

Analyse af gener relevante ved mistanke om kardiel arytm (udvidet panel)

Panelbeskrivelse

Navn: Analyse af gener relevante ved mistanke om kardiel arytm (udvidet panel)

Antal gener: 279

version: 2.0

Ibrugtagningsdato: 2026-01-23

Metodebeskrivelse: Prøveforberedelse og sekventering: Der udføres helgenomsekventering (WGS) med Illumina PCR free library prep (tagmentation) og Illumina sekventering (NovaSeq). Middelsekventeringsdybden for analysen er minimum 30X og minimumsandelen af genomet, der er dækket minimum 15X, er 95 %. Middelsekventeringsdybden og andelen, der i gennemsnit er dækket minimum 15X, for de enkelte gener kan ses i nedenstående tabel.

Dataanalyse: Referencegenom: hg38. Alignment og variantkald foretages med hhv. BWA og GATK. Data analyseres for strukturelle varianter vha. Manta og GATK-gCNV. Der foretages *in silico* filtrering af data til relevante genregioner baseret på nedenstående tabel. Varianter med allelfrekvens mindre end 15% filtreres som udgangspunkt fra. For alle, eller de specifikt anførte, gener undersøges der for varianter i de proteinkodende regioner og de nærmeste 10 bp i de tilstødende ikke-proteinkodende regioner, mtDNA, samt varianter i ikke-proteinkodende regioner indberettet som sandsynlig patogene (C4) eller patogene (C5) i ClinVar. Varianter klassificeres iht. ACMGs klassifikationsguideline (PMID: 25741868), og hvis tilgængelig og relevant benyttes ClinGens genspecifikke guideline. Varianter af ukendt klinisk betydning (C3) anføres kun i svaret, hvis de vurderes relevante i forhold til sygdomsbilledet. Anlægsbærerstatus for recessiv sygdom rapporteres ikke nødvendigvis. Genomsekventering er en screeningsmetode, og ikke alle varianter kan med sikkerhed identificeres. Den udførte analyse har ikke samme sensitivitet som DNA-microarrayanalyse ift. strukturelle varianter.

Genliste

Gener, hvor middelsekventeringsdybden er < 30X, er markeret med¹. Gener, hvor andelen af de kodede regioner, der dækket minimum 15X, er < 95 % er markeret med². Gener, der derudover vides at være suboptimalt dækket, er markeret med³. Værdier er angivet med +/- standardafvigelse.

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
AARS2	51.27 +/- 8.69	100.0 +/- 0.0	99.95 +/- 0.36	98.37 +/- 5.16
ABCC9	55.36 +/- 7.4	100.0 +/- 0.0	99.96 +/- 0.13	98.98 +/- 1.65
ACAD9	53.83 +/- 8.75	99.99 +/- 0.03	99.99 +/- 0.03	98.88 +/- 4.15
ACADVL	49.39 +/- 7.84	100.0 +/- 0.0	99.73 +/- 0.48	96.66 +/- 5.99
ACTA1	48.02 +/- 8.99	100.0 +/- 0.0	99.7 +/- 1.98	95.34 +/- 9.73
ACTN2	54.14 +/- 7.9	100.0 +/- 0.0	99.86 +/- 0.66	98.63 +/- 2.76
AGK	54.18 +/- 7.97	100.0 +/- 0.0	100.0 +/- 0.0	99.5 +/- 1.18
AGL	55.5 +/- 7.31	100.0 +/- 0.0	100.0 +/- 0.01	99.4 +/- 1.39
AKAP9	54.78 +/- 7.26	100.0 +/- 0.0	99.99 +/- 0.04	99.54 +/- 1.01
ALMS1	53.24 +/- 7.52	100.0 +/- 0.0	99.98 +/- 0.06	99.18 +/- 1.66
ALPK3	52.16 +/- 8.54	100.0 +/- 0.0	99.96 +/- 0.11	98.25 +/- 3.68
ANK2	54.32 +/- 7.66	100.0 +/- 0.0	100.0 +/- 0.01	99.3 +/- 2.01
ANKRD1	51.79 +/- 7.4	99.99 +/- 0.06	99.94 +/- 0.22	97.8 +/- 3.99
ANO5	54.74 +/- 7.51	99.97 +/- 0.05	99.96 +/- 0.05	99.22 +/- 1.81
ARSB	52.71 +/- 8.31	100.0 +/- 0.0	99.95 +/- 0.27	98.79 +/- 3.51
ATAD3A	48.89 +/- 8.14	99.97 +/- 0.21	99.32 +/- 1.57	93.42 +/- 7.9
ATP5F1D	50.27 +/- 8.99	100.0 +/- 0.0	100.0 +/- 0.0	97.49 +/- 5.86
ATPAF2	53.26 +/- 8.18	100.0 +/- 0.0	99.93 +/- 0.46	99.08 +/- 3.76
BAG3	49.59 +/- 8.11	99.95 +/- 0.05	99.68 +/- 1.08	96.74 +/- 7.05
BRAF	53.15 +/- 6.81	99.99 +/- 0.05	99.51 +/- 1.19	97.12 +/- 2.41

