

Kardiomyopathien			Bearbeitungszeit
Panel-ID			
ID007	Hypertrophe Kardiomyopathie (HCM, CMH)	Multigen-Panel (31 Gene) ACTC1, ACTN2, CALR3, CAV3, COX15, CSRP3, GAA, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PLN, PRKAG2, SCO2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL Basis-Panel (17 Gene) ACTC1, ACTN2, CSRP3, JPH2, MYBPC3, MYH7, MYL2, MYL3, NEXN, PLN, PRKAG2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN (spez. Exons)	4 – 6 Wochen
ID008	Dilatative Kardiomyopathie (DCM, CMD)	Multigen-Panel (39 Gene) ABCC9, ACTC1, ACTN2, BAG3, CRYAB, CSRP3, DES, DMD, DSG2, DSP, EYA4, FKTN, GATAD1, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEXN, PLN, PRDM16, RAF1, RBM20, SCN5A, SDHA, SGCD, TAZ, TMPO, TNNC1, TNNT2, TNNI3, TPM1, TTN, TTR, VCL Basis-Panel (10 Gene) BAG3, LMNA, MYBPC3, MYH6, MYH7, PLN, RBM20, TNNT2, TNNI3, TTN (spez. Exons)	4 – 6 Wochen
ID011	Linksventrikuläre Noncompaction-Kardiomyopathie (LVNC, NCCM)	Multigen-Panel (11 Gene) ACTC1, ACTN2, DTNA, LDB3, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1 Basis-Panel (10 Gene) ACTC1, DTNA, LDB3, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1	3 – 5 Wochen
ID010	Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD, ARVC)	Multigen-Panel (9 Gene) DES, DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3, TMEM43 Basis-Panel (8 Gene) DES, DSC2, DSG2, DSP, JUP, PKP2, TGFB3, TMEM43	3 – 5 Wochen
ID105	Restriktive Kardiomyopathie (RCM)	Multigen-Panel (10 Gene) ACTC1, BAG3, DES, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1	3 – 5 Wochen
ID027	Kardiomyopathien, umfassende Diagnostik	Multigen-Panel (96 Gene) ABCC9, ACTA1, ACTC1, ACTN2, ALMS1, ANKRD1, BAG3, BRAF, CALR3, CASQ2, CAV3, CBL, COX15, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FHL2, FKR, FKTN, FXN, GAA, GATAD1, GLA, HADHA, HFE, HRAS, ILK, JPH2, JUP, KLF10, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MIB1, MURC, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOZ2, MYPN, NEXN, NPPA, NRAS, PDLIM3, PKP2, PLN, PRDM16, PRKAG2, PTPN11, RAF1, RBM20, RYR2, SCN2B, SCN5A, SCO2, SDHA, SGCB, SGCD, SGCG, SHOC2, SLC25A4, SOS1, TAZ, TBX20, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TRPM4, TTN, TXNRD2, VCL	4 – 6 Wochen

Panel-ID			Bearbeitungszeit
ID149	Speichererkrankungen mit Herzbeteiligung	Multigen-Panel (12 Gene) ATP7B, FTH1, GAA, GLA, HAMP, HFE, HJV, LAMP2, PRKAG2, SLC40A1, TFR2, TTR	3 – 5 Wochen
ID123	Neuromuskuläre Erkrankungen mit Herzbeteiligung	Multigen-Panel (14 Gene) CAV3, DES, DMD, EMD, FHL1, FKRP, FKTN, LAMA2, LMNA, SGCB, SGCD, SGCG, TCAP, TTN Basis-Panel (14 Gene) CAV3, DES, DMD, EMD, FHL1, FKRP, FKTN, LAMA2, LMNA, SGCB, SGCD, SGCG, TCAP, TTN (spez. Exons)	4 – 6 Wochen

