

Analyse af gener relevante ved mistanke om Hereditær ataksi og spastisk paraplegi (udvidet panel)

Panelbeskrivelse

Navn: Analyse af gener relevante ved mistanke om Hereditær ataksi og spastisk paraplegi (udvidet panel)

Antal gener: 569

version: 2.1

Ibrugtagningsdato: 2023-06-01

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 fortages *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. Der er undersøgt for short tandem repeat ekspansioner i relevante gener ved brug af Expansion Hunter-softwaren. Metoden er beskrevet i følgende publikationer: PMID: 28887402 og 31134279. Expansion Hunter er et screeningsværktøj, og alle patogene ekspansioner skal bekræftes med anden metode. For generne *SMN1* og *SMN2* er der udelukkende undersøgt for kopitalsændringer ved brug af *SMN Caller* softwaren. Metoden er beskrevet i følgende publikation: PMID: 32066871. *SMN Caller* softwaren er et screeningsværktøj, og alle patogene kopitalsændringer skal bekræftes med anden metode. 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 [%]
AAAS	52.63 +/- 7.98	100.0 +/- 0.0	99.96 +/- 0.25	98.83 +/- 5.07
AARS1	52.22 +/- 8.24	100.0 +/- 0.0	99.99 +/- 0.09	98.83 +/- 2.63
ABCA2	51.5 +/- 8.93	99.99 +/- 0.05	99.51 +/- 1.09	95.66 +/- 6.11
ABCB7	44.2 +/- 16.13	99.99 +/- 0.03	96.19 +/- 8.22	73.19 +/- 32.92
ABCD1	40.69 +/- 15.58	99.84 +/- 0.77	90.9 +/- 15.14	66.18 +/- 38.73
ABHD12	50.57 +/- 8.62	100.0 +/- 0.01	99.74 +/- 0.8	96.96 +/- 6.38
ABHD16A	50.0 +/- 8.43	100.0 +/- 0.0	99.84 +/- 0.52	97.32 +/- 6.12
ACER3	54.6 +/- 7.7	99.88 +/- 0.1	99.85 +/- 0.13	98.96 +/- 1.53
ACO2	52.76 +/- 9.0	100.0 +/- 0.0	99.97 +/- 0.21	98.21 +/- 6.91
ADAR	52.76 +/- 8.27	100.0 +/- 0.0	99.94 +/- 0.2	98.73 +/- 3.46
ADCY5	51.85 +/- 8.42	100.0 +/- 0.0	99.86 +/- 0.44	97.15 +/- 7.11
ADGRG1	50.81 +/- 8.3	100.0 +/- 0.0	99.98 +/- 0.09	97.84 +/- 5.27
ADPRS	53.85 +/- 9.18	100.0 +/- 0.0	100.0 +/- 0.01	98.8 +/- 4.56
AFG2B	53.18 +/- 8.08	100.0 +/- 0.0	99.87 +/- 0.64	98.07 +/- 6.1
AFG3L2	54.26 +/- 7.85	100.0 +/- 0.0	99.96 +/- 0.25	98.97 +/- 2.49
AHI1	55.19 +/- 7.32	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 0.81
AIMP1	54.17 +/- 7.19	100.0 +/- 0.0	100.0 +/- 0.0	99.36 +/- 1.47