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>CACNA1C</i>	51.85 +/- 7.82	100.0 +/- 0.0	99.82 +/- 0.82	98.29 +/- 3.77
<i>CACNA1D</i>	53.11 +/- 8.01	99.99 +/- 0.03	99.86 +/- 0.41	98.66 +/- 3.17
<i>CACNA2D1</i>	55.07 +/- 7.4	100.0 +/- 0.0	99.99 +/- 0.08	99.34 +/- 1.35
<i>CACNB2</i>	54.16 +/- 7.67	100.0 +/- 0.0	99.99 +/- 0.04	98.89 +/- 2.61
<i>CALM1</i>	53.25 +/- 8.22	100.0 +/- 0.0	100.0 +/- 0.03	99.08 +/- 3.01
<i>CALM2</i>	53.59 +/- 8.08	100.0 +/- 0.0	100.0 +/- 0.0	98.86 +/- 2.99
<i>CALM3</i>	45.09 +/- 8.14	100.0 +/- 0.01	99.34 +/- 1.51	92.63 +/- 10.4
<i>CASQ2</i>	52.52 +/- 8.45	100.0 +/- 0.02	99.99 +/- 0.03	99.05 +/- 2.64
<i>CAV3</i>	50.31 +/- 8.29	100.0 +/- 0.01	99.75 +/- 1.61	98.3 +/- 6.89
<i>CAVIN4</i>	54.02 +/- 8.14	100.0 +/- 0.0	99.98 +/- 0.12	99.25 +/- 2.67
<i>CBL</i>	53.02 +/- 7.87	100.0 +/- 0.0	99.95 +/- 0.18	98.83 +/- 3.14
<i>CDH2</i>	55.66 +/- 7.43	99.99 +/- 0.06	99.92 +/- 0.31	99.21 +/- 1.51
<i>CHRM2</i>	54.51 +/- 7.85	100.0 +/- 0.0	99.99 +/- 0.05	99.34 +/- 2.4
<i>COA5</i>	55.2 +/- 8.31	100.0 +/- 0.0	99.96 +/- 0.24	99.39 +/- 1.74
<i>COA6</i>	56.07 +/- 7.27	100.0 +/- 0.0	99.97 +/- 0.18	99.75 +/- 0.97
<i>COX10</i>	55.39 +/- 8.65	100.0 +/- 0.0	99.96 +/- 0.28	98.27 +/- 6.06
<i>COX14</i>	54.56 +/- 8.99	100.0 +/- 0.0	100.0 +/- 0.0	98.46 +/- 9.88
<i>COX15</i>	54.03 +/- 7.18	100.0 +/- 0.0	100.0 +/- 0.0	99.61 +/- 0.85
<i>COX20</i>	53.46 +/- 7.67	100.0 +/- 0.0	99.9 +/- 0.69	99.04 +/- 3.19
<i>COX6B1</i>	50.95 +/- 8.8	100.0 +/- 0.0	99.92 +/- 0.55	98.48 +/- 4.99
<i>COX7B</i>	43.15 +/- 15.63	99.95 +/- 0.24	94.98 +/- 10.42	72.09 +/- 34.27
<i>CPT2</i>	52.9 +/- 8.76	100.0 +/- 0.0	99.99 +/- 0.04	98.76 +/- 4.32
<i>CRYAB</i>	51.85 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.0	99.16 +/- 3.52
<i>CSRP3</i>	55.56 +/- 7.24	100.0 +/- 0.0	100.0 +/- 0.0	99.74 +/- 0.84
<i>CTF1</i>	46.9 +/- 8.81	99.98 +/- 0.15	99.27 +/- 2.72	94.3 +/- 9.57
<i>CTNNA3</i>	55.0 +/- 7.3	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 1.21
<i>DES</i>	49.28 +/- 8.63	100.0 +/- 0.0	99.84 +/- 0.81	97.12 +/- 6.83
<i>DMD</i>	43.26 +/- 15.85	99.96 +/- 0.14	94.99 +/- 8.6	71.25 +/- 33.8
<i>DNAJC19</i>	54.85 +/- 7.36	100.0 +/- 0.0	100.0 +/- 0.0	98.86 +/- 2.88
<i>DOLK</i>	47.05 +/- 8.6	100.0 +/- 0.0	99.57 +/- 2.64	95.74 +/- 10.02
<i>DSC2</i>	54.17 +/- 7.27	100.0 +/- 0.0	100.0 +/- 0.03	99.31 +/- 1.7
<i>DSG2</i>	55.04 +/- 8.11	100.0 +/- 0.01	99.97 +/- 0.17	99.04 +/- 2.66
<i>DSP</i>	53.11 +/- 7.87	100.0 +/- 0.0	99.99 +/- 0.06	98.73 +/- 2.87
<i>DTNA</i>	53.93 +/- 7.86	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.61
<i>ELAC2</i>	52.76 +/- 7.36	99.88 +/- 0.1	99.83 +/- 0.12	98.86 +/- 3.07
<i>EMD</i>	39.58 +/- 15.58	99.34 +/- 2.41	88.72 +/- 16.69	62.52 +/- 39.24
<i>EPG5</i>	54.72 +/- 7.78	100.0 +/- 0.0	99.98 +/- 0.09	99.28 +/- 2.18
<i>ETFA</i>	54.01 +/- 6.71	100.0 +/- 0.03	99.99 +/- 0.04	99.47 +/- 1.15
<i>ETFB</i>	49.53 +/- 9.19	100.0 +/- 0.0	99.88 +/- 0.78	96.98 +/- 8.4
<i>ETFDH</i>	55.85 +/- 7.84	99.95 +/- 0.11	99.79 +/- 0.25	99.09 +/- 1.77
<i>EYA4</i>	54.97 +/- 7.78	100.0 +/- 0.0	99.96 +/- 0.22	99.34 +/- 1.81
<i>FAH</i>	51.3 +/- 8.58	100.0 +/- 0.0	99.76 +/- 0.84	97.67 +/- 6.81
<i>FASTKD2</i>	54.34 +/- 7.88	100.0 +/- 0.0	99.98 +/- 0.06	99.21 +/- 2.06
<i>FHL1</i>	42.18 +/- 16.05	99.99 +/- 0.09	93.3 +/- 11.78	68.92 +/- 36.87
<i>FHL2</i>	52.41 +/- 7.73	100.0 +/- 0.0	100.0 +/- 0.03	98.52 +/- 4.01
<i>FHOD3</i>	51.21 +/- 7.81	100.0 +/- 0.0	99.75 +/- 0.6	97.22 +/- 3.32