Kongenitale Herzfehler			
Panel-ID			Bearbeitungszeit
ID017	Angeborene nicht-syndromale Herzfehler	Multigen-Panel (33 Gene) ACTC1, ACVR2B, CFAP53, CFC1, CITED2, CRELD1, ELN, GATA4, GATA5, GATA6, GDF1, GJA1, JAG1, MMP21, MED13L, MYH6, ISL1, NKX2-5, NKX2-6, NR2F2, NODAL, NOTCH1, PKD1L1, PRDM6, ROBO4, SMAD6, TAB2, TBX1, TBX20, TFAP2B, TLL1, ZFPM2, ZIC3 Basis-Panel I (Atrium-/Ventrikelseptumdefekt) (12 Gene) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NR2F2, NKX2-5, TBX20, TLL1 Basis-Panel II (Fallot-Tetralogie) (11 Gene) GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2 Basis-Panel III (Heterotaxie) (9 Gene) ACVR2B, CFAP53, CFC1, CRELD1, GDF1, MMP21, NODAL, PKD1L1, ZIC3 Basis-Panel IV (Konotrunkaler Herzfehler) (10 Gene) CFC1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3 Basis-Panel V (Aortenklappenerkrankung) (7 Gene) ELN, GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2 Basis-Panel VI (Hypoplastisches Linksherzsyndrom) (4 Gene) GDF1, GJA1, NKX2-5, NR2F2 Basis-Panel VII (Multipler kongenitaler Herzfehler) (5 Gene) GATA5, GDF1, NR2F2, TAB2, ZIC3	3 – 5 Wochen
ID143	Atrium- und Ventrikelseptumdefekt (ASD, VSD, AVSD)	Multigen-Panel (12 Gene) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NKX2-5, NR2F2, TBX20, TLL1	3 – 5 Wochen

Panel-ID			Bearbeitungszeit
ID144	Falot-Tetralogie (TOF)	Multigen-Panel (11 Gene) GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2	3 – 5 Wochen
ID145	Viszerale Heterotaxie (HTX)	Multigen-Panel (9 Gene) ACVR2B, CFAP53, CFC1, CRELD1, GDF1, MMP21, NODAL, PKD1L1, ZIC3	3 – 5 Wochen
ID160	Konotrunkale Herzfehlbildung (CTHM)	Multigen-Panel (10 Gene) CFC1, GATA6, GATA5, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3	3 – 5 Wochen
ID015	RASopathien	Multigen-Panel (21 Gene) A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PTPN11, RAF1, RASA2, RRAS, PPP1CB, RIT1, SHOC2, SOS1, SOS2, SPRED1 Basis-Panel (17 Gene) [EBM + 25 kb] BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1, SOS2, SPRED1	3 – 5 Wochen
ID252	Angeborene syndromale Herzfehler	Multigen-Panel (49 Gene) ADAMTS10, ADAMTS17, AFF4, ARHGAP31, BMPR2, CDK13, CHD4, CHD7, CREBBP, DLL4, DOCK6, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, GPC3, JAG1, KDM6A, KMT2D, LTBP2, MED12, MGP, MYH11, NIPBL, NOTCH1, NOTCH2, NSD1, PITX2, PLD1, PRKAR1A, PRKD1, RBM10, RBPJ, SALL1, SALL4, SEMA3E, TBX3, TBX5, TGFBR1, TGFBR2, TMEM260, WDPCP, ZEB2 Basis-Panel I (CHARGE-Syndrom) (2 Gene) CHD7, SEMA3E Basis-Panel II (Weill-Marchesani-Syndrom) (4 Gene) ADAMTS10, ADAMTS17, FBN1, LTBP2 Basis-Panel III (Adams-Oliver-Syndrom) (6 Gene) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ Basis-Panel IV (Kabuki-Syndrom) (2 Gene) KDM6A, KMT2D Basis-Panel V (Alagille-Syndrom) (2 Gene) JAG1, NOTCH2 Basis-Panel VI (Marfan-Syndrom) (3 Gene) [EBM] FBN1, TGFBR1, TGFBR2 Basis-Panel VII (Rubinstein-Taybi-Syndrom) (2 Gene) CREBBP, EP300	4 – 6 Wochen

