

Analyse af gener relevante ved retinal dystrofi

Panelbeskrivelse

Navn: Analyse af gener relevante ved retinal dystrofi

Antal gener: 302

version: 2.0

Ibrugtagningsdato: 2025-06-10

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 [%]
ABCA4	52.6 +/- 7.91	100.0 +/- 0.0	99.94 +/- 0.32	98.75 +/- 4.1
ABCC6	50.08 +/- 7.67	100.0 +/- 0.0	99.85 +/- 0.9	97.68 +/- 5.84
ABHD12	50.57 +/- 8.62	100.0 +/- 0.01	99.74 +/- 0.8	96.96 +/- 6.38
ACBD5	55.14 +/- 7.48	99.95 +/- 0.05	99.94 +/- 0.05	99.26 +/- 1.38
ACO2	52.76 +/- 9.0	100.0 +/- 0.0	99.97 +/- 0.21	98.21 +/- 6.91
ADAM9	55.45 +/- 7.32	100.0 +/- 0.0	99.96 +/- 0.3	99.26 +/- 1.39
ADAMTS18	53.32 +/- 7.7	100.0 +/- 0.0	99.95 +/- 0.33	98.82 +/- 3.85
ADGRV1	54.8 +/- 7.41	100.0 +/- 0.0	99.99 +/- 0.06	99.49 +/- 1.01
ADIPOR1	52.08 +/- 7.57	100.0 +/- 0.0	99.99 +/- 0.09	98.22 +/- 3.82
AFG3L2	54.26 +/- 7.85	100.0 +/- 0.0	99.96 +/- 0.25	98.97 +/- 2.49
AGBL5	52.49 +/- 8.46	100.0 +/- 0.0	99.97 +/- 0.18	98.72 +/- 4.27
AHI1	55.19 +/- 7.32	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 0.81
AHR	53.52 +/- 7.74	100.0 +/- 0.0	99.82 +/- 0.66	97.82 +/- 3.03
AIPL1	54.48 +/- 8.32	99.96 +/- 0.05	99.95 +/- 0.05	99.04 +/- 3.5
AIRE	50.74 +/- 8.1	100.0 +/- 0.0	99.96 +/- 0.3	97.68 +/- 6.97
ALDH3A2	52.74 +/- 7.61	100.0 +/- 0.02	99.99 +/- 0.03	98.94 +/- 1.83
ALMS1	53.24 +/- 7.52	100.0 +/- 0.0	99.98 +/- 0.06	99.18 +/- 1.66
ALPK1	54.25 +/- 8.24	100.0 +/- 0.0	100.0 +/- 0.0	99.19 +/- 1.76
AMACR	54.74 +/- 7.59	100.0 +/- 0.0	99.91 +/- 0.38	99.09 +/- 1.99
ARRHGEF18	52.91 +/- 8.21	100.0 +/- 0.0	99.96 +/- 0.19	98.38 +/- 4.49
ARL13B	55.42 +/- 6.97	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 0.82