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>FKRP</i>	57.39 +/- 9.1	100.0 +/- 0.0	99.83 +/- 0.47	98.43 +/- 4.01
<i>FKTN</i>	54.54 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.0	99.4 +/- 1.72
<i>FLII</i>	52.18 +/- 8.63	100.0 +/- 0.0	99.77 +/- 0.74	97.35 +/- 5.91
<i>FLNC</i>	52.53 +/- 8.91	100.0 +/- 0.01	99.94 +/- 0.25	97.94 +/- 6.75
<i>FNIP1</i>	55.29 +/- 7.23	100.0 +/- 0.0	99.99 +/- 0.07	99.52 +/- 0.91
<i>FOXD4</i>	62.38 +/- 14.97	100.0 +/- 0.0	99.79 +/- 1.28	97.86 +/- 8.39
<i>FOXRED1</i>	50.43 +/- 8.93	100.0 +/- 0.0	99.84 +/- 1.07	97.64 +/- 7.2
<i>FXN</i>	53.72 +/- 7.79	99.99 +/- 0.03	99.96 +/- 0.14	99.15 +/- 2.07
<i>GAA</i>	51.67 +/- 8.38	100.0 +/- 0.0	99.95 +/- 0.27	98.28 +/- 5.14
<i>GATA4</i>	51.22 +/- 7.65	100.0 +/- 0.0	99.98 +/- 0.09	98.1 +/- 4.44
<i>GATA5</i>	52.14 +/- 8.69	100.0 +/- 0.0	99.83 +/- 0.73	97.2 +/- 6.42
<i>GATA6</i>	53.05 +/- 7.54	100.0 +/- 0.0	99.78 +/- 1.08	96.74 +/- 7.21
<i>GATAD1</i>	55.32 +/- 7.66	100.0 +/- 0.0	99.99 +/- 0.04	99.26 +/- 2.49
<i>GBE1</i>	55.41 +/- 7.39	100.0 +/- 0.0	100.0 +/- 0.0	99.5 +/- 1.34
<i>GFM1</i>	55.96 +/- 7.47	100.0 +/- 0.0	100.0 +/- 0.01	99.71 +/- 0.6
<i>GJA5</i>	53.59 +/- 8.52	99.94 +/- 0.16	99.74 +/- 0.53	98.85 +/- 2.67
<i>GLA</i>	43.47 +/- 15.87	99.99 +/- 0.04	95.69 +/- 8.38	73.05 +/- 32.79
<i>GLB1</i>	53.66 +/- 8.07	100.0 +/- 0.0	99.98 +/- 0.1	99.15 +/- 2.5
<i>GNB5</i>	51.66 +/- 7.44	99.69 +/- 0.71	97.26 +/- 1.87	93.43 +/- 3.44
<i>GPD1L</i>	54.31 +/- 7.92	100.0 +/- 0.0	99.91 +/- 0.5	98.77 +/- 3.19
<i>GSN</i>	50.81 +/- 8.49	100.0 +/- 0.0	99.98 +/- 0.13	97.93 +/- 5.86
<i>GUSB</i>	51.2 +/- 8.77	100.0 +/- 0.0	99.96 +/- 0.17	97.88 +/- 4.9
<i>GYG1</i>	55.51 +/- 8.27	99.99 +/- 0.03	99.99 +/- 0.03	99.3 +/- 1.64
<i>HADHA</i>	53.16 +/- 7.94	100.0 +/- 0.0	99.91 +/- 0.35	98.23 +/- 5.5
<i>HADHB</i>	55.8 +/- 7.59	100.0 +/- 0.0	100.0 +/- 0.0	99.73 +/- 0.95
<i>HAMP</i>	48.16 +/- 8.88	100.0 +/- 0.0	100.0 +/- 0.0	97.47 +/- 6.47
<i>HCN4</i>	49.55 +/- 8.05	100.0 +/- 0.0	99.82 +/- 0.92	96.09 +/- 8.54
<i>HGSNAT</i>	53.32 +/- 7.96	100.0 +/- 0.0	99.99 +/- 0.06	99.0 +/- 2.22
<i>HJV</i>	50.53 +/- 8.04	100.0 +/- 0.0	99.98 +/- 0.12	98.32 +/- 4.4
<i>HRAS</i>	49.95 +/- 9.48	99.95 +/- 0.17	99.17 +/- 2.84	94.67 +/- 10.68
<i>IDH2</i>	51.69 +/- 8.89	100.0 +/- 0.0	99.94 +/- 0.43	98.34 +/- 5.49
<i>IDS</i>	42.84 +/- 15.64	99.95 +/- 0.34	94.77 +/- 9.21	70.78 +/- 33.81
<i>IDUA</i>	50.12 +/- 9.17	100.0 +/- 0.0	99.59 +/- 1.62	95.77 +/- 9.73
<i>ILK</i>	51.64 +/- 9.11	100.0 +/- 0.0	99.98 +/- 0.11	98.05 +/- 6.09
<i>JPH2</i>	51.52 +/- 8.0	100.0 +/- 0.0	99.92 +/- 0.44	98.08 +/- 5.91
<i>JUP</i>	49.95 +/- 8.81	100.0 +/- 0.0	99.87 +/- 0.83	97.71 +/- 6.07
<i>KCNA5</i>	52.25 +/- 8.12	100.0 +/- 0.0	99.96 +/- 0.19	98.07 +/- 5.4
<i>KCND2</i>	53.24 +/- 7.83	99.99 +/- 0.06	99.9 +/- 0.56	98.37 +/- 4.14
<i>KCND3</i>	51.86 +/- 8.09	100.0 +/- 0.0	99.88 +/- 0.3	98.01 +/- 4.26
<i>KCNE1</i>	53.64 +/- 8.87	100.0 +/- 0.0	100.0 +/- 0.0	99.15 +/- 4.37
<i>KCNE2</i>	54.11 +/- 7.6	100.0 +/- 0.0	100.0 +/- 0.0	99.48 +/- 1.71
<i>KCNE3</i>	49.9 +/- 8.75	99.97 +/- 0.08	99.86 +/- 0.29	96.42 +/- 4.56
<i>KCNE5</i>	40.78 +/- 15.82	99.36 +/- 3.42	90.7 +/- 18.92	66.37 +/- 39.49
<i>KCNH2</i>	47.81 +/- 8.0	99.91 +/- 0.41	98.87 +/- 2.77	92.83 +/- 9.95
<i>KCNJ2</i>	54.68 +/- 7.95	100.0 +/- 0.0	99.98 +/- 0.16	99.36 +/- 1.9
<i>KCNJ5</i>	52.59 +/- 8.61	99.99 +/- 0.04	99.81 +/- 0.34	98.42 +/- 4.17

