

Augenerkrankungen			
Panel-ID			Bearbeitungszeit
ID050	Retinitis pigmentosa (RP), X-chromosomal oder autosomal-rezessiv	Multigen-Panel (57 Gene) ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, C8ORF37, CDHR1, CERKL, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, OFD1, PCARE, PDE6A*, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP2, RPE65, RPGR, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513 Basis-Panel I (autosomal-rezessiv) (2 Gene) EYS, USH2A Basis-Panel II (autosomal-rezessiv) (9 Gene) ABCA4, CERKL, CRB1, MERTK, PDE6A, PDE6B, RHO, RPE65, TULP1 Basis-Panel III (X-chromosomal) (3 Gene) RP2, RPGR, OFD1	4 - 6 Wochen
ID053	Retinitis pigmentosa (RP), X-chromosomal oder autosomal-dominant	Multigen-Panel (28 Gene) AIPL1, ARL3, BEST1, CA4, FSCN2, GUCA1B, HK1, IMPDH1, KLHL7, NR2E3, NRL, OFD1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RGR, RHO, RP1, RP2, RP9, RPE65, RPGR, SEMA4A, SNRNP200, TOPORS Basis-Panel I (autosomal-dominant) (10 Gene) GUCA1B, IMPDH1, KLHL7, NR2E3, PRPF31, PRPF8, PRPH2, RHO, RP1, SEMA4A Basis-Panel II (X-chromosomal) (3 Gene) RP2, RPGR, OFD1	3 - 5 Wochen
ID101	Zapfen- und Zapfen-Stäbchen-Dystrophie (COD, CORD)	Multigen-Panel (30 Gene) ABCA4, ADAM9, AIPL1, C8ORF37, CACNA1F, CACNA2D4, CDHR1, CEP78, CEP250, CNNM4, CRX, DRAM2, GUCA1A, GUCY2D, KCNV2, OPN1LW, OPN1MW, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RPGR, RPGRIP1, SEMA4A, TTL5, UNC119 Basis-Panel (12 Gene) ABCA4, C8ORF37, CDHR1, CRX, DRAM2, GUCA1A, GUCY2D, KCNV2, PDE6C, POC1B, PROM1, RAB28	3 - 5 Wochen
ID182	Dysgenesie des vorderen Augensegmentes (ASGD)	Multigen-Panel (8 Gene) CPAMD8, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN	3 - 5 Wochen

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ID034	Usher-Syndrom (USH)	Multigen-Panel (13 Gene) ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN Basis-Panel I (USH1) (5 Gene) CDH23, CIB2, MYO7A, PCDH15, USH1C Basis-Panel II (USH2) (3 Gene) ADGRV1, PDZD7, WHRN Basis-Panel II (USH2, USH3, USH4) (5 Gene) ARSG, CLRN1, HARS1, USH2A, WHRN	3 - 5 Wochen
ID187	Lebersche kongenitale Amaurose (LCA)	Multigen-Panel (19 Gene) AIPL1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1, USP45 Basis-Panel (9 Gene) AIPL1, CEP290, CRB1, CRX, GUCY2D, IMPDH1, RDH12, RPE65, RPGRIP1	3 - 5 Wochen
ID267	Kongenitale stationäre Nachtblindheit (CSNB)	Multigen-Panel (14 Gene) CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RHO, SAG, SLC24A1, TRPM1 Basis-Panel (7 Gene) CACNA1F, GPR179, GRK1, GRM6, NYX, SAG, TRPM1	3 - 5 Wochen
ID164	Achromatopsie (ACHM)	Multigen-Panel (8 Gene) ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, OPN1LW, OPN1MW	2 - 4 Wochen
ID079	Myopie (MYP)	Multigen-Panel (15 Gene) ARR3, CPSF1, COL11A1, COL2A1, COL9A1, COL9A2, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLTRK6, ZNF644 Basis-Panel I (11 Gene) ARR3, CPSF1, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLTRK6, ZNF644 Basis-Panel II (Stickler-Syndrom) (4 Gene) COL11A1, COL2A1, COL9A1, COL9A2	3 - 5 Wochen