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
ARL2BP	54.68 +/- 8.22	100.0 +/- 0.0	99.97 +/- 0.18	99.34 +/- 1.65
ARL3	51.14 +/- 7.99	100.0 +/- 0.0	99.89 +/- 0.29	98.1 +/- 3.82
ARL6	56.43 +/- 7.8	100.0 +/- 0.0	100.0 +/- 0.01	99.46 +/- 1.64
ARSG	52.08 +/- 8.05	100.0 +/- 0.0	99.87 +/- 0.6	98.46 +/- 3.84
ASRGL1	53.65 +/- 8.4	99.88 +/- 0.15	99.87 +/- 0.15	98.81 +/- 3.81
ATF6	54.55 +/- 7.39	100.0 +/- 0.02	99.95 +/- 0.05	99.38 +/- 1.46
ATOH7	56.3 +/- 8.98	100.0 +/- 0.0	99.95 +/- 0.33	98.88 +/- 4.06
ATXN7	50.36 +/- 7.01	99.85 +/- 0.18	97.84 +/- 1.94	92.92 +/- 4.38
BBIP1	55.31 +/- 8.01	100.0 +/- 0.0	100.0 +/- 0.03	99.44 +/- 2.48
BBS1	52.34 +/- 8.31	100.0 +/- 0.0	100.0 +/- 0.03	98.98 +/- 3.48
BBS10	55.8 +/- 7.93	100.0 +/- 0.0	99.96 +/- 0.27	99.53 +/- 1.25
BBS12	56.62 +/- 8.74	100.0 +/- 0.0	99.99 +/- 0.07	99.45 +/- 1.89
BBS2	54.23 +/- 7.64	100.0 +/- 0.0	100.0 +/- 0.0	99.72 +/- 0.61
BBS4	53.66 +/- 8.02	100.0 +/- 0.0	99.99 +/- 0.09	99.52 +/- 1.37
BBS5	52.67 +/- 7.75	99.98 +/- 0.12	99.54 +/- 1.04	96.14 +/- 5.26
BBS7	56.19 +/- 7.17	100.0 +/- 0.0	99.99 +/- 0.06	99.64 +/- 0.95
BBS9	54.64 +/- 7.47	100.0 +/- 0.0	99.99 +/- 0.03	99.15 +/- 1.85
BEST1	50.94 +/- 8.34	100.0 +/- 0.0	99.96 +/- 0.2	98.39 +/- 4.93
C1QTNF5	48.61 +/- 8.26	100.0 +/- 0.0	99.85 +/- 0.66	96.2 +/- 8.75
CABP4	54.68 +/- 8.49	100.0 +/- 0.0	99.93 +/- 0.48	98.97 +/- 5.23
CACNA1F	38.22 +/- 15.33	99.65 +/- 1.14	87.16 +/- 18.94	61.64 +/- 41.54
CACNA2D4	51.07 +/- 8.02	100.0 +/- 0.0	99.98 +/- 0.13	98.33 +/- 4.63
CAPN5	51.83 +/- 8.41	100.0 +/- 0.0	99.97 +/- 0.22	98.64 +/- 3.81
CC2D2A	54.85 +/- 8.01	100.0 +/- 0.0	99.99 +/- 0.09	99.36 +/- 2.46
CCT2	55.38 +/- 7.31	100.0 +/- 0.0	99.94 +/- 0.37	99.39 +/- 1.49
CDH23	51.33 +/- 8.46	100.0 +/- 0.0	99.9 +/- 0.35	97.81 +/- 5.4
CDH3	51.59 +/- 8.43	100.0 +/- 0.0	99.9 +/- 0.66	98.47 +/- 4.73
CDHR1	52.68 +/- 8.48	100.0 +/- 0.0	99.97 +/- 0.17	98.94 +/- 2.78
CEP164	49.98 +/- 8.18	100.0 +/- 0.0	99.82 +/- 0.5	96.83 +/- 6.45
CEP19	52.74 +/- 8.12	99.98 +/- 0.04	99.95 +/- 0.11	98.88 +/- 2.94
CEP250	52.33 +/- 8.0	100.0 +/- 0.0	99.95 +/- 0.34	98.65 +/- 4.62
CEP290	54.71 +/- 7.35	100.0 +/- 0.0	100.0 +/- 0.01	99.48 +/- 1.25
CEP78	55.8 +/- 7.38	100.0 +/- 0.0	99.98 +/- 0.1	99.49 +/- 1.18
CERKL	55.16 +/- 7.11	100.0 +/- 0.0	100.0 +/- 0.0	99.4 +/- 1.38
CFAP410	53.88 +/- 8.44	100.0 +/- 0.0	99.91 +/- 0.5	98.16 +/- 3.71
CFAP418	56.07 +/- 8.09	100.0 +/- 0.0	100.0 +/- 0.0	99.64 +/- 1.22
CFH	58.11 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.0	99.82 +/- 0.52
CHM	43.26 +/- 15.79	99.98 +/- 0.08	95.26 +/- 7.71	70.91 +/- 34.37
CIB2	49.14 +/- 8.82	99.9 +/- 0.0	99.68 +/- 1.13	96.08 +/- 8.18
CLCC1	54.56 +/- 7.39	100.0 +/- 0.0	100.0 +/- 0.0	99.37 +/- 1.41
CLN3	49.41 +/- 8.56	100.0 +/- 0.0	99.93 +/- 0.41	97.49 +/- 6.6
CLN5	55.95 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.0	99.7 +/- 0.68
CLN6	48.22 +/- 8.66	99.97 +/- 0.07	99.81 +/- 0.66	95.66 +/- 8.17
CLN8	51.36 +/- 7.62	98.57 +/- 0.67	97.01 +/- 1.09	94.5 +/- 3.81
CLRN1	54.46 +/- 7.43	100.0 +/- 0.0	100.0 +/- 0.0	99.64 +/- 1.37
CLUAP1	53.47 +/- 8.04	100.0 +/- 0.0	100.0 +/- 0.03	99.3 +/- 2.16

