD4, **TGFB2**, TGFB3, **TGFBR1***, **TGFBR2***, ZNF469* (32 genes, 101.4 kb)

☐ **Complete panel (#143): Connective tissue disorders (Marfan, Ehlers-Danlos and Loeys-Dietz syndromes) according to NCBI *Genereviews*, *clinical utility gene cards***

ACTA2**, ADAMTS2, B3GALT6, B4GALT7, C1R,C1S, CBS, CHST14, COL1A1*, COL1A2*, **COL3A1, COL4A5*, COL5A1, COL5A2, COL12A1, DSE, EFEMP2, ELN, **FBN1***, FBN2, FKBP14, FLNA, GATA5, MAT2A, MFAP5, **MYH11**, **MYLK**, NOTCH1, PLOD1*, PRDM5, PRKG1, SKI, SLC2A10, SLC39A13, **SMAD3**, SMAD4, **TGFB2**, TGFB3, **TGFBR1***, **TGFBR2***, TNXB, ZNF469* (42 genes, 149.79 kb)

☐ **Complete panel (#144): Ehlers-Danlos syndrome, genes according to *clinical utility gene card*, EDS type I-IV**

ADAMTS2**, B3GALT6, B4GALT7, C1R, C1S, CBS, CHST14, COL1A1*, COL1A2*, **COL3A1, **COL5A1**, **COL5A2**, COL12A1, DSE, EFEMP2, ELN, **FKBP14**, FLNA, **PLOD1***, PRDM5, SLC2A10, SLC39A13, TNXB, ZNF469* (24 genes, 91.13 kb)

☐ **Standard panel (#014): Ehlers-Danlos syndrome, main genes according to NCBI *Genereviews*, 2017 International Classification of the EDS**

ADAMTS2**, COL1A2*, **COL3A1, **COL5A1**, **COL5A2**, **FKBP14**, **PLOD1*** (7 genes, 24.98 kb)

☐ **Standard panel (#015): Ehlers-Danlos syndrome, dominant**

COL1A1, COL1A2*, **COL3A1***, **COL5A1**, **COL5A2*** (5 genes, 22.9 kb)

☐ **Standard panel (#016): Ehlers-Danlos syndrome, recessive**

***ADAMTS2**, B3GALT6, B4GALT7, CBS, CHST14, COL1A2*, DSE, EFEMP2, **FKBP14**, **PLOD1**, SLC2A10, SLC39A13*
(12 genes, 22.2 kb)

☐ **Single gene diagnostics**

☐ *ACTA2*	☐ *COL3A1**	☐ *MYH11*	☐ *TGFB2*
☐ *CBS*	☐ *COL4A5**	☐ *MYLK*	☐ *TGFBR1**
☐ *COL1A1**	☐ *FBN1**	☐ *SMAD3*	☐ *TGFBR2**
☐ *COL1A2**			

Request for molecular genetic testing-
Marfan syndrome and other connective tissue disorders

You can choose between the complete panel Marfan syndrome and other connective tissue disordersor single gene diagnostics.

Contact person for clinical questions: 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:
*: for these genes a quantitative analysis by MLPA is also available.

☐ **Complete panel Marfan syndrome, Loeys-Dietz syndrome, Ehlers-Danlos syndrome and genes associated with aortic dilatation (#141)**

☐ **Single gene diagnostic by Sanger-sequencing technology**

Gene	kb	Single gene diagnostics
ACTA2	1.13	☐
ADAMTS2	3.64	not available
B3GALT6	0.99	not available
B4GALT7	0.98	not available
C1R	2.12	not available
C1S	2.07	not available
CBS	1.66	☐
CHST14	1.13	not available
COL1A1*	4.40	☐
COL1A2*	4.10	☐
COL12A1	9.19	not available
COL3A1*	4.40	☐
COL4A5*	5.06	☐
COL5A1	5.52	not available
COL5A2	4.50	not available
DSE	2.88	not available
EFEMP2	1.33	not available
ELN	2.36	not available
FBN1*	8.62	☐
FBN2	8.74	not available
FKBP14	0.64	not available
FLNA	7.92	not available
GATA5	1.19	not available
MAT2A	1.19	not available
MFAP5	0.52	not available
MYH11	5.92	☐
MYLK	5.75	☐
NOTCH1	7.67	not available
PLOD1*	2.18	not available
PRDM5	1.89	not available
PRKG1	2.06	not available
SKI	2.19	not available
SLC2A10	1.63	not available
SLC39A13	1.10	not available
SMAD3	1.28	☐
SMAD4	1.66	not available
TGFB2	1.25	☐
TGFB3	1.24	not available

Gene	kb	Single gene diagnostics
TGFBR1*	1.51	☐
TGFBR2*	1.70	☐
TNXB	12.74	not available
ZNF469	11.78	not available