

### Sample type

- Blood (EDTA)  
 DNA, Specify Source: \_\_\_\_\_  
 Others, Specify: \_\_\_\_\_

### Prenatal<sup>1)</sup>

- Native Amniotic Fluid (10-15 ml)  
 Native Chorionic Villi (20-40 mg)  
 Fetal DNA

<sup>1)</sup>Please add maternal EDTA blood or DNA for maternal cell contamination testing in case of prenatal analysis.

FOR LAB USE ONLY - DO NOT COVER

### Requesting Health Care Professional Information

Physician Name: \_\_\_\_\_ Institution: \_\_\_\_\_  
Street: \_\_\_\_\_ City: \_\_\_\_\_ Postal Code: \_\_\_\_\_  
Country: \_\_\_\_\_ Email: \_\_\_\_\_

### Patient Information

First Name: \_\_\_\_\_ Last Name: \_\_\_\_\_  
Date of Birth: Year \_\_\_\_ /Month \_\_\_\_ /Day \_\_\_\_ Your Reference Number: \_\_\_\_\_  
Sex:  Male  Female  Diverse  Unknown Ethnicity: \_\_\_\_\_

### Patient History

Unaffected  Affected  Age of Onset: \_\_\_\_\_  
Has the patient received hematopoietic stem cell transplantation?  Yes  No

Suspected Diagnosis: \_\_\_\_\_ ICD-10: \_\_\_\_\_

Relevant Clinical Findings (attach copies of clinical reports if available)

### Family History

Parental Consanguinity:  Yes  No  
Affected Siblings/Family Members:  Yes Relation to Patient: \_\_\_\_\_  No

Clinical Information of Affected Family Members (attach pedigree if available)

### Test Information

We offer a wide spectrum of molecular genetic analyses. In general, we will perform the most suitable analysis and gene composition testing, based on the patient's medical history you provide us, or alternatively, perform the customized analyses you request. Please contact us at info@medgen-mainz.de in case of any questions.

- Exome (Solo)  Exome (Trio) (Use separate request forms for each family member to be analysed)  
 Multi-Gene Panel corresponding to the suspected diagnosis  
 Customized Panel, Specify: \_\_\_\_\_

Single Gene Testing Gene Name: \_\_\_\_\_  Sequencing  Del/Dup Analysis (MLPA)  Repeat Analysis  
 Single Variant Testing Gene Name: \_\_\_\_\_ Variant (Mutation): \_\_\_\_\_  
Family Member Tested by Us:  No  Yes Our Patient ID: \_\_\_\_\_ Relation to Patient: \_\_\_\_\_

Regarding array CGH or cytogenetics, please find detailed information on our website.

Please Remember the Obligatory Declaration of Informed Consent (Back Page).

## Billing Information

Invoice to:  Patient  Institution

Institution/Last Name: \_\_\_\_\_ Department/First Name: \_\_\_\_\_

Street: \_\_\_\_\_ City: \_\_\_\_\_

Postal Code: \_\_\_\_\_ Country: \_\_\_\_\_

Phone: \_\_\_\_\_ Fax: \_\_\_\_\_

Email (mandatory): \_\_\_\_\_

VAT (mandatory for institutional customers in the EU): \_\_\_\_\_

The institution or the patient or the legal representative has been informed about the resulting costs and has approved them.

A quotation is required prior to starting the analysis.

### Declaration of Informed Consent

The German Genetic Diagnostics Act (GenDG) requires detailed information and written consent for all genetic analyses as well as additional genetic counselling prior to prenatal and predictive genetic analyses. Please read this form carefully and check any statements that may apply to you.

My attending physician \_\_\_\_\_ has informed me about the nature, importance and implications of the genetic testing to be performed. All my questions have been answered to my satisfaction and I have had enough time for consideration to make an informed decision. I wish to be informed about the results of the genetic testing and I have been informed of my right not to know. I agree to the requested test being subcontracted to a cooperating specialized medical laboratory, if necessary.

With my signature below I give my consent to genetic analyses according to the enclosed request or which are necessary to clarify the above mentioned disease/disorder/diagnosis in question and I agree with the blood/tissue collection required for this.

#### I give my additional consent to:

##### Reporting of secondary findings

I am aware that genetic evaluation focuses on those changes directly related to my clinical indication or suspected diagnosis. However, comprehensive analyses, e.g. whole exome sequencing (WES), examine numerous genes in parallel and can possibly identify genetic variants not related to my existing clinical symptoms, but which could lead to an increased risk of disease and knowledge of which might be of medical value for my personal health and treatment in the future (ACMG guideline: Kalia et al., Genetic Medicine; 2017). I understand the significance of such secondary findings for myself and my biological relatives and I agree to be informed about those changes from which practical consequences may be deduced.

Yes  No

##### Extended storage and use of my data

I understand that the collected data/test results are stored for 10 years according to legal requirements and must be destroyed afterwards. However, these data may still be of great importance to me or my relatives (e.g. my children) even after this period. Therefore, I give my consent to store my data beyond the legal period of 10 years.

Yes  No

I agree that the collected data/test results may be used in encrypted (pseudonymised) form for scientific and quality assurance purposes, may be published anonymously in scientific journals and transferred in the context of scientific projects.

Yes  No

##### Extended storage and use of my sample material

The Genetic Diagnostics Act requires that unused patient sample material be destroyed after the test is completed. With my consent, however, it may be stored. I authorize any remaining sample material to be transferred to the Medizinische Genetik Mainz, consent to its use for scientific and quality assurance purposes and to the transfer within the framework of scientific projects in encrypted (pseudonymised) form.

Yes  No

I agree that for a more precise assessment/classification of possibly causative changes detected in my child in the course of the genetic analysis performed, testing of my genetic material will be performed to confirm the kinship. The results of this testing will only be used for internal variant assessment, do not correspond to forensic analysis and will not be communicated as part of the results reporting.

Yes  No

I understand that I may withdraw this consent at any time, in whole or in part, without giving reasons and without risk of personal disadvantage. I understand that I have the right not to know the results of the genetic testing. I am aware that I can interrupt the analyses at any time until the results have been reported. I am aware that, once reported, the results are subjected to the legal retention period of 10 years and cannot be destroyed before expiration of this time even if requested.

\_\_\_\_\_  
Date

X

\_\_\_\_\_  
Signature of Patient or (Legal) Representative

I hereby confirm that the consent as shown above, including all mentioned sub-items, has been declared by the patient or their legal representative.

\_\_\_\_\_  
Date

X

\_\_\_\_\_  
Signature of Physician