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>CNGA1</i>	53.48 +/- 7.42	100.0 +/- 0.0	99.96 +/- 0.24	99.34 +/- 1.21
<i>CNGA3</i>	52.44 +/- 8.89	100.0 +/- 0.0	99.95 +/- 0.33	98.83 +/- 3.49
<i>CNGB1</i>	50.8 +/- 7.96	99.98 +/- 0.04	99.88 +/- 0.59	97.88 +/- 4.89
<i>CNGB3</i>	54.27 +/- 7.71	100.0 +/- 0.0	99.99 +/- 0.06	99.45 +/- 1.8
<i>CNNM4</i>	50.18 +/- 8.43	100.0 +/- 0.0	99.77 +/- 1.01	96.95 +/- 9.02
<i>COL18A1</i>	53.15 +/- 9.25	99.96 +/- 0.08	99.63 +/- 0.71	97.24 +/- 5.86
<i>COL4A1</i>	53.49 +/- 7.61	100.0 +/- 0.0	99.99 +/- 0.04	98.83 +/- 3.9
<i>COQ2</i>	55.89 +/- 7.92	100.0 +/- 0.0	100.0 +/- 0.0	99.01 +/- 3.42
<i>COQ5</i>	52.16 +/- 8.46	100.0 +/- 0.0	100.0 +/- 0.01	99.11 +/- 2.67
<i>CRB1</i>	54.71 +/- 7.34	100.0 +/- 0.0	100.0 +/- 0.0	99.47 +/- 1.33
<i>CRX</i>	50.0 +/- 8.2	99.98 +/- 0.04	99.91 +/- 0.45	97.79 +/- 5.12
<i>CSPP1</i>	54.76 +/- 7.34	100.0 +/- 0.0	100.0 +/- 0.0	99.43 +/- 0.86
<i>CTC1</i>	50.95 +/- 7.99	100.0 +/- 0.0	99.94 +/- 0.24	98.52 +/- 4.55
<i>CTNNA1</i>	54.32 +/- 7.63	100.0 +/- 0.01	99.96 +/- 0.22	99.33 +/- 2.67
<i>CTNNB1</i>	55.04 +/- 7.68	100.0 +/- 0.0	100.0 +/- 0.03	99.09 +/- 1.44
<i>CTSD</i>	51.59 +/- 9.55	100.0 +/- 0.0	99.88 +/- 0.69	97.16 +/- 7.32
<i>CWC27</i>	53.67 +/- 7.14	100.0 +/- 0.0	99.9 +/- 0.47	98.86 +/- 2.56
<i>CYP2R1</i>	54.26 +/- 7.44	100.0 +/- 0.0	100.0 +/- 0.03	99.37 +/- 1.94
<i>CYP4V2</i>	54.14 +/- 7.94	100.0 +/- 0.0	99.96 +/- 0.25	98.98 +/- 2.92
<i>DHDDS</i>	51.93 +/- 8.36	100.0 +/- 0.0	100.0 +/- 0.0	98.46 +/- 4.24
<i>DHX38</i>	52.66 +/- 8.19	100.0 +/- 0.0	99.98 +/- 0.12	98.9 +/- 4.16
<i>DMD</i>	43.26 +/- 15.85	99.96 +/- 0.14	94.99 +/- 8.6	71.25 +/- 33.8
<i>DRAM2</i>	55.02 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 1.46
<i>EFEMP1</i>	55.23 +/- 8.02	100.0 +/- 0.0	99.94 +/- 0.29	99.04 +/- 2.21
<i>ELOVL1</i>	50.06 +/- 9.24	100.0 +/- 0.0	99.97 +/- 0.18	98.24 +/- 4.37
<i>ELOVL4</i>	54.94 +/- 7.66	100.0 +/- 0.0	99.82 +/- 1.01	98.68 +/- 2.86
<i>ERCC6</i>	54.53 +/- 7.25	100.0 +/- 0.0	99.99 +/- 0.06	99.41 +/- 1.57
<i>ERCC8</i>	54.54 +/- 7.59	100.0 +/- 0.0	100.0 +/- 0.03	99.42 +/- 1.25
<i>ESPN</i>	47.81 +/- 8.29	99.99 +/- 0.05	99.26 +/- 1.81	92.86 +/- 11.26
<i>EXOSC2</i>	51.86 +/- 8.31	100.0 +/- 0.0	99.98 +/- 0.12	98.82 +/- 4.51
<i>EYS</i>	56.44 +/- 7.49	100.0 +/- 0.0	100.0 +/- 0.0	99.58 +/- 0.99
<i>FAM161A</i>	54.03 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.01	99.27 +/- 2.32
<i>FLVCR1</i>	54.95 +/- 7.43	99.83 +/- 0.23	99.66 +/- 0.39	98.6 +/- 2.83
<i>FRMD7</i>	42.14 +/- 16.08	99.99 +/- 0.07	94.18 +/- 9.36	68.02 +/- 37.95
<i>FZD4</i>	52.86 +/- 7.97	99.98 +/- 0.04	99.91 +/- 0.16	98.5 +/- 3.35
<i>GDF6</i>	51.23 +/- 7.45	100.0 +/- 0.0	99.91 +/- 0.29	97.75 +/- 5.85
<i>GNAT1</i>	50.38 +/- 8.53	100.0 +/- 0.0	99.94 +/- 0.43	97.74 +/- 6.81
<i>GNAT2</i>	52.1 +/- 8.63	99.76 +/- 0.56	99.73 +/- 0.59	97.86 +/- 6.31
<i>GNB3</i>	49.1 +/- 8.64	100.0 +/- 0.0	99.89 +/- 0.75	97.7 +/- 6.67
<i>GNPTG</i>	52.91 +/- 8.59	100.0 +/- 0.0	99.77 +/- 1.05	98.06 +/- 4.33
<i>GPR143</i>	40.07 +/- 15.64	99.57 +/- 2.01	91.05 +/- 15.77	63.84 +/- 41.09
<i>GPR179</i>	51.37 +/- 8.18	100.0 +/- 0.0	99.86 +/- 0.7	98.0 +/- 6.16
<i>GRK1</i>	51.45 +/- 8.48	100.0 +/- 0.03	99.88 +/- 0.73	98.11 +/- 6.08
<i>GRM6</i>	54.14 +/- 8.23	100.0 +/- 0.0	99.92 +/- 0.51	98.46 +/- 5.29
<i>GRN</i>	53.75 +/- 8.59	100.0 +/- 0.0	100.0 +/- 0.0	98.96 +/- 3.52
<i>GUCA1A</i>	51.96 +/- 9.16	100.0 +/- 0.0	100.0 +/- 0.0	98.63 +/- 4.91
<i>GUCA1B</i>	51.93 +/- 7.56	100.0 +/- 0.0	100.0 +/- 0.0	98.41 +/- 5.81

