

CARDIAC AND AORTIC PANELS KNOW&MANAGE ORDER FORM



BARCODE

PHYSICIAN INFORMATION / [ADD TRANSLATION IN LOCAL LANGUAGE]

Institution/Practice / [Add translation in local language]

First Name / [Add translation in local language]

Last Name / [Add translation in local language]

Address (street name, no., city, postal code, country) /
[Add translation in local language]

Telephone Number (country code & number) / [Add translation in local language]

E-mail Address (for report access) / [Add translation in local language]

PATIENT INFORMATION / [ADD TRANSLATION IN LOCAL LANGUAGE]

First Name / [Add translation in local language]

Last Name / [Add translation in local language]

Date of Birth (DD/MM/YYYY) /
[Add translation in local language]

Personal Identification No. /
[Add translation in local language]

Gender (male/female/other - specify karyotype) / [Add translation in local language]

Address (street name, no., city, postal code, country) /
[Add translation in local language]

Telephone Number (country code & number) / [Add translation in local language]

Reason for Test (diagnosis, predictive, carrier) /
[Add translation in local language]

Sample Collection Date (DD/MM/YYYY) / [Add translation in local language]

DECLARATION OF CONSENT (ACCORDING TO GERMAN GENETIC DIAGNOSTICS ACT, GenDG)

APPLICABLE only for the determination of genetic (hereditary) characteristics

The GenDG requires provision of detailed information and a written consent for all genetic investigations as well as genetic counselling prior to both predictive (applies to healthy individuals) and prenatal testing (with restrictions: prenatal testing is not performed for late manifesting disorders, including Hereditary Cancer Panels). The German Society of Human Genetics (GfH) and the Association of German Human Geneticists (BVDH) recommend clarifying the issues listed below during the information process. Please read the declaration of consent carefully and tick the boxes, in accordance with your consent.

By signing the form below I confirm that I:

- Have been fully informed by my physician about the significance and consequences of the genetic investigation, in compliance with GenDG.
- Have read/have been read the Informed Consent which is attached to this form and which I fully understand.
- Have been given sufficient opportunity to discuss open questions.
- Authorize [insert legal entity here] to collect the necessary samples for investigation (blood, tissue, chorionic villus cells or amniotic fluid for prenatal diagnosis) and to send this form to MVZ Martinsried GmbH, Lochhamer Str. 29, 82152 Martinsried, Germany, in order to perform the tests requested through this form.
- Consent to the genetic test being carried out in order to clarify the disease/dysfunction/suspected diagnosis below.

In addition,

- I agree that a copy of the results of the analysis may be sent to the following physician(s), in accordance with my express requests and according to [insert legal entity here] internal procedures. YES NO

Dr(s) Name

Street

Postcode/City

Country

Place

Signature of Parent
or Legal Guardian

- I agree that the investigation or parts of the investigation may be forwarded to collaborating medical laboratories, if necessary. YES NO
- I agree with the evaluation of additional genes in the same indication group as part of the research. YES NO
- I agree that the remaining specimens may be stored for further investigations after the examination is completed, yet not claiming storage. YES NO
- I agree that the specimens, and if applicable DNA sequence information, may be made available anonymously for quality management and scientific purposes. YES NO
- I agree that the results of the analysis may be stored for a longer period than the statutory period of 10 years, yet not claiming storage of results. YES NO
- I agree to the storage and use of my test results under the protection of anonymity in a statistical database used for scientific purposes and to help diagnose genetic diseases. I understand that I will remain under the protection of anonymity and I cannot be identified during the analysis of the data and that any personal information will be transformed into information of a non-personal nature. YES NO

By signing the form below I confirm that:

- I may stop the investigation at any time and ask for the results available until that time to be destroyed.
- I may withdraw any of my consents given through this form entirely or in part at any time without giving reasons.
- I will be charged for the costs incurred until the time of withdrawal of consent.
- I may choose not to be informed about the test results (right not to know).
- The genetic investigation and evaluation is limited to the requested indication and no statements will be made about other diseases.
- All information in this form is true. YES NO

Communication of additional findings found during the course of the research

Yes, I wish to be informed about additional findings.

No, I do not wish to be informed about additional findings.

Date

Physician's Signature



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RELEVANT CLINICAL INFORMATION / [ADD TRANSLATION IN LOCAL LANGUAGE]

Interpretation of the genetic results relies on an accurate and complete clinical picture of the patient, including clinical manifestations, family medical history and previous diagnoses.

Check all boxes that apply to your patient:

Patient has a confirmed or suspected diagnosis of an inherited cardiac disease OR Patient with a clinical index of suspicion for an inherited cardiac disease.

Diagnosis:

Patient has a family member (first or second degree relative) diagnosed with an inherited cardiac disease. Disease:

Patient has a family member who had an unexplained sudden cardiac arrest, aortic event, or who died of an unexplained sudden cardiac death.