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>KCNJ8</i>	55.47 +/- 8.43	100.0 +/- 0.0	100.0 +/- 0.0	99.62 +/- 1.18
<i>KCNQ1</i>	51.39 +/- 9.07	100.0 +/- 0.0	99.69 +/- 1.36	96.43 +/- 6.72
<i>KLF10</i>	53.78 +/- 7.61	99.93 +/- 0.1	99.85 +/- 0.38	98.63 +/- 3.54
<i>KLHL24</i>	55.55 +/- 8.01	100.0 +/- 0.0	100.0 +/- 0.0	99.44 +/- 1.27
<i>KRAS</i>	55.79 +/- 7.63	99.98 +/- 0.04	99.92 +/- 0.22	99.08 +/- 2.05
<i>LAMA2</i>	54.96 +/- 7.47	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 1.56
<i>LAMA4</i>	55.18 +/- 7.59	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.58
<i>LAMP2</i>	43.14 +/- 15.76	99.97 +/- 0.17	95.17 +/- 8.15	71.26 +/- 34.03
<i>LDB3</i>	52.07 +/- 7.84	100.0 +/- 0.0	99.79 +/- 0.52	97.7 +/- 3.31
<i>LMNA</i>	49.82 +/- 8.35	100.0 +/- 0.0	99.96 +/- 0.16	97.29 +/- 6.96
<i>LMOD2</i>	50.2 +/- 7.93	99.99 +/- 0.06	99.6 +/- 0.89	96.22 +/- 5.42
<i>LRPPRC</i>	55.15 +/- 7.76	100.0 +/- 0.0	99.98 +/- 0.12	99.25 +/- 1.49
<i>LZTR1</i>	51.86 +/- 8.39	100.0 +/- 0.0	99.95 +/- 0.34	98.54 +/- 6.12
<i>MAP2K1</i>	51.3 +/- 8.2	100.0 +/- 0.0	99.8 +/- 0.72	97.54 +/- 4.52
<i>MAP2K2</i>	50.63 +/- 8.38	100.0 +/- 0.0	99.73 +/- 1.15	95.96 +/- 8.4
<i>MIB1</i>	54.44 +/- 7.41	100.0 +/- 0.03	99.89 +/- 0.42	98.92 +/- 2.07
<i>MLYCD</i>	52.28 +/- 8.19	99.98 +/- 0.1	99.7 +/- 0.56	96.63 +/- 5.15
<i>MMACHC</i>	49.18 +/- 8.8	100.0 +/- 0.0	99.81 +/- 0.88	96.27 +/- 8.17
<i>MMUT</i>	55.42 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.67 +/- 1.04
<i>MRAS</i>	52.12 +/- 7.74	100.0 +/- 0.0	99.84 +/- 0.85	98.34 +/- 4.13
<i>MRPL3</i>	54.68 +/- 7.73	100.0 +/- 0.0	100.0 +/- 0.0	99.51 +/- 1.71
<i>MRPL44</i>	55.94 +/- 6.97	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.23
<i>MT-TI</i>	3658.9 +/- 771.18	100.0 +/- 0.0	100.0 +/- 0.0	100.0 +/- 0.0
<i>MT-TK</i>	3538.39 +/- 702.04	100.0 +/- 0.0	100.0 +/- 0.0	100.0 +/- 0.0
<i>MTO1</i>	54.49 +/- 7.52	99.95 +/- 0.05	99.92 +/- 0.18	99.18 +/- 1.88
<i>MYBPC3</i>	52.81 +/- 8.63	100.0 +/- 0.0	99.97 +/- 0.15	98.43 +/- 4.04
<i>MYBPHL</i>	51.88 +/- 9.13	100.0 +/- 0.0	99.96 +/- 0.25	98.49 +/- 7.62
<i>MYH6</i>	50.26 +/- 8.21	100.0 +/- 0.0	99.75 +/- 0.71	96.64 +/- 7.29
<i>MYH7</i>	51.0 +/- 8.09	100.0 +/- 0.0	100.0 +/- 0.0	98.14 +/- 5.3
<i>MYL2</i>	51.82 +/- 8.78	100.0 +/- 0.0	99.93 +/- 0.48	98.17 +/- 4.62
<i>MYL3</i>	49.83 +/- 8.78	100.0 +/- 0.0	99.99 +/- 0.09	97.32 +/- 9.92
<i>MYL4</i>	51.33 +/- 9.24	100.0 +/- 0.0	100.0 +/- 0.01	98.49 +/- 4.43
<i>MYLK2</i>	49.4 +/- 8.99	100.0 +/- 0.0	99.83 +/- 0.96	97.14 +/- 9.35
<i>MYLK3</i>	54.2 +/- 7.97	100.0 +/- 0.0	99.99 +/- 0.06	99.27 +/- 2.29
<i>MYOM1</i>	53.52 +/- 7.36	100.0 +/- 0.0	99.92 +/- 0.31	98.89 +/- 2.08
<i>MYOT</i>	55.2 +/- 7.54	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.52
<i>MYOZ2</i>	54.78 +/- 7.37	100.0 +/- 0.0	100.0 +/- 0.0	99.82 +/- 0.64
<i>MYPN</i>	53.44 +/- 7.82	100.0 +/- 0.0	99.99 +/- 0.04	99.42 +/- 2.08
<i>NAA15</i>	56.35 +/- 8.03	100.0 +/- 0.0	100.0 +/- 0.0	99.64 +/- 0.91
<i>NAGLU</i>	53.56 +/- 8.47	100.0 +/- 0.0	99.96 +/- 0.3	98.55 +/- 4.21
<i>NDUFA1</i>	45.51 +/- 17.89	100.0 +/- 0.0	95.43 +/- 10.56	72.42 +/- 34.32
<i>NDUFA10</i>	53.79 +/- 7.01	100.0 +/- 0.0	99.94 +/- 0.21	98.71 +/- 2.72
<i>NDUFA11</i>	52.56 +/- 8.82	100.0 +/- 0.0	100.0 +/- 0.0	98.73 +/- 4.65
<i>NDUFA2</i>	53.81 +/- 9.93	100.0 +/- 0.0	100.0 +/- 0.0	99.0 +/- 3.71
<i>NDUFA4</i>	54.65 +/- 7.29	100.0 +/- 0.0	100.0 +/- 0.0	99.81 +/- 0.59
<i>NDUFAF1</i>	53.61 +/- 8.71	100.0 +/- 0.0	100.0 +/- 0.0	98.94 +/- 3.36
<i>NDUFAF2</i>	57.01 +/- 8.31	100.0 +/- 0.0	100.0 +/- 0.0	99.31 +/- 2.6