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>GUCY2D</i>	52.39 +/- 8.39	99.9 +/- 0.0	99.74 +/- 0.66	97.63 +/- 5.27
<i>HARS1</i>	52.54 +/- 8.15	100.0 +/- 0.0	99.96 +/- 0.28	98.46 +/- 6.7
<i>HCCS</i>	42.84 +/- 16.27	99.83 +/- 1.1	93.94 +/- 11.68	70.0 +/- 35.47
<i>HGSNAT</i>	53.32 +/- 7.96	100.0 +/- 0.0	99.99 +/- 0.06	99.0 +/- 2.22
<i>HK1</i>	52.77 +/- 8.71	100.0 +/- 0.0	99.93 +/- 0.39	98.5 +/- 5.51
<i>HMX1</i>	48.36 +/- 9.52	100.0 +/- 0.0	99.38 +/- 2.74	93.21 +/- 11.78
<i>IDH3A</i>	56.06 +/- 8.01	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.47
<i>IDH3B</i>	50.47 +/- 8.28	100.0 +/- 0.0	99.99 +/- 0.09	98.42 +/- 4.22
<i>IFT140</i>	55.06 +/- 8.53	100.0 +/- 0.0	100.0 +/- 0.01	99.16 +/- 2.44
<i>IFT172</i>	51.73 +/- 7.98	100.0 +/- 0.0	99.98 +/- 0.09	98.92 +/- 3.1
<i>IFT27</i>	48.35 +/- 8.62	99.89 +/- 0.75	97.97 +/- 2.78	91.88 +/- 7.4
<i>IFT74</i>	54.98 +/- 7.35	100.0 +/- 0.03	99.98 +/- 0.06	99.66 +/- 0.72
<i>IFT81</i>	53.91 +/- 7.36	100.0 +/- 0.0	99.97 +/- 0.17	98.55 +/- 2.42
<i>IMPDH1</i>	49.93 +/- 8.55	100.0 +/- 0.0	99.93 +/- 0.45	97.32 +/- 7.41
<i>IMPG1</i>	55.35 +/- 7.91	100.0 +/- 0.0	100.0 +/- 0.01	99.59 +/- 0.69
<i>IMPG2</i>	54.07 +/- 7.62	99.88 +/- 0.14	99.58 +/- 0.28	98.47 +/- 2.24
<i>INPP5E</i>	50.81 +/- 8.98	100.0 +/- 0.0	99.5 +/- 2.37	95.13 +/- 8.84
<i>IQCB1</i>	54.8 +/- 7.29	99.92 +/- 0.1	99.89 +/- 0.1	99.46 +/- 0.92
<i>JAG1</i>	54.24 +/- 7.78	100.0 +/- 0.0	99.94 +/- 0.37	98.84 +/- 2.65
<i>KCNJ13</i>	54.45 +/- 7.97	100.0 +/- 0.0	100.0 +/- 0.01	99.48 +/- 1.52
<i>KCNV2</i>	58.57 +/- 8.43	100.0 +/- 0.0	100.0 +/- 0.0	99.7 +/- 0.93
<i>KIAA1549</i>	54.12 +/- 8.21	100.0 +/- 0.0	99.91 +/- 0.27	98.75 +/- 3.37
<i>KIF11</i>	55.52 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.0	99.74 +/- 0.86
<i>KIF3B</i>	51.97 +/- 8.1	100.0 +/- 0.0	99.99 +/- 0.04	98.86 +/- 2.14
<i>KIZ</i>	54.43 +/- 7.3	100.0 +/- 0.0	99.98 +/- 0.16	99.49 +/- 1.55
<i>KLHL7</i>	54.71 +/- 8.04	100.0 +/- 0.0	99.94 +/- 0.37	99.55 +/- 1.22
<i>LAMA1</i>	55.01 +/- 7.48	100.0 +/- 0.0	100.0 +/- 0.0	99.52 +/- 1.26
<i>LAMP2</i>	43.14 +/- 15.76	99.97 +/- 0.17	95.17 +/- 8.15	71.26 +/- 34.03
<i>LCA5</i>	53.98 +/- 7.16	100.0 +/- 0.0	100.0 +/- 0.01	99.63 +/- 0.92
<i>LIG3</i>	52.98 +/- 7.94	100.0 +/- 0.0	99.97 +/- 0.09	98.6 +/- 3.19
<i>LRAT</i>	54.61 +/- 7.2	100.0 +/- 0.0	99.97 +/- 0.13	99.31 +/- 1.97
<i>LRIT3</i>	53.86 +/- 7.89	100.0 +/- 0.0	100.0 +/- 0.0	99.47 +/- 1.21
<i>LRP2</i>	54.31 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.01	99.41 +/- 1.36
<i>LRP5</i>	52.36 +/- 8.32	99.98 +/- 0.07	99.55 +/- 1.19	96.62 +/- 5.5
<i>LZTFL1</i>	56.16 +/- 8.07	100.0 +/- 0.0	99.99 +/- 0.03	99.54 +/- 1.34
<i>MAK</i>	54.87 +/- 7.86	100.0 +/- 0.0	100.0 +/- 0.0	99.62 +/- 0.93
<i>MAPKAPK3</i>	53.56 +/- 8.56	100.0 +/- 0.03	99.9 +/- 0.61	97.8 +/- 5.7
<i>MED12</i>	38.72 +/- 15.13	99.63 +/- 2.0	88.91 +/- 17.25	62.9 +/- 40.62
<i>MERTK</i>	53.65 +/- 7.9	100.0 +/- 0.0	99.95 +/- 0.36	99.04 +/- 2.33
<i>MFRP</i>	48.77 +/- 8.23	100.0 +/- 0.0	99.75 +/- 1.58	96.88 +/- 8.76
<i>MFSD8</i>	55.98 +/- 7.76	100.0 +/- 0.0	100.0 +/- 0.0	99.73 +/- 0.66
<i>MKKS</i>	55.12 +/- 7.37	100.0 +/- 0.0	100.0 +/- 0.01	99.57 +/- 1.06
<i>MKS1</i>	50.76 +/- 8.19	100.0 +/- 0.0	99.84 +/- 1.1	98.54 +/- 4.1
<i>MMACHC</i>	49.18 +/- 8.8	100.0 +/- 0.0	99.81 +/- 0.88	96.27 +/- 8.17
<i>MSTO1</i>	40.98 +/- 6.05	99.91 +/- 0.26	94.8 +/- 5.32	77.79 +/- 12.42
<i>MTRFR</i>	54.41 +/- 8.59	100.0 +/- 0.0	99.98 +/- 0.12	99.3 +/- 1.67

