

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: 278

version: 1.3

Ibrugtagningsdato: 2023-05-22

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 de anførte gener undersøges der for varianter i de proteinkodende regioner og de nærmeste 10 bp i de tilstødende ikke-proteinkodende regioner, 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
ANOS	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
BRAF	53.15 +/- 6.81	99.99 +/- 0.05	99.51 +/- 1.19	97.12 +/- 2.41
CACNA1C	51.85 +/- 7.82	100.0 +/- 0.0	99.82 +/- 0.82	98.29 +/- 3.77
CACNA1D	53.11 +/- 8.01	99.99 +/- 0.03	99.86 +/- 0.41	98.66 +/- 3.17

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<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
<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

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<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
<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

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<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
<i>NDUFAF3</i>	49.77 +/- 8.61	100.0 +/- 0.0	99.98 +/- 0.16	97.77 +/- 7.86
<i>NDUFAF4</i>	54.64 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.0	99.15 +/- 1.98

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>NDUFAF5</i>	55.53 +/- 7.82	99.89 +/- 0.17	99.84 +/- 0.19	99.13 +/- 1.58
<i>NDUFB11</i>	35.85 +/- 13.88	97.85 +/- 4.02	82.84 +/- 20.6	55.78 +/- 38.92
<i>NDUFB3</i>	55.93 +/- 9.08	100.0 +/- 0.0	99.99 +/- 0.09	99.63 +/- 1.83
<i>NDUFB8</i>	47.88 +/- 8.9	100.0 +/- 0.0	99.87 +/- 0.89	96.86 +/- 10.1
<i>NDUFS1</i>	54.35 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.02	99.09 +/- 2.2
<i>NDUFS2</i>	51.61 +/- 8.73	100.0 +/- 0.0	99.99 +/- 0.07	98.89 +/- 3.06
<i>NDUFS3</i>	53.23 +/- 8.99	100.0 +/- 0.0	100.0 +/- 0.0	99.37 +/- 2.91
<i>NDUFS4</i>	55.0 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.0	99.82 +/- 0.87
<i>NDUFS6</i>	56.37 +/- 9.2	100.0 +/- 0.0	100.0 +/- 0.0	98.99 +/- 2.5
<i>NDUFS7</i>	57.32 +/- 9.67	100.0 +/- 0.0	100.0 +/- 0.0	98.55 +/- 3.68
<i>NDUFS8</i>	53.59 +/- 9.05	100.0 +/- 0.0	100.0 +/- 0.0	99.0 +/- 3.0
<i>NDUFV1</i>	48.59 +/- 8.66	100.0 +/- 0.0	99.84 +/- 0.89	96.41 +/- 7.96
<i>NDUFV2</i>	55.3 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.0	99.49 +/- 1.42
<i>NEBL</i>	55.1 +/- 7.33	100.0 +/- 0.0	99.99 +/- 0.09	99.7 +/- 0.76
<i>NEXN</i>	53.32 +/- 8.04	100.0 +/- 0.0	99.92 +/- 0.4	99.0 +/- 2.25
<i>NF1</i>	54.62 +/- 7.56	100.0 +/- 0.0	99.92 +/- 0.24	98.77 +/- 1.81
<i>NKX2-5</i>	53.83 +/- 9.3	100.0 +/- 0.0	99.97 +/- 0.18	98.71 +/- 4.48
<i>NONO</i>	41.92 +/- 16.4	99.9 +/- 0.38	93.02 +/- 13.02	67.29 +/- 38.46
<i>NOS1AP</i>	51.68 +/- 8.1	99.97 +/- 0.04	99.93 +/- 0.19	98.16 +/- 4.47
<i>NPPA</i>	50.03 +/- 8.43	100.0 +/- 0.0	99.82 +/- 0.84	98.18 +/- 9.14
<i>NRAP</i>	53.11 +/- 7.88	100.0 +/- 0.0	99.99 +/- 0.04	99.11 +/- 2.86
<i>NRAS</i>	55.99 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 0.83
<i>NUBPL</i>	54.57 +/- 8.26	100.0 +/- 0.0	100.0 +/- 0.02	99.39 +/- 2.17
<i>OBSCN</i>	53.36 +/- 8.77	100.0 +/- 0.0	99.94 +/- 0.33	98.37 +/- 4.66
<i>PCCA</i>	54.77 +/- 7.57	100.0 +/- 0.0	99.99 +/- 0.04	98.73 +/- 2.9
<i>PCCB</i>	53.66 +/- 7.65	100.0 +/- 0.0	99.97 +/- 0.21	99.15 +/- 3.22
<i>PDLIM3</i>	53.99 +/- 7.92	100.0 +/- 0.0	100.0 +/- 0.0	99.28 +/- 1.51
<i>PET100</i>	49.16 +/- 9.24	100.0 +/- 0.0	100.0 +/- 0.0	97.5 +/- 10.89
<i>PKP2</i>	54.9 +/- 7.37	100.0 +/- 0.0	99.99 +/- 0.06	99.04 +/- 3.01
<i>PLD1</i>	54.99 +/- 7.95	100.0 +/- 0.01	100.0 +/- 0.02	99.57 +/- 1.02
<i>PLEKHM2</i>	49.8 +/- 8.12	100.0 +/- 0.01	98.88 +/- 2.45	94.06 +/- 7.48
<i>PLN</i>	54.41 +/- 7.27	100.0 +/- 0.0	100.0 +/- 0.0	99.58 +/- 1.05
<i>PMM2</i>	53.79 +/- 8.67	100.0 +/- 0.0	99.87 +/- 0.85	98.37 +/- 4.98
<i>PNPLA2</i>	51.63 +/- 9.27	99.9 +/- 0.1	99.69 +/- 1.28	96.55 +/- 8.33
<i>PPA2</i>	54.11 +/- 7.07	100.0 +/- 0.0	100.0 +/- 0.0	99.51 +/- 1.51
<i>PPCS</i>	55.63 +/- 8.62	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 1.53
<i>PPP1CB</i>	55.13 +/- 7.65	100.0 +/- 0.03	99.76 +/- 0.67	97.96 +/- 2.01
<i>PPP1R13L</i>	47.85 +/- 8.43	99.98 +/- 0.1	98.48 +/- 3.12	91.8 +/- 10.93
<i>PRDM16</i>	53.96 +/- 8.27	100.0 +/- 0.0	99.8 +/- 0.5	97.91 +/- 4.55
<i>PRKAG2</i>	54.31 +/- 7.74	100.0 +/- 0.0	99.95 +/- 0.22	98.33 +/- 3.12
<i>PSEN1</i>	54.65 +/- 8.13	100.0 +/- 0.0	99.98 +/- 0.13	99.51 +/- 1.56
<i>PSEN2</i>	51.42 +/- 7.81	100.0 +/- 0.0	99.97 +/- 0.14	98.55 +/- 4.25
<i>PTPN11</i>	55.1 +/- 7.94	100.0 +/- 0.0	99.96 +/- 0.18	99.28 +/- 1.99
<i>QRSL1</i>	54.02 +/- 7.14	99.99 +/- 0.03	99.99 +/- 0.03	99.38 +/- 1.41
<i>RAF1</i>	54.46 +/- 7.88	100.0 +/- 0.01	99.97 +/- 0.19	98.64 +/- 4.74
<i>RANGRF</i>	49.05 +/- 8.96	100.0 +/- 0.0	100.0 +/- 0.0	97.89 +/- 7.18