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
NDUFAF3	49.77 +/- 8.61	100.0 +/- 0.0	99.98 +/- 0.16	97.77 +/- 7.86
NDUFAF4	54.64 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.0	99.15 +/- 1.98
NDUFAF5	55.53 +/- 7.82	99.89 +/- 0.17	99.84 +/- 0.19	99.13 +/- 1.58
NDUFB11	35.85 +/- 13.88	97.85 +/- 4.02	82.84 +/- 20.6	55.78 +/- 38.92
NDUFB3	55.93 +/- 9.08	100.0 +/- 0.0	99.99 +/- 0.09	99.63 +/- 1.83
NDUFB8	47.88 +/- 8.9	100.0 +/- 0.0	99.87 +/- 0.89	96.86 +/- 10.1
NDUFS1	54.35 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.02	99.09 +/- 2.2
NDUFS2	51.61 +/- 8.73	100.0 +/- 0.0	99.99 +/- 0.07	98.89 +/- 3.06
NDUFS3	53.23 +/- 8.99	100.0 +/- 0.0	100.0 +/- 0.0	99.37 +/- 2.91
NDUFS4	55.0 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.0	99.82 +/- 0.87
NDUFS6	56.37 +/- 9.2	100.0 +/- 0.0	100.0 +/- 0.0	98.99 +/- 2.5
NDUFS7	57.32 +/- 9.67	100.0 +/- 0.0	100.0 +/- 0.0	98.55 +/- 3.68
NDUFS8	53.59 +/- 9.05	100.0 +/- 0.0	100.0 +/- 0.0	99.0 +/- 3.0
NDUFV1	48.59 +/- 8.66	100.0 +/- 0.0	99.84 +/- 0.89	96.41 +/- 7.96
NDUFV2	55.3 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.0	99.49 +/- 1.42
NEBL	55.1 +/- 7.33	100.0 +/- 0.0	99.99 +/- 0.09	99.7 +/- 0.76
NEXN	53.32 +/- 8.04	100.0 +/- 0.0	99.92 +/- 0.4	99.0 +/- 2.25
NF1	54.62 +/- 7.56	100.0 +/- 0.0	99.92 +/- 0.24	98.77 +/- 1.81
NKX2-5	53.83 +/- 9.3	100.0 +/- 0.0	99.97 +/- 0.18	98.71 +/- 4.48
NONO	41.92 +/- 16.4	99.9 +/- 0.38	93.02 +/- 13.02	67.29 +/- 38.46
NOS1AP	51.68 +/- 8.1	99.97 +/- 0.04	99.93 +/- 0.19	98.16 +/- 4.47
NPPA	50.03 +/- 8.43	100.0 +/- 0.0	99.82 +/- 0.84	98.18 +/- 9.14
NRAP	53.11 +/- 7.88	100.0 +/- 0.0	99.99 +/- 0.04	99.11 +/- 2.86
NRAS	55.99 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 0.83
NUBPL	54.57 +/- 8.26	100.0 +/- 0.0	100.0 +/- 0.02	99.39 +/- 2.17
OBSCN	53.36 +/- 8.77	100.0 +/- 0.0	99.94 +/- 0.33	98.37 +/- 4.66
PCCA	54.77 +/- 7.57	100.0 +/- 0.0	99.99 +/- 0.04	98.73 +/- 2.9
PCCB	53.66 +/- 7.65	100.0 +/- 0.0	99.97 +/- 0.21	99.15 +/- 3.22
PDLIM3	53.99 +/- 7.92	100.0 +/- 0.0	100.0 +/- 0.0	99.28 +/- 1.51
PET100	49.16 +/- 9.24	100.0 +/- 0.0	100.0 +/- 0.0	97.5 +/- 10.89
PKP2	54.9 +/- 7.37	100.0 +/- 0.0	99.99 +/- 0.06	99.04 +/- 3.01
PLD1	54.99 +/- 7.95	100.0 +/- 0.01	100.0 +/- 0.02	99.57 +/- 1.02
PLEKHM2	49.8 +/- 8.12	100.0 +/- 0.01	98.88 +/- 2.45	94.06 +/- 7.48
PLN	54.41 +/- 7.27	100.0 +/- 0.0	100.0 +/- 0.0	99.58 +/- 1.05
PMM2	53.79 +/- 8.67	100.0 +/- 0.0	99.87 +/- 0.85	98.37 +/- 4.98
PNPLA2	51.63 +/- 9.27	99.9 +/- 0.1	99.69 +/- 1.28	96.55 +/- 8.33
PPA2	54.11 +/- 7.07	100.0 +/- 0.0	100.0 +/- 0.0	99.51 +/- 1.51
PPCS	55.63 +/- 8.62	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 1.53
PPP1CB	55.13 +/- 7.65	100.0 +/- 0.03	99.76 +/- 0.67	97.96 +/- 2.01
PPP1R13L	47.85 +/- 8.43	99.98 +/- 0.1	98.48 +/- 3.12	91.8 +/- 10.93
PRDM16	53.96 +/- 8.27	100.0 +/- 0.0	99.8 +/- 0.5	97.91 +/- 4.55
PRKAG2	54.31 +/- 7.74	100.0 +/- 0.0	99.95 +/- 0.22	98.33 +/- 3.12
PSEN1	54.65 +/- 8.13	100.0 +/- 0.0	99.98 +/- 0.13	99.51 +/- 1.56
PSEN2	51.42 +/- 7.81	100.0 +/- 0.0	99.97 +/- 0.14	98.55 +/- 4.25
PTPN11	55.1 +/- 7.94	100.0 +/- 0.0	99.96 +/- 0.18	99.28 +/- 1.99
QRSL1	54.02 +/- 7.14	99.99 +/- 0.03	99.99 +/- 0.03	99.38 +/- 1.41
RAF1	54.46 +/- 7.88	100.0 +/- 0.01	99.97 +/- 0.19	98.64 +/- 4.74