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>MTTP</i>	55.42 +/- 7.93	100.0 +/- 0.0	99.98 +/- 0.12	99.39 +/- 1.55
<i>MVK</i>	51.88 +/- 8.0	100.0 +/- 0.0	99.94 +/- 0.39	98.24 +/- 5.57
<i>MYO7A</i>	51.81 +/- 8.53	100.0 +/- 0.0	99.92 +/- 0.47	97.97 +/- 5.44
<i>NBAS</i>	55.51 +/- 7.41	100.0 +/- 0.0	100.0 +/- 0.0	99.52 +/- 1.16
<i>NDP</i>	42.79 +/- 16.25	99.96 +/- 0.25	93.61 +/- 13.18	70.24 +/- 35.63
<i>NEUROD1</i>	52.33 +/- 6.66	100.0 +/- 0.0	100.0 +/- 0.01	98.98 +/- 2.61
<i>NMNAT1</i>	52.26 +/- 8.65	100.0 +/- 0.0	99.89 +/- 0.73	98.65 +/- 4.56
<i>NPHP1</i>	55.34 +/- 9.45	100.0 +/- 0.0	99.84 +/- 1.06	98.35 +/- 7.05
<i>NPHP3</i>	55.11 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.01	99.33 +/- 1.56
<i>NPHP4</i>	53.37 +/- 7.99	100.0 +/- 0.0	99.92 +/- 0.44	98.44 +/- 3.59
<i>NR2E3</i>	53.73 +/- 8.51	100.0 +/- 0.0	99.95 +/- 0.24	97.79 +/- 6.4
<i>NRL</i>	51.76 +/- 8.65	100.0 +/- 0.0	99.94 +/- 0.33	98.62 +/- 4.48
<i>NYX</i>	45.5 +/- 17.68	99.96 +/- 0.24	93.82 +/- 14.67	71.89 +/- 34.41
<i>OAT</i>	55.98 +/- 8.06	100.0 +/- 0.0	100.0 +/- 0.0	99.32 +/- 1.48
<i>OFD1</i>	42.18 +/- 15.83	99.93 +/- 0.36	93.98 +/- 9.99	68.58 +/- 36.59
<i>OPN1SW</i>	54.73 +/- 8.94	100.0 +/- 0.0	99.99 +/- 0.04	98.7 +/- 5.91
<i>OTX2</i>	52.15 +/- 8.58	100.0 +/- 0.0	100.0 +/- 0.0	98.91 +/- 2.84
<i>P3H2</i>	53.41 +/- 7.77	100.0 +/- 0.01	99.86 +/- 0.61	98.5 +/- 4.09
<i>PANK2</i>	52.0 +/- 7.82	100.0 +/- 0.02	99.89 +/- 0.27	97.84 +/- 4.21
<i>PAX2</i>	45.55 +/- 7.82	100.0 +/- 0.01	99.64 +/- 1.68	93.13 +/- 12.12
<i>PCARE</i>	51.23 +/- 7.92	100.0 +/- 0.0	99.83 +/- 0.5	97.36 +/- 4.45
<i>PCDH15</i>	56.23 +/- 7.46	100.0 +/- 0.0	100.0 +/- 0.0	99.64 +/- 1.09
<i>PCYT1A</i>	53.31 +/- 8.04	100.0 +/- 0.0	99.98 +/- 0.1	99.23 +/- 2.6
<i>PDE6A</i>	54.08 +/- 8.03	100.0 +/- 0.0	99.99 +/- 0.07	98.96 +/- 2.98
<i>PDE6B</i>	54.18 +/- 8.93	100.0 +/- 0.0	99.89 +/- 0.49	98.52 +/- 5.47
<i>PDE6C</i>	53.01 +/- 7.61	100.0 +/- 0.0	99.94 +/- 0.42	98.91 +/- 3.03
<i>PDE6G</i>	47.07 +/- 9.53	100.0 +/- 0.01	99.76 +/- 1.01	94.21 +/- 13.16
<i>PDE6H</i>	55.74 +/- 9.01	100.0 +/- 0.0	100.0 +/- 0.0	99.84 +/- 0.53
<i>PDSS1</i>	55.54 +/- 6.89	100.0 +/- 0.0	99.86 +/- 0.74	99.08 +/- 2.49
<i>PDZD7</i>	47.1 +/- 8.19	100.0 +/- 0.0	99.44 +/- 3.11	94.46 +/- 11.79
<i>PEX1</i>	55.38 +/- 6.94	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.11
<i>PEX2</i>	54.88 +/- 8.0	100.0 +/- 0.0	100.0 +/- 0.0	99.74 +/- 0.88
<i>PEX6</i>	49.64 +/- 7.85	99.96 +/- 0.05	99.82 +/- 0.41	96.66 +/- 7.29
<i>PEX7</i>	56.22 +/- 7.44	100.0 +/- 0.0	100.0 +/- 0.0	99.46 +/- 1.84
<i>PGK1</i>	43.93 +/- 16.0	99.94 +/- 0.2	96.29 +/- 7.81	72.91 +/- 33.11
<i>PHYH</i>	55.25 +/- 7.86	99.96 +/- 0.05	99.95 +/- 0.05	99.45 +/- 1.74
<i>PITPNM3</i>	49.57 +/- 8.05	99.98 +/- 0.04	99.66 +/- 0.77	96.64 +/- 4.97
<i>PLA2G5</i>	51.77 +/- 7.98	100.0 +/- 0.0	99.98 +/- 0.13	98.63 +/- 4.94
<i>PLK4</i>	55.66 +/- 7.66	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 0.99
<i>PNPLA6</i>	52.2 +/- 8.63	100.0 +/- 0.0	99.94 +/- 0.24	97.88 +/- 5.9
<i>POC1B</i>	55.28 +/- 7.97	100.0 +/- 0.0	99.97 +/- 0.18	99.32 +/- 1.38
<i>POC5</i>	54.42 +/- 7.6	100.0 +/- 0.0	100.0 +/- 0.01	99.19 +/- 2.08
<i>POMGNT1</i>	52.11 +/- 9.18	100.0 +/- 0.0	99.94 +/- 0.42	98.61 +/- 4.01
<i>POMGNT2</i>	54.16 +/- 8.27	100.0 +/- 0.0	99.93 +/- 0.49	98.39 +/- 3.97
<i>POMT1</i>	53.59 +/- 8.44	100.0 +/- 0.0	99.98 +/- 0.12	99.07 +/- 3.38
<i>PPT1</i>	53.27 +/- 7.55	100.0 +/- 0.0	99.97 +/- 0.22	99.2 +/- 2.6