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
ALAS2	41.66 +/- 16.0	99.71 +/- 1.95	93.23 +/- 11.56	67.25 +/- 38.72
ALDH18A1	54.07 +/- 7.8	100.0 +/- 0.0	99.96 +/- 0.28	99.09 +/- 2.87
ALDH3A2	52.74 +/- 7.61	100.0 +/- 0.02	99.99 +/- 0.03	98.94 +/- 1.83
ALDH5A1	53.79 +/- 7.43	100.0 +/- 0.0	99.96 +/- 0.22	99.18 +/- 2.73
ALG1	52.41 +/- 8.45	99.96 +/- 0.05	99.95 +/- 0.06	99.05 +/- 4.0
ALG10	53.38 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.01	99.25 +/- 2.15
ALG11	54.73 +/- 7.73	100.0 +/- 0.0	99.99 +/- 0.06	99.53 +/- 1.15
ALG12	56.13 +/- 8.89	99.98 +/- 0.13	99.68 +/- 0.73	97.99 +/- 4.33
ALG13	42.18 +/- 15.57	99.92 +/- 0.25	93.61 +/- 9.57	68.82 +/- 34.14
ALG14	53.71 +/- 7.77	100.0 +/- 0.0	99.99 +/- 0.05	99.37 +/- 1.7
ALG2	55.37 +/- 8.44	100.0 +/- 0.0	100.0 +/- 0.0	99.49 +/- 1.63
ALG3	50.28 +/- 8.39	100.0 +/- 0.0	99.87 +/- 0.54	97.91 +/- 7.37
ALG6	54.48 +/- 7.68	100.0 +/- 0.03	100.0 +/- 0.03	99.47 +/- 1.23
ALG8	55.44 +/- 7.77	100.0 +/- 0.0	99.95 +/- 0.31	99.55 +/- 1.3
ALG9	54.81 +/- 8.3	99.99 +/- 0.05	99.99 +/- 0.05	99.35 +/- 1.81
ALS2	54.23 +/- 7.53	100.0 +/- 0.0	99.99 +/- 0.06	99.49 +/- 1.44
AMPD2	51.27 +/- 8.22	100.0 +/- 0.0	99.88 +/- 0.59	98.16 +/- 4.33
ANO10	53.61 +/- 7.6	100.0 +/- 0.0	99.99 +/- 0.04	99.2 +/- 3.18
AP1S2	43.34 +/- 16.23	99.77 +/- 0.96	94.39 +/- 11.4	72.04 +/- 33.13
AP4B1	54.44 +/- 8.15	100.0 +/- 0.0	100.0 +/- 0.0	99.4 +/- 2.46
AP4E1	55.73 +/- 7.25	100.0 +/- 0.0	100.0 +/- 0.0	99.69 +/- 0.65
AP4M1	50.12 +/- 8.82	100.0 +/- 0.0	99.87 +/- 0.78	97.77 +/- 7.54
AP4S1	54.2 +/- 7.87	100.0 +/- 0.0	99.99 +/- 0.09	99.38 +/- 1.18
AP5Z1	54.6 +/- 8.9	100.0 +/- 0.0	99.93 +/- 0.43	98.57 +/- 4.88
APTX	53.55 +/- 8.23	100.0 +/- 0.0	100.0 +/- 0.01	99.62 +/- 0.82
ARG1	57.77 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.0	99.88 +/- 0.46
ARL13B	55.42 +/- 6.97	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 0.82
ARL3	51.14 +/- 7.99	100.0 +/- 0.0	99.89 +/- 0.29	98.1 +/- 3.82
ARL6IP1	57.22 +/- 8.33	100.0 +/- 0.0	100.0 +/- 0.0	99.64 +/- 0.79
ARMC9	52.98 +/- 8.18	100.0 +/- 0.01	99.98 +/- 0.09	98.97 +/- 3.48
ARSA	48.47 +/- 7.85	99.65 +/- 0.28	99.27 +/- 0.98	96.16 +/- 5.99
ARSI	52.02 +/- 8.83	100.0 +/- 0.0	99.98 +/- 0.1	97.54 +/- 5.83