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>RANGRF</i>	49.05 +/- 8.96	100.0 +/- 0.0	100.0 +/- 0.0	97.89 +/- 7.18
<i>RASA2</i>	54.15 +/- 7.33	100.0 +/- 0.0	99.93 +/- 0.25	99.15 +/- 1.33
<i>RBM20</i>	51.96 +/- 7.99	100.0 +/- 0.0	99.97 +/- 0.1	98.38 +/- 4.4
<i>RHBDF1</i>	55.57 +/- 9.2	100.0 +/- 0.0	99.91 +/- 0.6	98.84 +/- 3.61
<i>RIT1</i>	53.38 +/- 7.23	100.0 +/- 0.0	99.97 +/- 0.09	99.0 +/- 2.2
<i>RNF220</i>	50.97 +/- 9.18	100.0 +/- 0.0	99.91 +/- 0.38	98.22 +/- 4.89
<i>RPL3L</i>	53.07 +/- 7.83	100.0 +/- 0.0	99.95 +/- 0.31	98.54 +/- 5.62
<i>RRAGD</i>	53.32 +/- 7.55	99.99 +/- 0.09	99.64 +/- 1.25	97.93 +/- 2.9
<i>RRAS</i>	46.2 +/- 7.88	100.0 +/- 0.0	99.3 +/- 2.07	93.15 +/- 12.2
<i>RYR2</i>	54.31 +/- 7.57	100.0 +/- 0.0	99.93 +/- 0.23	98.87 +/- 2.23
<i>SCN10A</i>	52.47 +/- 8.11	100.0 +/- 0.0	100.0 +/- 0.0	99.17 +/- 2.12
<i>SCN1B</i>	45.33 +/- 7.64	99.9 +/- 0.64	98.74 +/- 4.7	90.01 +/- 13.8
<i>SCN2B</i>	51.46 +/- 8.26	100.0 +/- 0.01	99.94 +/- 0.19	98.13 +/- 5.62
<i>SCN3B</i>	51.74 +/- 8.08	100.0 +/- 0.0	99.93 +/- 0.35	98.66 +/- 3.7
<i>SCN4B</i>	48.74 +/- 8.52	100.0 +/- 0.0	99.81 +/- 1.13	96.08 +/- 8.49
<i>SCN5A</i>	51.21 +/- 8.03	100.0 +/- 0.0	99.95 +/- 0.17	98.18 +/- 3.72
<i>SCO1</i>	54.33 +/- 7.15	99.99 +/- 0.03	99.92 +/- 0.15	98.99 +/- 1.61
<i>SCO2</i>	51.71 +/- 8.16	100.0 +/- 0.0	99.99 +/- 0.07	98.51 +/- 4.1
<i>SDHA</i>	54.85 +/- 7.73	100.0 +/- 0.0	99.96 +/- 0.2	99.11 +/- 2.34
<i>SDHAF1</i>	53.24 +/- 9.14	100.0 +/- 0.0	99.88 +/- 0.83	98.53 +/- 4.56
<i>SDHD</i>	55.56 +/- 8.26	100.0 +/- 0.0	100.0 +/- 0.0	99.68 +/- 1.43
<i>SGCB</i>	56.04 +/- 7.84	100.0 +/- 0.0	99.9 +/- 0.35	99.14 +/- 1.34
<i>SGCD</i>	53.75 +/- 7.31	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.14