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<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
<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

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
TCAP	49.93 +/- 8.98	100.0 +/- 0.0	99.88 +/- 0.82	97.24 +/- 6.84
TECRL	57.49 +/- 7.82	99.98 +/- 0.04	99.98 +/- 0.04	99.61 +/- 1.32
TFR2	48.57 +/- 7.98	100.0 +/- 0.0	99.76 +/- 1.15	95.88 +/- 9.14
TGFB3	51.27 +/- 8.32	100.0 +/- 0.0	99.99 +/- 0.05	98.79 +/- 4.24
TMEM126B	56.97 +/- 8.13	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.59
TMEM43	53.09 +/- 7.39	100.0 +/- 0.0	99.96 +/- 0.28	98.86 +/- 4.44
TMEM70	55.07 +/- 8.4	100.0 +/- 0.0	99.98 +/- 0.1	98.99 +/- 2.65
TMPO	55.24 +/- 7.54	100.0 +/- 0.0	99.92 +/- 0.54	99.12 +/- 3.01
TNNC1	47.47 +/- 9.49	100.0 +/- 0.0	99.86 +/- 0.91	95.73 +/- 13.22
TNNI3	47.53 +/- 8.64	100.0 +/- 0.0	99.48 +/- 3.4	95.8 +/- 11.48
TNNI3K	56.41 +/- 7.57	100.0 +/- 0.0	100.0 +/- 0.0	99.78 +/- 0.71
TNNT2	48.97 +/- 8.98	100.0 +/- 0.0	99.89 +/- 0.53	97.36 +/- 8.88
TOR1AIP1	53.34 +/- 7.84	100.0 +/- 0.0	100.0 +/- 0.02	98.68 +/- 3.06
TPM1	52.87 +/- 7.59	100.0 +/- 0.0	99.79 +/- 1.4	98.36 +/- 5.52
TRDN	55.85 +/- 7.38	100.0 +/- 0.0	100.0 +/- 0.03	99.67 +/- 0.73
TRIM63	51.0 +/- 9.3	100.0 +/- 0.0	99.9 +/- 0.6	98.31 +/- 3.63
TRPM4	50.11 +/- 8.25	100.0 +/- 0.0	99.84 +/- 0.65	97.12 +/- 5.92
TSFM	54.37 +/- 8.59	100.0 +/- 0.0	100.0 +/- 0.0	99.25 +/- 1.86
TTN	54.55 +/- 7.64	99.91 +/- 0.1	99.5 +/- 0.22	98.62 +/- 1.32
TTR	55.5 +/- 8.45	100.0 +/- 0.0	100.0 +/- 0.0	99.52 +/- 1.77
TXNRD2	52.07 +/- 8.53	100.0 +/- 0.0	100.0 +/- 0.01	98.57 +/- 4.31
UQCC2	55.03 +/- 9.29	100.0 +/- 0.0	99.94 +/- 0.43	98.79 +/- 5.82
VCL	52.06 +/- 7.88	100.0 +/- 0.0	99.99 +/- 0.09	98.68 +/- 3.15
XK	43.27 +/- 16.22	99.96 +/- 0.25	94.56 +/- 9.0	70.3 +/- 34.89
ZBTB17	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: 09/10-2025