ATAD3A	48.89 +/- 8.14	99.97 +/- 0.21	99.32 +/- 1.57	93.42 +/- 7.9
ATCAY	52.19 +/- 8.1	100.0 +/- 0.0	99.95 +/- 0.33	98.74 +/- 3.93
ATG7	53.52 +/- 8.19	100.0 +/- 0.0	99.98 +/- 0.16	98.52 +/- 4.62
ATL1	54.36 +/- 7.58	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.26
ATM	55.4 +/- 7.19	100.0 +/- 0.0	99.99 +/- 0.03	99.49 +/- 0.98
ATN1	44.43 +/- 7.51	99.96 +/- 0.14	98.08 +/- 3.62	88.3 +/- 12.51
ATP13A2	51.75 +/- 8.62	100.0 +/- 0.0	99.88 +/- 0.45	97.48 +/- 6.81
ATP1A2	51.77 +/- 7.93	100.0 +/- 0.0	100.0 +/- 0.01	98.68 +/- 5.18
ATP1A3	48.22 +/- 8.29	100.0 +/- 0.0	99.67 +/- 0.84	95.22 +/- 8.1
ATP2B3	41.62 +/- 16.01	99.74 +/- 1.21	92.54 +/- 13.05	67.42 +/- 37.11
ATP6AP1	39.35 +/- 15.09	99.76 +/- 1.1	90.64 +/- 16.23	64.66 +/- 40.72
ATP6V0A1	52.07 +/- 7.57	100.0 +/- 0.0	100.0 +/- 0.0	98.89 +/- 3.11
ATP6V0A2	53.61 +/- 7.71	100.0 +/- 0.0	99.99 +/- 0.07	99.26 +/- 1.69
ATP7B	53.21 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.03	99.16 +/- 2.06

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>ATP8A2</i>	53.93 +/- 7.33	100.0 +/- 0.0	100.0 +/- 0.0	99.14 +/- 1.76
<i>ATXN1</i>	54.83 +/- 7.83	100.0 +/- 0.0	99.98 +/- 0.14	98.95 +/- 3.57
<i>ATXN10</i>	54.25 +/- 7.49	100.0 +/- 0.0	99.88 +/- 0.45	98.3 +/- 2.98
<i>ATXN2</i>	52.92 +/- 7.5	99.93 +/- 0.05	99.51 +/- 0.81	96.45 +/- 4.75
<i>ATXN3</i>	54.36 +/- 7.44	100.0 +/- 0.01	99.87 +/- 0.3	98.21 +/- 2.29
<i>ATXN7</i>	50.36 +/- 7.01	99.85 +/- 0.18	97.84 +/- 1.94	92.92 +/- 4.38
<i>ATXN8OS</i>	54.36 +/- 7.64	100.0 +/- 0.0	100.0 +/- 0.0	99.23 +/- 2.31
<i>AUH</i>	54.68 +/- 7.55	100.0 +/- 0.0	100.0 +/- 0.0	99.27 +/- 2.11
<i>B3GALNT2</i>	54.46 +/- 7.74	100.0 +/- 0.0	99.96 +/- 0.2	99.41 +/- 1.46
<i>B3GALT6</i>	54.13 +/- 9.42	100.0 +/- 0.0	99.68 +/- 1.64	97.2 +/- 6.81
<i>B3GAT3</i>	50.4 +/- 8.02	100.0 +/- 0.0	99.96 +/- 0.24	98.49 +/- 3.89
<i>B3GLCT</i>	54.52 +/- 7.58	99.86 +/- 0.09	99.86 +/- 0.09	99.11 +/- 1.48
<i>B4GALNT1</i>	49.58 +/- 8.27	100.0 +/- 0.0	99.92 +/- 0.46	97.44 +/- 7.28
<i>B4GALT1</i>	52.1 +/- 7.87	100.0 +/- 0.0	99.9 +/- 0.58	98.37 +/- 4.93
<i>B4GALT7</i>	50.02 +/- 8.54	99.99 +/- 0.07	99.76 +/- 1.15	96.99 +/- 6.52
<i>B4GAT1</i>	51.39 +/- 8.44	100.0 +/- 0.0	99.84 +/- 1.09	97.87 +/- 8.35
<i>B9D1</i>	51.09 +/- 8.08	100.0 +/- 0.0	100.0 +/- 0.0	98.7 +/- 5.02