<i>SGCG</i>	55.97 +/- 7.92	100.0 +/- 0.0	100.0 +/- 0.0	99.58 +/- 1.69
<i>SGSH</i>	52.42 +/- 8.24	100.0 +/- 0.0	99.97 +/- 0.21	98.45 +/- 4.85
<i>SHMT2</i>	52.64 +/- 8.92	100.0 +/- 0.0	99.99 +/- 0.07	98.84 +/- 3.39
<i>SHOC2</i>	54.91 +/- 7.5	100.0 +/- 0.0	99.92 +/- 0.29	99.21 +/- 0.87
<i>SLC22A5</i>	52.51 +/- 8.16	100.0 +/- 0.0	99.96 +/- 0.16	98.43 +/- 4.25
<i>SLC25A20</i>	53.26 +/- 8.15	100.0 +/- 0.0	100.0 +/- 0.0	99.27 +/- 2.76
<i>SLC25A3</i>	53.74 +/- 7.99	100.0 +/- 0.0	99.99 +/- 0.05	99.1 +/- 2.83
<i>SLC25A4</i>	52.35 +/- 7.87	100.0 +/- 0.0	99.98 +/- 0.16	98.33 +/- 4.02
<i>SLC30A5</i>	53.87 +/- 7.09	100.0 +/- 0.0	99.8 +/- 0.96	98.96 +/- 2.67
<i>SLC40A1</i>	55.65 +/- 8.05	100.0 +/- 0.0	99.99 +/- 0.07	99.57 +/- 1.31
<i>SLC4A3</i>	48.87 +/- 8.45	100.0 +/- 0.0	99.73 +/- 0.82	96.28 +/- 6.56
<i>SLC6A6</i>	51.33 +/- 7.94	100.0 +/- 0.0	99.88 +/- 0.73	98.01 +/- 4.59
<i>SLMAP</i>	54.75 +/- 7.75	100.0 +/- 0.0	100.0 +/- 0.03	98.97 +/- 2.56
<i>SNTA1</i>	47.99 +/- 7.59	100.0 +/- 0.0	99.64 +/- 1.31	95.95 +/- 8.77
<i>SOS1</i>	53.84 +/- 7.54	100.0 +/- 0.0	99.69 +/- 0.98	97.96 +/- 3.04
<i>SOS2</i>	54.73 +/- 7.38	99.99 +/- 0.04	99.78 +/- 0.76	98.52 +/- 2.17
<i>SPEG</i>	50.43 +/- 8.78	100.0 +/- 0.0	99.81 +/- 0.86	96.85 +/- 8.55
<i>SPRED1</i>	54.98 +/- 7.33	100.0 +/- 0.0	99.95 +/- 0.23	99.04 +/- 1.38
<i>SPRED2</i>	53.17 +/- 7.71	99.99 +/- 0.09	99.81 +/- 0.74	97.61 +/- 4.14
<i>SURF1</i>	51.24 +/- 8.42	100.0 +/- 0.0	99.78 +/- 1.21	97.56 +/- 4.81
<i>TAB2</i>	53.46 +/- 7.72	99.91 +/- 0.03	99.7 +/- 0.85	97.77 +/- 2.65
<i>TAFAZZIN</i>	39.63 +/- 15.85	99.51 +/- 2.23	88.68 +/- 18.75	63.59 +/- 41.35
<i>TANGO2</i>	53.93 +/- 8.6	100.0 +/- 0.0	100.0 +/- 0.0	98.97 +/- 3.83