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ID206	Katarakt (CTRCT)	<p>Multigen-Panel (37 Gene)</p> <p>AGK, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, DNMBP, EPHA2, FOXE3, FYCO1, GCNT2, GJA3, GJA8, HSF4, LEMD2, LIM2, LSS, MAF, MIP, NHS, PITX3, SIPA1L3, SLC16A12, TDRD7, UNC45B, VIM, WFS1</p> <p>Basis-Panel I (autosomal-dominant) (24 Gene)</p> <p>BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, EPHA2, GJA3, GJA8, HSF4, MAF, MIP, PITX3, SLC16A12, VIM</p> <p>Basis-Panel II (autosomal-rezessiv) (14 Gene)</p> <p>AGK, BFSP1, CRYAA, CRYAB, CRYBB1, CRYBB3, DNMBP, FOXE3, FYCO1, GCNT2, LEMD2, LIM2, LSS, TDRD7</p>	3 - 5 Wochen
ID263	Mikrophthalmie, Anophthalmie und Kolobom	<p>Multigen-Panel (47 Gene)</p> <p>ABC6, ACTB, ACTG1, ALDH1A3, BCOR, BMP4, CHD7, C12ORF57, CC2D2A, CRYAA, FOXE3, GDF3, GDF6, GLI2, HCCS, HMGB3, HMX1, MAB21L2, MAF, MFRP, MITF, NAA10, OTX2, PAX2, PAX6, PDE6D, PITX3, PTCH1, PRSS56, RBP4, RARB, RAX, RPPRIP1L, SALL2, SEMA3A, SHH, SIX3, SMOC1, SOX2, SRD5A3, STRA6, TENM3, TFAP2A, TMEM67, VAX1, VSX2, YAP1</p> <p>Basis-Panel I (Mikrophthalmie) (17 Gene)</p> <p>ABC6, ALDH1A3, BCOR, BMP4, HCCS, HMGB3, MAB21L2, MFRP, NAA10, OTX2, PRSS56, RBP4, RARB, RAX, SOX2, STRA6, VSX2</p> <p>Basis-Panel II (Kolobom) (11 Gene)</p> <p>ABC6, GDF3, GDF6*, PAX6, RBP4, SALL2, SHH, STRA6, TENM3, VSX2, YAP1</p>	3 - 5 Wochen
ID186	Altersbedingte Makuladegeneration (ARMD)	<p>Multigen-Panel (16 Gene)</p> <p>ABCA4, APOE, ARMS2, C2, C3, C9, CFB, CFH, CFI, CST3, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, RAX2</p> <p>Basis-Panel (11 Gene)</p> <p>ABCA4, ARMS2, C3, C9, CFB, CFH, CFI, FBLN5, HTRA1, RAX2</p>	3 - 5 Wochen
ID139	Makuladystrophie (MD)	<p>Multigen-Panel (15 Gene)</p> <p>ABCA4, BEST1, CDH3, CNGB3, CTNNA1, ELOVL4, IMPG1, IMPG*, MAPKAPK3, MFSD8, PRDM13, PROM1, PRPH2, RP1L1, TIMP3</p> <p>Basis-Panel (10 Gene)</p> <p>ABCA4, BEST1, CDH3, CNGB3, ELOVL4, IMPG1, IMPG2, PROM1, PRPH2, TIMP3</p>	3 - 5 Wochen

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ID102	Morbus Stargardt (STGD)	Multigen-Panel (5 Gene) ABCA4, CNGB3, ELOVL4, PROM1, PRPH2	2 - 4 Wochen
ID261	Fuchs-Endotheldystrophie (FECD)	Multigen-Panel (5 Gene) AGBL1, COL8A2, SLC4A11, TCF4, ZEB1	2 - 4 Wochen
ID063	Kongenitale extraokuläre Muskelfibrose (CFEOM)	Multigen-Panel (5 Gene) COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3	2 - 4 Wochen
ID081	Optikusatrophie (OPA)	Multigen-Panel (22 Gene) ACO2, ATP1A3, ANTXR1, C12ORF65, CISD2, DNM1L, FDXR, MECR, MFN2, NR2F1, OPA1, OPA3, PDXK, PRPS1, RTN4IP1, SLC25A46, TBCE, TMEM126A, TIMM8A, UCHL1, WFS1, YME1L1 Basis-Panel (18 Gene) ACO2, C12ORF65, CISD2, DNM1L, FDXR, MECR, MFN2, NR2F1, OPA1, OPA3, PDXK, PRPS1, RTN4IP1, SLC25A46, TMEM126A, TIMM8A, WFS1, YME1L1	3 - 5 Wochen
ID082	Okulokutaner Albinismus (OCA)	Multigen-Panel (8 Gene) GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	2 - 4 Wochen
ID175	Syndromaler Albinismus	Multigen-Panel (22 Gene) AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, EDN3, EDNRB, HPS1, HPS3, HPS4, HPS5, HPS6, KITLG, LYST, MITF, MLPH, MYO5A, PAX3, RAB27A, SNAI2, SOX10, TYR Basis-Panel I (Hermansky-Pudlak-Syndrom) (10 Gene) AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6 Basis-Panel II (Waardenburg-Syndrom) (8 Gene) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Basis-Panel III (Griscelli-Syndrom) (3 Gene) MLPH, MYO5A, RAB27A	3 - 5 Wochen
ID062	Stickler-Syndrom (STL)	Multigen-Panel (6 Gene) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2	3 - 5 Wochen
ID230	Weill-Marchesani-Syndrom (WMS)	Multigen-Panel (4 Gene) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 Wochen

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ID178	Walker-Warburg-Syndrom (MDDGA)	Multigen-Panel (14 Gene) B3GALNT2, B4GAT1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 Wochen
ID029	Senior-Loken-Syndrom (SLSN)	Multigen-Panel (8 Gene) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19 Basis-Panel (7 Gene) CEP290, IQCB1, NPHP1, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 Wochen
ID093	Bardet-Biedl-Syndrom (BBS)	Multigen-Panel (21 Gene) ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP Basis-Panel (16 Gene) ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS1*, IFT27, LZTFL1, MKKS, MKS1, TTC8, WDPCP	3 - 5 Wochen
ID084	Zellweger-Syndrom (ZWS)	Multigen-Panel (14 Gene) ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	3 - 5 Wochen
ID083	Störung der peroxisomalen Biogenese (PBD)	Multigen-Panel (14 Gene) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Basis-Panel I (Zellweger-Syndrom) (12 Gene) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Basis-Panel II (Neonatale Adrenoleukodystrophie/Infantiles Refsum-Syndrom) (12 Gene) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX26	3 - 5 Wochen

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.