<i>B9D2</i>	51.21 +/- 8.93	100.0 +/- 0.0	100.0 +/- 0.0	98.53 +/- 5.79
<i>BBS1</i>	52.34 +/- 8.31	100.0 +/- 0.0	100.0 +/- 0.03	98.98 +/- 3.48
<i>BCAS3</i>	52.53 +/- 7.81	100.0 +/- 0.0	99.93 +/- 0.4	98.24 +/- 4.54
<i>BEAN1</i>	52.97 +/- 8.14	100.0 +/- 0.0	99.91 +/- 0.32	98.42 +/- 3.17
<i>BLOC1S1</i>	48.85 +/- 8.79	100.0 +/- 0.0	99.86 +/- 0.95	96.06 +/- 10.09
<i>BRF1</i>	52.87 +/- 9.02	100.0 +/- 0.0	99.8 +/- 0.98	97.4 +/- 7.85
<i>BSCL2</i>	52.38 +/- 8.46	100.0 +/- 0.0	99.99 +/- 0.06	98.79 +/- 6.57
<i>C19ORF12</i>	53.52 +/- 7.82	99.95 +/- 0.05	99.88 +/- 0.3	98.47 +/- 3.97
<i>C1GALT1C1</i>	44.72 +/- 16.04	99.99 +/- 0.05	96.5 +/- 7.69	74.67 +/- 31.45
<i>C2CD3</i>	53.05 +/- 7.83	100.0 +/- 0.0	100.0 +/- 0.03	99.02 +/- 2.45
<i>CA8</i>	54.27 +/- 7.46	100.0 +/- 0.0	100.0 +/- 0.01	99.35 +/- 1.59
<i>CACNA1A</i>	49.1 +/- 7.77	99.98 +/- 0.05	99.52 +/- 1.75	95.06 +/- 8.13
<i>CACNA1G</i>	49.87 +/- 8.16	100.0 +/- 0.0	99.9 +/- 0.41	96.71 +/- 6.9
<i>CACNA2D2</i>	49.95 +/- 8.77	100.0 +/- 0.01	99.61 +/- 1.41	96.11 +/- 7.62
<i>CACNB4</i>	55.4 +/- 7.13	100.0 +/- 0.0	99.98 +/- 0.09	99.42 +/- 1.14
<i>CAD</i>	52.53 +/- 8.24	100.0 +/- 0.0	99.96 +/- 0.28	98.58 +/- 4.76
<i>CAMLG</i>	55.34 +/- 8.16	100.0 +/- 0.0	99.96 +/- 0.25	99.17 +/- 2.77
<i>CAMTA1</i>	53.41 +/- 7.82	100.0 +/- 0.0	99.75 +/- 0.45	98.54 +/- 2.78
<i>CAPN1</i>	51.1 +/- 8.17	100.0 +/- 0.0	99.94 +/- 0.35	98.39 +/- 4.69
<i>CAPRIN1</i>	54.6 +/- 7.78	100.0 +/- 0.0	99.89 +/- 0.38	98.67 +/- 2.34
<i>CASK</i>	42.28 +/- 15.64	99.79 +/- 0.76	93.75 +/- 10.69	69.36 +/- 34.73
<i>CC2D2A</i>	54.85 +/- 8.01	100.0 +/- 0.0	99.99 +/- 0.09	99.36 +/- 2.46
<i>CCDC88C</i>	52.15 +/- 8.34	100.0 +/- 0.0	99.87 +/- 0.54	98.18 +/- 4.86
<i>CCT5</i>	54.77 +/- 7.56	100.0 +/- 0.0	99.97 +/- 0.21	99.48 +/- 1.06
<i>CDK16</i>	38.28 +/- 15.38	99.46 +/- 1.78	87.38 +/- 19.99	61.6 +/- 42.84
<i>CDK5</i>	50.49 +/- 8.84	100.0 +/- 0.0	99.92 +/- 0.51	98.45 +/- 5.47
<i>CENPF</i>	54.2 +/- 7.49	100.0 +/- 0.0	100.0 +/- 0.0	99.6 +/- 1.06
<i>CEP104</i>	52.61 +/- 7.68	100.0 +/- 0.0	99.69 +/- 0.8	96.19 +/- 4.4
<i>CEP290</i>	54.71 +/- 7.35	100.0 +/- 0.0	100.0 +/- 0.01	99.48 +/- 1.25
<i>CEP41</i>	53.27 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.01	99.35 +/- 1.63