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>TBX20</i>	50.84 +/- 7.71	100.0 +/- 0.0	99.97 +/- 0.1	98.21 +/- 6.83
<i>TBX5</i>	52.02 +/- 7.59	100.0 +/- 0.0	99.97 +/- 0.21	98.92 +/- 2.83
<i>TCAP</i>	49.93 +/- 8.98	100.0 +/- 0.0	99.88 +/- 0.82	97.24 +/- 6.84
<i>TECRL</i>	57.49 +/- 7.82	99.98 +/- 0.04	99.98 +/- 0.04	99.61 +/- 1.32
<i>TFR2</i>	48.57 +/- 7.98	100.0 +/- 0.0	99.76 +/- 1.15	95.88 +/- 9.14
<i>TGFB3</i>	51.27 +/- 8.32	100.0 +/- 0.0	99.99 +/- 0.05	98.79 +/- 4.24
<i>TMEM126B</i>	56.97 +/- 8.13	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.59
<i>TMEM43</i>	53.09 +/- 7.39	100.0 +/- 0.0	99.96 +/- 0.28	98.86 +/- 4.44
<i>TMEM70</i>	55.07 +/- 8.4	100.0 +/- 0.0	99.98 +/- 0.1	98.99 +/- 2.65
<i>TMPO</i>	55.24 +/- 7.54	100.0 +/- 0.0	99.92 +/- 0.54	99.12 +/- 3.01
<i>TNNC1</i>	47.47 +/- 9.49	100.0 +/- 0.0	99.86 +/- 0.91	95.73 +/- 13.22
<i>TNNI3</i>	47.53 +/- 8.64	100.0 +/- 0.0	99.48 +/- 3.4	95.8 +/- 11.48
<i>TNNI3K</i>	56.41 +/- 7.57	100.0 +/- 0.0	100.0 +/- 0.0	99.78 +/- 0.71
<i>TNNT2</i>	48.97 +/- 8.98	100.0 +/- 0.0	99.89 +/- 0.53	97.36 +/- 8.88
<i>TOR1AIP1</i>	53.34 +/- 7.84	100.0 +/- 0.0	100.0 +/- 0.02	98.68 +/- 3.06
<i>TPM1</i>	52.87 +/- 7.59	100.0 +/- 0.0	99.79 +/- 1.4	98.36 +/- 5.52
<i>TRDN</i>	55.85 +/- 7.38	100.0 +/- 0.0	100.0 +/- 0.03	99.67 +/- 0.73
<i>TRIM63</i>	51.0 +/- 9.3	100.0 +/- 0.0	99.9 +/- 0.6	98.31 +/- 3.63
<i>TRPM4</i>	50.11 +/- 8.25	100.0 +/- 0.0	99.84 +/- 0.65	97.12 +/- 5.92
<i>TSFM</i>	54.37 +/- 8.59	100.0 +/- 0.0	100.0 +/- 0.0	99.25 +/- 1.86
<i>TTN</i>	54.55 +/- 7.64	99.91 +/- 0.1	99.5 +/- 0.22	98.62 +/- 1.32
<i>TTR</i>	55.5 +/- 8.45	100.0 +/- 0.0	100.0 +/- 0.0	99.52 +/- 1.77
<i>TXNRD2</i>	52.07 +/- 8.53	100.0 +/- 0.0	100.0 +/- 0.01	98.57 +/- 4.31
<i>UQCC2</i>	55.03 +/- 9.29	100.0 +/- 0.0	99.94 +/- 0.43	98.79 +/- 5.82
<i>VCL</i>	52.06 +/- 7.88	100.0 +/- 0.0	99.99 +/- 0.09	98.68 +/- 3.15
<i>XK</i>	43.27 +/- 16.22	99.96 +/- 0.25	94.56 +/- 9.0	70.3 +/- 34.89
<i>ZBTB17</i>	56.27 +/- 8.95	100.0 +/- 0.0	100.0 +/- 0.03	99.21 +/- 2.9

For supplerende oplysninger vedrørende analysen kan afdelingen kontaktes på mail: mol-dia@rn.dk. Rapport genereret: 23/01-2026