Arrhythmien / Ionenkanalerkrankungen			
Panel-ID			Bearbeitungszeit
ID013	Long-QT-Syndrom (LQT)	Multigen-Panel (14 Gene) AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1 Basis-Panel (12 Gene) CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1	3 – 5 Wochen
ID233	Short-QT-Syndrom (SQT)	Multigen-Panel (6 Gene) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1	3 – 5 Wochen
ID014	Brugada-Syndrom (BRGDA)	Multigen-Panel (15 Gene) CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, TRPM4 Basis-Panel (12 Gene) CACNA1C, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNJ8, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A	3 – 5 Wochen
ID012	Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT)	Multigen-Panel (6 Gene) ANK2, CALM1, CASQ2, KCNJ2, RYR2, TRDN Basis-Panel (5 Gene) CALM1, CASQ2, KCNJ2, RYR2, TRDN	3 – 5 Wochen
ID016	Familiäres Vorhofflimmern (ATFB)	Multigen-Panel (14 Gene) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, NPPA, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A	3 – 5 Wochen
ID107	Sick-Sinus-Syndrom (SSS)	Multigen-Panel (3 Gene) HCN4, MYH6, SCN5A	3 – 5 Wochen
ID026	Arrhythmien, umfassende Diagnostik	Multigen-Panel (49 Gene) ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CASQ2, CAV3, DES, DSC2, DSG2, DSP, EMD, GJA5, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LMNA, MYH6, NKX2-5, NPPA, PKP2, PRKAG2, RANGRF, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SNTA1, TBX5, TGFB3, TMEM43, TNNI3, TRPM4, TRDN	4 – 6 Wochen

Arterien-/Aortenerkrankungen			
Panel-ID			Bearbeitungszeit
ID020	Thorakales Aortenaneurysma und Aortendissektion (AAT, TAAD)	Multigen-Panel (9 Gene) [EBM] ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2	3 – 5 Wochen
ID009	Loeys-Dietz-Syndrom (LDS) und ähnliche Aortenerkrankungen	Multigen-Panel (31 Gene) ACTA2, BGN, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FLNA, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR2 Basis-Panel I (21 Gene) [EBM + 25 kb] ACTA2, BGN, COL3A1, FBN1, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PRKG1, ROBO4, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBR1, TGFBR* Basis-Panel II (Thorakales Aortenaneurysma/-dissektion) (9 Gene) [EBM] ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2 Basis-Panel III (Thorakales Aortenaneurysma, nicht-syndromal) (10 Gene) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2 Basis-Panel IV (Loeys-Dietz-Syndrom) (8 Gene) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Basis-Panel V (Aortenklappenerkrankung) (3 Gene) NOTCH1, ROBO4, SMAD6	4 – 6 Wochen
ID234	Hämorrhagischer und Ischämischer Schlaganfall	Multigen-Panel (39 Gene) ACE, ACTA2, ADA2, ALOX5AP, APOE, APP, CBS, CCM2, COL3A1, COL4A1, COL4A2, CST3, F2, F5, FBN1, FLNA, GUCYA1, GLA, HTRA1, ITM2B, JAG1, KRIT1, MTHFR, MYH11, MYLK, NOS3, NOTCH3, OTC, PDCD10, POLG, PRKCH, RNF213, SLC2A10, SMAD3, TGFB2, TGFBR1, TGFBR2, TREX1, TTR Basis-Panel I (17 Gene) ACTA2, APP, CBS, COL3A1, COL4A1, COL4A2, FBN1, GLA, HTRA1, MYH11, MYLK, NOTCH3, SMAD3, TGFB2, TGFBR1, TGFBR2, TREX1 Basis-Panel II (Intrazerebrale Hämorrhagie) (5 Gene) ACE, APP, CST3, COL4A1, COL4A2 Basis-Panel III (Moyamoya-Erkrankung) (4 Gene) ACTA2, JAG1, GUCY1A1, RNF213 Basis-Panel IV (Aortenaneurysma/-dissektion) (9 Gene) [EBM] ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2 Basis-Panel V (CADASIL, CARASIL) (2 Gene) HTRA1, NOTCH3	4 – 6 Wochen