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>CHMP1A</i>	52.69 +/- 9.04	100.0 +/- 0.0	99.97 +/- 0.21	98.41 +/- 3.19
<i>CHP1</i>	52.34 +/- 8.23	100.0 +/- 0.0	99.98 +/- 0.12	98.95 +/- 2.77
<i>CHST14</i>	46.39 +/- 8.4	99.99 +/- 0.04	98.75 +/- 3.5	91.65 +/- 12.65
<i>CHST3</i>	52.89 +/- 8.96	100.0 +/- 0.0	99.95 +/- 0.23	98.19 +/- 4.71
<i>CHST6</i>	52.56 +/- 8.53	100.0 +/- 0.03	99.6 +/- 0.92	96.62 +/- 5.91
<i>CHSY1</i>	52.66 +/- 7.1	99.93 +/- 0.38	99.26 +/- 2.56	96.97 +/- 5.41
<i>CILK1</i>	54.96 +/- 7.86	100.0 +/- 0.0	99.96 +/- 0.17	99.4 +/- 1.42
<i>CLCN2</i>	51.74 +/- 8.94	100.0 +/- 0.0	99.86 +/- 0.5	97.4 +/- 5.55
<i>CLN5</i>	55.95 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.0	99.7 +/- 0.68
<i>CLN6</i>	48.22 +/- 8.66	99.97 +/- 0.07	99.81 +/- 0.66	95.66 +/- 8.17
<i>CLP1</i>	52.87 +/- 9.02	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 2.66
<i>CLPP</i>	51.45 +/- 7.89	100.0 +/- 0.0	99.93 +/- 0.38	98.56 +/- 4.26
<i>COA7</i>	53.37 +/- 7.66	100.0 +/- 0.0	100.0 +/- 0.0	99.15 +/- 2.42
<i>COASY</i>	49.99 +/- 8.71	100.0 +/- 0.0	99.97 +/- 0.17	97.35 +/- 9.13
<i>COG1</i>	52.01 +/- 7.84	99.99 +/- 0.06	99.57 +/- 1.63	98.24 +/- 3.79
<i>COG2</i>	55.97 +/- 7.91	100.0 +/- 0.0	100.0 +/- 0.0	99.47 +/- 1.67
<i>COG4</i>	51.68 +/- 7.92	100.0 +/- 0.0	100.0 +/- 0.0	98.88 +/- 3.12
<i>COG5</i>	55.33 +/- 7.94	100.0 +/- 0.0	99.99 +/- 0.04	99.46 +/- 1.87
<i>COG6</i>	56.49 +/- 7.32	100.0 +/- 0.0	99.97 +/- 0.17	99.5 +/- 1.46
<i>COG7</i>	53.44 +/- 7.55	100.0 +/- 0.0	100.0 +/- 0.0	99.31 +/- 2.05
<i>COG8</i>	51.73 +/- 7.89	100.0 +/- 0.0	99.93 +/- 0.33	98.54 +/- 3.53
<i>COQ4</i>	51.78 +/- 8.16	100.0 +/- 0.0	99.65 +/- 2.12	97.65 +/- 8.57
<i>COQ8A</i>	55.02 +/- 9.11	100.0 +/- 0.0	100.0 +/- 0.0	99.28 +/- 2.55
<i>COX20</i>	53.46 +/- 7.67	100.0 +/- 0.0	99.9 +/- 0.69	99.04 +/- 3.19
<i>CP</i>	54.85 +/- 7.93	100.0 +/- 0.0	99.99 +/- 0.06	99.46 +/- 0.98
<i>CPLANE1</i>	55.19 +/- 7.49	100.0 +/- 0.0	99.98 +/- 0.08	99.5 +/- 0.87
<i>CPT1C</i>	47.62 +/- 8.44	100.0 +/- 0.0	99.76 +/- 1.26	95.56 +/- 11.14
<i>CRB2</i>	51.5 +/- 8.23	99.95 +/- 0.05	99.88 +/- 0.24	97.72 +/- 5.88
<i>CRPPA</i>	54.79 +/- 7.38	99.99 +/- 0.03	99.89 +/- 0.48	99.09 +/- 2.38