Patient has family members who have had genetic testing that identified a specific variant. Gene: Variant:

Patient (child) born with heart defects. Defect:

Patient (child) with (neuro)developmental delay. Symptoms:

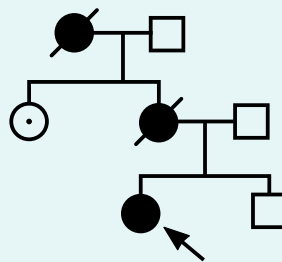
Patients with systemic features of an aortic disorder. Symptoms:

Testing the index patient will improve data interpretation. If this is not the index patient, is he/she available for genetic testing? Yes No N/A

Additional clinical information: e.g., imaging results (X-Ray, MRI, CT), echocardiogram, electrocardiogram, stress test, Holter and event monitor, cardiac catheterization, pulse oximetry, etc. Please provide all relevant medical reports.

PEDIGREE / [ADD TRANSLATION IN LOCAL LANGUAGE]

Example of a pedigree:
sudden cardiac death



SYMBOLS

female		male
	unaffected	
	affected	
	deceased	
	carrier	
	unknown sex	
	spontaneous abortion	
	termination of pregnancy	
	identical twins	
	fraternal twins	
	index patient/ proband	
		infertile

OUR PANELS / [ADD TRANSLATION IN LOCAL LANGUAGE]

- 1 COMPREHENSIVE ARRYTHMIAS & CARDIOMYOPATHIES PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CASQ2, CAV3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, FHL1, FKTN, FLNC, GLA, GPD1L, HCN4, ILK, JPH2, JUP, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ8, KCNQ1, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEBL, NEXN, PKP2, PLN, PRDM16, PRKAG2, RAF1, RANGRF, RBM20, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN5A, SGCD, TAZ, TCAP, TECRL, TGFB3, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRPM4, TTN, VCL
- 2 BRUGADA SYNDROME PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
CACNA1C, CACNB2, GPD1L, HCN4, KCNE3, SCN1B, SCN3B, SCN5A, TRPM4
- 3 CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN
- 4 SHORT QT SYNDROME PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1
- 5 LONG QT SYNDROME PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN
- 6 ARRYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
DSC2, DSG2, DSP, JUP, LMNA, PKP2, TGFB3, TMEM43
- 7 DILATED CARDIOMYOPATHY PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ABCC9, ACADVL, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, CAV3, CHRM2, CPT2, CRYAB, CSRP3, CTF1, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FHL2, FLNC, FKR, FKTN, GATA4, GATA6, GATAD1, ILK, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEBL, NEXN, NKX2-5, NPPA, PKP2, PLN, PDLIM5, RAF1, RBM20, RYR2, SCN5A, SDHA, SGCD, SLC22A5, TAZ, TCAP, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL
- 8 HYPERTROPHIC CARDIOMYOPATHY PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ANKRD1, BAG3, BRAF, CACNA1C, CAV3, CALR3, CBL, CHRM2, CPT2, CSRP3, CTF1, DES, ELAC2, FHL1, FHL2, FLNC, GAA, GATA4, GLA, HRAS, JPH2, KRAS, LAMP2, LDB3, MAP2K1, MAP2K2, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NF1, PLN, PDLIM5, PRKAG2, PTPN11, RAF1, RASA1, SHOC2, SOS1, SPRED1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL
- 9 RESTRICTIVE CARDIOMYOPATHY PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
DES, FLNC, MYBPC3, MYH7, TNNI3, TNNT2
- 10 LEFT VENTRICULAR/NONCOMPACTION CARDIOMYOPATHY PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ACTC1, CASQ2, HCN4, LDB3, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1
- 11 COMPREHENSIVE CONGENITAL HEART DEFECTS PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ACTC1, ACVR2B, ADAMTS10, ARHGAP31, BMPR2, BRAF, CBL, CFAP53, CHD7, CITED2, CREBBP, CRELD1, DNAH11, DNAH5, DNAI1, DOCK6, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HRAS, JAG1, KDM6A, KMT2D, KRAS, LEFTY2, LZTR1, MAP2K1, MAP2K2, MED12, MED13L, MGP, MMP21, MRAS, MYH11, MYH6, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NOTCH1, NOTCH2, NPH4, NR2F2, NRAS, NSD1, PITX2, PKD1L1, PPP1CB, PTPN11, RAF1, RBM10, RBPJ, RIT1, RRAS, SALL1, SALL4, SEMA3E, SHOC2, SMAD6, SOS1, SOS2, SPRED1, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TGFB1, TGFB2, TLL1, ZEB2, ZFPM2, ZIC3
- 12 ALAGILLE SYNDROME PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
JAG1, NOTCH2
- 13 RASOPATHIES WITH HEART DEFECTS PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, SHOC2, SOS1, SOS2
- 14 SYNDROMIC CONGENITAL HEART DEFECTS PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ADAMTS10, ARHGAP31, CHD7, CREBBP, DOCK6, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, GPC3, JAG1, KDM6A, KMT2D, MED12, MGP, MYH11, NIPBL, NOTCH1, NOTCH2, NSD1, PITX2, RBM10, RBPJ, SALL1, SALL4, SEMA3E, TBX3, TBX5, TFAP2B, TGFB1, TGFB2, ZEB2
- 15 ISOLATED CONGENITAL HEART DEFECTS PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ACTC1, BMPR2, CITED2, DTNA, ELN, FOXH1, FOXP1, GATA4, GATA5, GATA6, GJA1, MED13L, MYH6, NKX2-5, NKX2-6, NR2F2, SMAD6, TAB2, TBX1, TBX20, TLL1, ZFPM2
- 16 HETEROTAXY PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
CRELD1, DNAI1, DNAH5, DNAH11, GDF1
- 17 COMPREHENSIVE AORTIC DISORDERS PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ACTA2, BGN, CBS, COL1A1, COL3A1, COL4A5, COL5A1, COL5A2, EFEMP2, ELN, EMILIN1, FBLN5, FBN1, FBN2, FLNA, FOXE3, GATA5, LOX, LTBP3, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2
- 18 EHLERS-DANLOS SYNDROME PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, DSE, EMILIN1, FKBP14, FLNA, PHYKPL, PIEZO2, PLOD1, PLOD3, PRDM5, SLC2A10, SLC39A13, TNXB, ZNF469
- 19 MARFAN SYNDROME PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
FBN1, TGFB1, TGFB2
- 20 MARFAN-LIKE DISORDERS PANEL / [ADD TRANSLATION IN LOCAL LANGUAGE]**
ADAMTS10, ADAMTS17, ADAMTSL2, ADAMTSL4, FBN1, FBN2, LTBP2, LTBP3, MED12, SKI, UPF3B, ZDHHC9