Panel-ID			Bearbeitungszeit
ID109	Cutis laxa (ADCL, ARCL)	Multigen-Panel (10 Gene) ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP4, PYCR1	3 – 5 Wochen
ID039	Ehlers-Danlos-Syndrom (EDS)	Multigen-Panel (20 Gene) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Basis-Panel I (EDS, dominant) (7 Gene) [EBM + 25 kb] C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2 Basis-Panel II (EDS, rezessiv) (11 Gene) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13	3 – 5 Wochen
ID022	Marfan-Syndrom (MFS)	Multigen-Panel (3 Gene) [EBM] FBN1, TGFB2, TGFBR1	3 – 5 Wochen
ID194	Marfan-Syndrom (MFS) und ähnliche Krankheitsbilder	Multigen-Panel (42 Gene) ABCD4, ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL11A1, FBN1, FBN2, FKBP14, FLNA, LIG4, LMBRD1, LOX, LTBP2, PLOD, MED12, MMACHC, MMADHC, MTHFR, MTR, MTRR, MYH11, MYLK, PRDM5, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469 Basis-Panel I (18 Gene) [EBM + 25 kb] ACTA2, BGN, CBS, COL3A1, FBN1, FBN2, LOX, LTBP2, MYLK, MYH11, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Basis-Panel II (Marfan-Syndrom) (3 Gene) [EBM] FBN1, TGFB2, TGFBR1 Basis-Panel III (Aortopathie) (9 Gene) [EBM] ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFBR1, TGFBR2 Basis-Panel IV (Homocystinurie) (8 Gene) ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR Basis-Panel V (Weill-Marchesani-Syndrom) (4 Gene) ADAMTS10, ADAMTS17, FBN1, LTBP2 Basis-Panel VI (Stickler-Syndrom) (4 Gene) COL2A1, COL9A1, COL9A2, COL11A1 Basis-Panel VII (Ehlers-Danlos-Syndrom) (6 Gene) [EBM + 25 kb] COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PRDM5 Basis-Panel VIII (Brittle-Cornea-Syndrom) (2 Gene) PRDM5, ZNF469	4 – 6 Wochen

<p>ID137</p>	<p>Bindegewebserkrankungen mit Aortenbeteiligung</p>	<p>Multigen-Panel (60 Gene) ABCC6, ACTA2, ADAMTS2, AEBP1, ALDH18A1, ATP6VOA2, ATP6V1, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL1A1, COL1A2, COL3A1, COL4A1, COL5A1, COL5A2, COL12A1, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, ITPKC, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PLOD3, PYCR1, RIN2, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2, TNXB, ZNF469</p> <p>Basis-Panel I (Thorakales Aortenaneurysma/-dissektion) (9 Gene) [EBM] ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB1, TGFB2</p> <p>Basis-Panel II (Marfan-Syndrom) (3 Gene) [EBM] FBN1, TGFB2, TGFB1</p> <p>Basis-Panel III (Ehlers-Danlos-Syndrom, dominant) (7 Gene) [EBM + 25 kb] C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2</p> <p>Basis-Panel IV (Ehlers-Danlos-Syndrom, rezessiv) (11 Gene) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13</p> <p>Basis-Panel V (Cutis laxa) (8 Gene) ALDH18A1, ATP6VOA2, ATP7A, EFEMP2, ELN, FBLN5, LTBP4, PYCR1</p> <p>Basis-Panel VI (Loeys-Dietz-Syndrom) (8 Gene) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2</p> <p>Basis-Panel VII (Aortenaneurysma, nicht-syndromal) (10 Gene) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2</p> <p>Basis-Panel VIII (Aortenklappenerkrankung) (3 Gene) NOTCH1, ROBO4, SMAD6</p>	<p>4 – 6 Wochen</p>
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Kosten

Die Kosten werden bei bestehender medizinischer Indikation über einen Überweisungsschein Typ 10 (EBM) abgerechnet. Humangenetische Leistungen sind nicht budgetrelevant.

Es können bis zu 25 Kilobasen (kb) kodierende Sequenzen im Rahmen einer Stufendiagnostik abgerechnet werden (siehe ggf. Basispanels). Mutationssuchen in mehr als 25 kb unterliegen aktuell noch einer Genehmigungspflicht durch die gesetzliche Krankenversicherung.

Für privatversicherte Patienten sowie private Kostenträger (Krankenhäuser etc.) können auf Wunsch entsprechende Kostenvoranschläge erstellt werden.