<i>CSGALNACT1</i>	53.51 +/- 8.14	100.0 +/- 0.0	99.96 +/- 0.16	98.81 +/- 2.6
<i>CSPP1</i>	54.76 +/- 7.34	100.0 +/- 0.0	100.0 +/- 0.0	99.43 +/- 0.86
<i>CSTB</i>	54.12 +/- 8.48	100.0 +/- 0.0	99.71 +/- 1.67	97.4 +/- 6.07
<i>CTBP1</i>	53.86 +/- 8.57	99.95 +/- 0.27	99.16 +/- 1.86	95.22 +/- 5.92
<i>CTNNB1</i>	55.04 +/- 7.68	100.0 +/- 0.0	100.0 +/- 0.03	99.09 +/- 1.44
<i>CWF19L1</i>	53.45 +/- 8.56	100.0 +/- 0.0	100.0 +/- 0.0	98.97 +/- 3.34
<i>CYP27A1</i>	51.99 +/- 8.71	100.0 +/- 0.0	99.86 +/- 0.92	98.15 +/- 6.72
<i>CYP2U1</i>	53.26 +/- 7.53	100.0 +/- 0.0	99.99 +/- 0.05	98.78 +/- 3.06
<i>CYP7B1</i>	54.37 +/- 7.69	100.0 +/- 0.0	100.0 +/- 0.0	99.36 +/- 1.84
<i>DAB1</i>	53.94 +/- 7.43	100.0 +/- 0.0	99.94 +/- 0.23	99.14 +/- 1.66
<i>DAG1</i>	54.41 +/- 8.0	100.0 +/- 0.0	100.0 +/- 0.0	99.27 +/- 2.62
<i>DARS1</i>	54.82 +/- 7.28	100.0 +/- 0.0	100.0 +/- 0.01	99.74 +/- 0.79
<i>DARS2</i>	53.0 +/- 7.67	100.0 +/- 0.0	99.98 +/- 0.12	99.09 +/- 2.37
<i>DCC</i>	54.14 +/- 7.33	100.0 +/- 0.0	99.92 +/- 0.21	98.98 +/- 1.67
<i>DDHD1</i>	54.01 +/- 7.84	100.0 +/- 0.0	99.94 +/- 0.24	98.89 +/- 2.15
<i>DDHD2</i>	55.06 +/- 7.7	100.0 +/- 0.0	99.95 +/- 0.18	99.32 +/- 1.64
<i>DDOST</i>	51.9 +/- 8.2	99.91 +/- 0.19	99.88 +/- 0.24	98.49 +/- 4.37
<i>DDX3X</i>	43.37 +/- 16.0	99.96 +/- 0.19	95.67 +/- 8.3	71.16 +/- 34.63

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>DDX59</i>	52.36 +/- 7.29	100.0 +/- 0.0	99.84 +/- 0.68	98.54 +/- 2.78
<i>DHCR7</i>	53.79 +/- 9.03	99.97 +/- 0.12	99.79 +/- 0.86	98.57 +/- 5.94
<i>DHDDS</i>	51.93 +/- 8.36	100.0 +/- 0.0	100.0 +/- 0.0	98.46 +/- 4.24
<i>DKC1</i>	42.52 +/- 16.09	99.94 +/- 0.32	93.3 +/- 13.12	69.97 +/- 35.49
<i>DMXL2</i>	55.67 +/- 7.64	100.0 +/- 0.0	100.0 +/- 0.01	99.54 +/- 1.34
<i>DNAI3</i>	55.25 +/- 7.49	100.0 +/- 0.0	99.98 +/- 0.1	99.69 +/- 0.97
<i>DNAJC19</i>	54.85 +/- 7.36	100.0 +/- 0.0	100.0 +/- 0.0	98.86 +/- 2.88
<i>DNAJC5</i>	54.07 +/- 8.61	100.0 +/- 0.0	99.9 +/- 0.46	98.54 +/- 4.57
<i>DNMT1</i>	54.52 +/- 8.27	100.0 +/- 0.0	99.98 +/- 0.11	99.37 +/- 1.94
<i>DOCK3</i>	52.56 +/- 8.1	100.0 +/- 0.0	99.87 +/- 0.63	98.3 +/- 4.03
<i>DOLK</i>	47.05 +/- 8.6	100.0 +/- 0.0	99.57 +/- 2.64	95.74 +/- 10.02
<i>DPAGT1</i>	51.6 +/- 8.57	100.0 +/- 0.0	99.91 +/- 0.58	98.28 +/- 6.05