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>PRCD</i>	50.39 +/- 8.08	100.0 +/- 0.0	99.85 +/- 0.58	98.15 +/- 4.65
<i>PRDM13</i>	52.75 +/- 7.97	99.98 +/- 0.1	99.51 +/- 2.39	97.3 +/- 6.89
<i>PROM1</i>	54.31 +/- 7.9	100.0 +/- 0.0	100.0 +/- 0.03	99.28 +/- 2.32
<i>PRPF3</i>	54.11 +/- 7.76	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.41
<i>PRPF31</i>	51.37 +/- 8.12	100.0 +/- 0.0	99.88 +/- 0.83	98.07 +/- 6.25
<i>PRPF4</i>	53.86 +/- 8.37	100.0 +/- 0.0	100.0 +/- 0.01	99.42 +/- 2.41
<i>PRPF6</i>	52.61 +/- 7.9	100.0 +/- 0.0	100.0 +/- 0.0	98.94 +/- 3.24
<i>PRPF8</i>	51.5 +/- 8.41	100.0 +/- 0.0	99.99 +/- 0.06	98.75 +/- 4.15
<i>PRPH2</i>	52.8 +/- 7.63	100.0 +/- 0.0	99.99 +/- 0.03	98.72 +/- 3.8
<i>PRPS1</i>	39.1 +/- 15.28	99.38 +/- 1.99	89.0 +/- 16.91	63.4 +/- 37.45
<i>RAB28</i>	54.23 +/- 7.15	100.0 +/- 0.0	100.0 +/- 0.0	98.6 +/- 2.57
<i>RAX2</i>	51.68 +/- 9.15	100.0 +/- 0.0	99.81 +/- 1.25	97.18 +/- 6.57
<i>RBP3</i>	55.27 +/- 8.07	100.0 +/- 0.0	99.97 +/- 0.18	99.31 +/- 3.61
<i>RBP4</i>	51.7 +/- 8.16	100.0 +/- 0.0	99.86 +/- 0.91	97.65 +/- 4.38
<i>RCBTB1</i>	54.48 +/- 7.67	99.93 +/- 0.14	99.82 +/- 0.43	98.96 +/- 1.59
<i>RD3</i>	51.49 +/- 8.06	100.0 +/- 0.0	100.0 +/- 0.01	98.65 +/- 3.52
<i>RDH11</i>	53.51 +/- 7.58	100.0 +/- 0.0	100.0 +/- 0.03	99.38 +/- 1.06
<i>RDH12</i>	52.02 +/- 7.92	100.0 +/- 0.0	100.0 +/- 0.0	98.95 +/- 3.84
<i>RDH5</i>	49.35 +/- 8.82	100.0 +/- 0.0	99.9 +/- 0.69	96.95 +/- 8.79
<i>REEP6</i>	50.01 +/- 8.46	100.0 +/- 0.0	99.52 +/- 2.01	95.97 +/- 8.53
<i>RGR</i>	52.6 +/- 8.33	100.0 +/- 0.0	99.99 +/- 0.06	98.57 +/- 5.14
<i>RGS9</i>	51.38 +/- 7.73	100.0 +/- 0.0	100.0 +/- 0.0	98.65 +/- 3.79
<i>RGS9BP</i>	52.71 +/- 8.78	100.0 +/- 0.0	99.95 +/- 0.15	98.2 +/- 3.72
<i>RHO</i>	52.26 +/- 8.52	99.98 +/- 0.1	99.96 +/- 0.18	98.88 +/- 3.97
<i>RIMS2</i>	55.06 +/- 7.33	100.0 +/- 0.0	99.88 +/- 0.6	98.98 +/- 1.77
<i>RLBP1</i>	53.25 +/- 8.83	100.0 +/- 0.0	100.0 +/- 0.0	99.14 +/- 3.64
<i>RNU4ATAC</i>	58.08 +/- 10.68	100.0 +/- 0.0	100.0 +/- 0.0	99.88 +/- 0.8
<i>ROM1</i>	50.78 +/- 8.19	100.0 +/- 0.0	100.0 +/- 0.0	98.09 +/- 6.06
<i>RP1</i>	52.29 +/- 7.5	100.0 +/- 0.0	99.96 +/- 0.23	98.46 +/- 3.11
<i>RP1L1</i>	51.31 +/- 8.88	99.99 +/- 0.04	99.85 +/- 0.82	97.21 +/- 6.12
<i>RP2</i>	42.75 +/- 15.83	99.95 +/- 0.24	94.59 +/- 10.43	69.56 +/- 35.43
<i>RP9</i>	52.59 +/- 8.38	99.94 +/- 0.39	99.36 +/- 2.54	97.55 +/- 5.03
<i>RPE65</i>	54.67 +/- 7.5	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 1.17
<i>RPGR²</i>	35.4 +/- 12.99	91.12 +/- 5.75	77.91 +/- 12.72	54.81 +/- 30.0
<i>RPGRIP1</i>	53.96 +/- 7.82	100.0 +/- 0.0	100.0 +/- 0.01	99.15 +/- 2.94
<i>RPGRIP1L</i>	54.65 +/- 7.42	100.0 +/- 0.0	100.0 +/- 0.0	99.66 +/- 0.88
<i>RS1</i>	40.86 +/- 15.91	99.84 +/- 0.81	91.2 +/- 13.99	65.91 +/- 37.74
<i>RTN4IP1</i>	53.39 +/- 8.09	100.0 +/- 0.0	100.0 +/- 0.02	99.08 +/- 2.82
<i>SAG</i>	53.19 +/- 8.06	100.0 +/- 0.0	99.99 +/- 0.04	99.16 +/- 3.05
<i>SAMD11</i>	46.95 +/- 8.82	100.0 +/- 0.0	99.36 +/- 2.04	92.92 +/- 12.36
<i>SAMD7</i>	51.78 +/- 7.62	100.0 +/- 0.0	99.94 +/- 0.19	98.38 +/- 3.05
<i>SCAPER</i>	55.09 +/- 7.32	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 0.84
<i>SDCCAG8</i>	55.16 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.53 +/- 1.56
<i>SEMA4A</i>	49.69 +/- 7.86	99.98 +/- 0.13	99.52 +/- 1.03	95.8 +/- 6.46
<i>SLC24A1</i>	52.36 +/- 7.77	100.0 +/- 0.0	99.96 +/- 0.21	98.99 +/- 3.2
<i>SLC25A46</i>	54.42 +/- 7.38	100.0 +/- 0.0	99.99 +/- 0.07	99.43 +/- 1.22