PATIENT INFORMATION / [ADD TRANSLATION IN LOCAL LANGUAGE]

First Name / [Add translation in local language]

Last Name / [Add translation in local language]

Date of Birth (DD/MM/YYYY) / [Add translation in local language]

Gender (male/female/other - specify karyotype) / [Add translation in local language]

Telephone Number (country code & number) / [Add translation in local language]

E-mail Address / [Add translation in local language]

Clinical Diagnosis / [Add translation in local language]

Genetic counselling or counselling by the ordering Physician is necessary before ordering a test in order to inform the patient of all of the possible outcomes and the limitations of the genetic test.

I understand that I will be tested for:

(to be filled in by physician)

I understand that the biological sample will be used to determine if I, or members of my family, are carriers of a genetic variant causing the disease or are carriers of the disease or have an increased risk for developing a disease.

The role of genetic testing. In many cases, a genetic test can directly detect a genetic alteration. Molecular tests can identify structural changes in the DNA (variants). Cytogenetic tests identify the chromosomal changes (structural or numerical). The sensitivity and specificity of each test varies.

The tests offered are complex analyses and are performed using high-end equipment. The methods are externally validated, but there is a minimal possibility of errors.

The significance of the results. If the result is identified as being directly causative of the clinical manifestations, it is considered to be **conclusive**. If the test does not identify the causative mutations of the clinical manifestations, it is considered to be **inconclusive** and this does not preclude other genetic changes (or non-genetic factors) responsible for the disease (a genetic disease or susceptibility to a genetic condition is not excluded). Therefore, an inconclusive result (no causative mutation identified) does not exclude the existence of other pathogenic genetic changes (variants) not tested through the current analysis.

Interpretation of the genetic results relies on a **complete clinical picture of the patient**, including clinical manifestations, family medical history and previous diagnoses. An error in diagnosis could occur due to a clinical picture that is different from that declared. In addition, the test can identify a possible nonpaternity. The test results will be forwarded to the patient by the geneticist or ordering physician and are **confidential**.

By my signature, I hereby certify that:

1. I have been informed of the nature and purpose of the genetic test.
2. I have been informed of the benefits and limitations of the genetic test
by (name of physician)
3. I have been informed that the genetic test can provide information/results which have no connection with the purpose of testing. I understand that only I decide if I want those additional results to be provided.
4. I have received clear answers to my questions in relation to the genetic test.
5. I have received a copy of this form.
6. I agree to provide a sample for the above mentioned genetic test.

I have explained the risks and benefits of the test as well as alternative test methods to the patient. I have answered all the questions from the patients or parent/legal guardian.

Name of the ordering physician

First Name

Signature of the
Ordering Physician

Incidental findings. Genetic testing can provide information unrelated to the purpose of the test, but that may have medical importance for the patient or family (information correlated with an increased risk for incurable disorders).

Use of the sample/result. The sample provided will be used solely for the purpose of the test and for which I have given my written consent.

Test results can also be used for research and to improve the diagnosis and treatment of genetic diseases.

The genetic material can be used for other purposes only with my prior express written consent.

Post-testing genetic counselling. A conclusive result may offer the patient information on the susceptibility, diagnosis, possible prognosis and/or heritability of the disease. An inconclusive result may lead to confusion and anxiety or may suggest the need for further genetic testing. Therefore, post-testing genetic counselling is advised for the clinical interpretation of the results.

Completed by: Patient Parent/Legal Guardian

First Name

Last Name

Date of Completion

Signature

Last Name

Date of Signature