<i>DPM1</i>	55.3 +/- 8.03	100.0 +/- 0.0	100.0 +/- 0.0	99.73 +/- 1.26
<i>DPM2</i>	48.79 +/- 8.39	100.0 +/- 0.0	99.99 +/- 0.08	97.15 +/- 11.75
<i>DPM3</i>	50.7 +/- 8.09	100.0 +/- 0.0	99.82 +/- 1.21	98.89 +/- 3.95
<i>DPYSL5</i>	52.13 +/- 8.4	100.0 +/- 0.0	99.99 +/- 0.06	98.68 +/- 4.42
<i>DSTYK</i>	52.8 +/- 7.84	100.0 +/- 0.0	100.0 +/- 0.03	98.96 +/- 3.66
<i>DYNC1H1</i>	53.26 +/- 7.91	100.0 +/- 0.0	99.97 +/- 0.18	98.9 +/- 3.38
<i>EBF3</i>	54.46 +/- 8.19	100.0 +/- 0.0	99.9 +/- 0.67	98.31 +/- 3.95
<i>EDEM3</i>	55.22 +/- 7.47	100.0 +/- 0.0	99.98 +/- 0.06	99.07 +/- 1.26
<i>EEF2</i>	54.66 +/- 8.94	100.0 +/- 0.0	99.99 +/- 0.06	98.71 +/- 6.1
<i>EIF2B1</i>	51.35 +/- 7.56	99.83 +/- 0.32	99.28 +/- 0.81	96.39 +/- 4.09
<i>EIF2B2</i>	53.36 +/- 7.69	100.0 +/- 0.0	99.98 +/- 0.06	99.07 +/- 2.4
<i>EIF2B3</i>	51.31 +/- 7.76	100.0 +/- 0.0	99.9 +/- 0.69	98.38 +/- 4.44
<i>EIF2B4</i>	52.54 +/- 8.45	100.0 +/- 0.0	99.97 +/- 0.22	99.29 +/- 3.39
<i>EIF2B5</i>	51.46 +/- 8.19	99.97 +/- 0.05	99.94 +/- 0.14	99.03 +/- 3.31
<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>ELOVL5</i>	55.02 +/- 7.65	100.0 +/- 0.0	100.0 +/- 0.0	99.71 +/- 0.97
<i>ENTPD1</i>	54.45 +/- 8.02	100.0 +/- 0.0	99.98 +/- 0.12	99.14 +/- 2.5
<i>EOGT</i>	55.69 +/- 7.68	100.0 +/- 0.0	100.0 +/- 0.0	99.48 +/- 1.39
<i>EPM2A</i>	53.46 +/- 7.4	99.96 +/- 0.27	99.47 +/- 1.35	96.81 +/- 3.23
<i>ERCC4</i>	53.69 +/- 7.57	100.0 +/- 0.0	99.99 +/- 0.04	99.22 +/- 1.61
<i>ERLIN1</i>	53.02 +/- 7.89	100.0 +/- 0.0	99.94 +/- 0.34	98.85 +/- 3.13
<i>ERLIN2</i>	52.79 +/- 8.04	99.99 +/- 0.07	99.69 +/- 0.54	97.65 +/- 3.24
<i>EVC</i>	51.23 +/- 8.25	99.99 +/- 0.09	99.71 +/- 0.94	96.84 +/- 5.85
<i>EVC2</i>	52.51 +/- 7.89	100.0 +/- 0.0	99.89 +/- 0.54	98.29 +/- 5.23
<i>EXOC3L2</i>	48.14 +/- 8.27	99.99 +/- 0.03	99.86 +/- 0.41	96.1 +/- 8.33
<i>EXOC8</i>	53.22 +/- 7.24	100.0 +/- 0.0	99.99 +/- 0.06	98.92 +/- 3.01
<i>EXOSC1</i>	55.96 +/- 8.36	100.0 +/- 0.0	99.96 +/- 0.25	99.36 +/- 1.87
<i>EXOSC3</i>	53.66 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.0	99.33 +/- 2.05
<i>EXOSC5</i>	52.0 +/- 8.01	100.0 +/- 0.0	100.0 +/- 0.0	98.08 +/- 6.23
<i>EXOSC8</i>	55.21 +/- 7.44	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 1.26