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>SLC37A3</i>	53.48 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.01	99.02 +/- 2.51
<i>SLC38A8</i>	51.3 +/- 8.37	100.0 +/- 0.0	99.95 +/- 0.3	97.96 +/- 6.97
<i>SLC6A6</i>	51.33 +/- 7.94	100.0 +/- 0.0	99.88 +/- 0.73	98.01 +/- 4.59
<i>SLC7A14</i>	55.08 +/- 8.02	100.0 +/- 0.0	99.96 +/- 0.16	99.17 +/- 1.93
<i>SNRNP200</i>	52.63 +/- 9.1	100.0 +/- 0.0	99.84 +/- 0.87	97.71 +/- 8.58
<i>SPATA7</i>	55.16 +/- 7.41	100.0 +/- 0.0	100.0 +/- 0.0	99.53 +/- 1.42
<i>SPP2</i>	54.5 +/- 7.98	100.0 +/- 0.0	100.0 +/- 0.0	99.11 +/- 2.74
<i>SPTLC1</i>	55.52 +/- 7.78	100.0 +/- 0.0	100.0 +/- 0.0	99.46 +/- 1.27
<i>SRD5A3</i>	54.39 +/- 7.94	99.99 +/- 0.04	99.92 +/- 0.42	99.21 +/- 1.59
<i>SSBP1</i>	55.54 +/- 7.85	100.0 +/- 0.0	100.0 +/- 0.0	99.75 +/- 0.92
<i>STN1</i>	54.12 +/- 8.47	100.0 +/- 0.0	99.99 +/- 0.04	99.28 +/- 2.46
<i>TEAD1</i>	54.55 +/- 7.73	100.0 +/- 0.0	99.78 +/- 0.6	98.7 +/- 2.0
<i>TIMM8A</i>	40.71 +/- 14.41	99.9 +/- 0.5	93.69 +/- 13.01	68.13 +/- 36.85
<i>TIMP3</i>	52.28 +/- 7.98	100.0 +/- 0.0	99.95 +/- 0.21	98.69 +/- 3.27
<i>TINF2</i>	51.29 +/- 8.06	100.0 +/- 0.0	100.0 +/- 0.01	97.99 +/- 6.43
<i>TLCD3B</i>	44.51 +/- 7.58	99.87 +/- 0.45	97.05 +/- 4.2	86.93 +/- 9.55
<i>TMEM216</i>	51.73 +/- 9.05	100.0 +/- 0.0	100.0 +/- 0.0	98.86 +/- 2.76
<i>TMEM218</i>	54.07 +/- 8.21	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 1.69
<i>TMEM231</i>	52.27 +/- 8.24	100.0 +/- 0.01	99.59 +/- 0.76	96.3 +/- 4.7
<i>TMEM237</i>	54.62 +/- 7.76	100.0 +/- 0.0	99.97 +/- 0.22	99.43 +/- 1.88
<i>TOPORS</i>	53.59 +/- 7.26	100.0 +/- 0.0	100.0 +/- 0.03	99.42 +/- 1.59
<i>TPP1</i>	50.74 +/- 8.83	100.0 +/- 0.0	100.0 +/- 0.0	98.62 +/- 3.99
<i>TRAF3IP1</i>	54.04 +/- 8.11	100.0 +/- 0.0	99.91 +/- 0.5	98.43 +/- 3.04
<i>TREX1</i>	51.23 +/- 9.75	100.0 +/- 0.0	100.0 +/- 0.0	97.72 +/- 6.99
<i>TRIM32</i>	55.0 +/- 8.0	100.0 +/- 0.0	99.83 +/- 0.45	98.45 +/- 2.54
<i>TRNT1</i>	56.47 +/- 7.62	100.0 +/- 0.0	100.0 +/- 0.0	99.75 +/- 0.55
<i>TRPM1</i>	54.84 +/- 7.72	100.0 +/- 0.0	99.99 +/- 0.06	99.44 +/- 1.76
<i>TSPAN12</i>	55.02 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.0 +/- 2.78
<i>TTC8</i>	55.08 +/- 7.81	100.0 +/- 0.0	99.87 +/- 0.7	99.14 +/- 2.09
<i>TTL5</i>	53.77 +/- 7.49	100.0 +/- 0.0	99.95 +/- 0.33	99.28 +/- 2.03
<i>TTPA</i>	54.78 +/- 7.23	99.98 +/- 0.04	99.97 +/- 0.05	99.0 +/- 3.25
<i>TUB</i>	52.68 +/- 7.98	100.0 +/- 0.0	99.8 +/- 0.74	98.28 +/- 4.17
<i>TUBB4B</i>	53.33 +/- 8.7	100.0 +/- 0.0	99.86 +/- 0.45	97.22 +/- 5.93
<i>TUBGCP4</i>	53.0 +/- 8.1	100.0 +/- 0.0	99.92 +/- 0.46	98.82 +/- 3.44
<i>TUBGCP6</i>	55.06 +/- 8.96	100.0 +/- 0.0	99.94 +/- 0.36	98.93 +/- 3.92
<i>TULP1</i>	48.92 +/- 8.38	100.0 +/- 0.0	99.9 +/- 0.66	96.84 +/- 7.55
<i>UNC119</i>	48.43 +/- 9.0	100.0 +/- 0.0	99.77 +/- 1.55	95.28 +/- 11.64
<i>USH1C</i>	48.75 +/- 8.86	100.0 +/- 0.0	99.32 +/- 1.28	94.2 +/- 7.31
<i>USH1G</i>	52.0 +/- 8.88	100.0 +/- 0.0	99.68 +/- 0.98	95.85 +/- 6.19
<i>USH2A</i>	55.66 +/- 7.82	100.0 +/- 0.0	100.0 +/- 0.01	99.6 +/- 1.15
<i>USP45</i>	56.57 +/- 7.55	100.0 +/- 0.02	99.99 +/- 0.03	99.75 +/- 0.53
<i>VCAN</i>	54.23 +/- 7.5	99.97 +/- 0.05	99.94 +/- 0.05	99.47 +/- 1.4
<i>VPS13B</i>	55.06 +/- 7.31	100.0 +/- 0.0	100.0 +/- 0.01	99.62 +/- 1.05
<i>WDPCP</i>	55.08 +/- 7.68	100.0 +/- 0.0	99.96 +/- 0.16	99.43 +/- 1.45
<i>WDR19</i>	55.29 +/- 7.59	100.0 +/- 0.0	99.98 +/- 0.13	99.6 +/- 1.17
<i>WHRN</i>	50.55 +/- 8.43	99.94 +/- 0.09	99.81 +/- 0.63	97.06 +/- 7.63
<i>ZFYVE26</i>	52.88 +/- 7.91	100.0 +/- 0.0	99.94 +/- 0.29	98.83 +/- 3.35

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
ZNF408	53.44 +/- 8.18	100.0 +/- 0.0	99.98 +/- 0.06	98.84 +/- 4.72
ZNF423	52.79 +/- 8.22	99.99 +/- 0.08	99.68 +/- 0.81	97.53 +/- 3.64
ZNF513	50.39 +/- 8.93	100.0 +/- 0.0	99.85 +/- 0.59	96.29 +/- 7.07

For supplerende oplysninger vedrørende analysen kan afdelingen kontaktes på mail: mol-dia@rn.dk. Rapport genereret: 09/10-2025