<i>EXOSC9</i>	54.6 +/- 7.36	100.0 +/- 0.0	100.0 +/- 0.0	99.61 +/- 1.17
<i>EXT1</i>	52.25 +/- 7.63	100.0 +/- 0.0	99.91 +/- 0.19	98.56 +/- 2.6
<i>EXT2</i>	54.18 +/- 8.18	100.0 +/- 0.0	99.98 +/- 0.06	99.22 +/- 2.38

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>FA2H</i>	49.37 +/- 8.86	100.0 +/- 0.0	99.78 +/- 1.27	97.02 +/- 6.84
<i>FAM149B1</i>	54.76 +/- 7.56	100.0 +/- 0.0	100.0 +/- 0.02	99.3 +/- 1.6
<i>FAR1</i>	56.36 +/- 7.12	100.0 +/- 0.01	99.99 +/- 0.04	99.64 +/- 0.75
<i>FARS2</i>	53.78 +/- 8.03	100.0 +/- 0.0	99.98 +/- 0.07	98.88 +/- 3.7
<i>FBXL4</i>	55.91 +/- 7.3	100.0 +/- 0.0	100.0 +/- 0.03	99.58 +/- 0.98
<i>FBXO7</i>	52.32 +/- 7.96	100.0 +/- 0.0	99.94 +/- 0.42	98.82 +/- 2.4
<i>FCSK</i>	52.64 +/- 8.2	100.0 +/- 0.0	99.97 +/- 0.14	98.79 +/- 4.73
<i>FGF14</i>	55.05 +/- 7.82	100.0 +/- 0.0	100.0 +/- 0.03	99.43 +/- 1.27
<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>FLVCR1</i>	54.95 +/- 7.43	99.83 +/- 0.23	99.66 +/- 0.39	98.6 +/- 2.83
<i>FMR1</i>	42.17 +/- 15.12	99.72 +/- 0.98	94.07 +/- 10.43	70.04 +/- 34.02
<i>FOLR1</i>	50.63 +/- 9.04	100.0 +/- 0.0	100.0 +/- 0.0	98.91 +/- 4.71
<i>FRMD4A</i>	52.74 +/- 7.96	100.0 +/- 0.0	99.99 +/- 0.03	98.96 +/- 2.57
<i>FRMD5</i>	54.31 +/- 8.17	99.99 +/- 0.04	99.47 +/- 0.92	97.46 +/- 3.18
<i>FUT8</i>	53.66 +/- 7.97	100.0 +/- 0.0	99.98 +/- 0.15	98.83 +/- 3.4
<i>FXN</i>	53.72 +/- 7.79	99.99 +/- 0.03	99.96 +/- 0.14	99.15 +/- 2.07
<i>G6PC3</i>	50.93 +/- 8.98	100.0 +/- 0.0	99.78 +/- 1.37	97.89 +/- 6.54
<i>GAD1</i>	53.19 +/- 7.72	100.0 +/- 0.0	100.0 +/- 0.03	98.98 +/- 2.05
<i>GALC</i>	56.77 +/- 8.19	99.97 +/- 0.05	99.96 +/- 0.05	99.54 +/- 1.38
<i>GALNT12</i>	52.56 +/- 7.43	100.0 +/- 0.0	99.86 +/- 0.5	98.16 +/- 3.53
<i>GALNT2</i>	52.01 +/- 8.08	100.0 +/- 0.0	99.92 +/- 0.48	98.0 +/- 3.84
<i>GALNT3</i>	55.35 +/- 7.5	100.0 +/- 0.0	100.0 +/- 0.0	99.36 +/- 1.49
<i>GBA2</i>	50.09 +/- 8.82	100.0 +/- 0.0	99.77 +/- 1.07	96.91 +/- 7.5
<i>GBE1</i>	55.41 +/- 7.39	100.0 +/- 0.0	100.0 +/- 0.0	99.5 +/- 1.34
<i>GCH1</i>	55.33 +/- 7.23	100.0 +/- 0.0	100.0 +/- 0.0	99.45 +/- 1.69
<i>GDAP2</i>	55.34 +/- 7.62	100.0 +/- 0.0	99.98 +/- 0.12	99.54 +/- 1.27
<i>GEMIN5</i>	54.71 +/- 7.72	100.0 +/- 0.0	100.0 +/- 0.0	99.33 +/- 2.15
<i>GFAP</i>	50.43 +/- 8.48	100.0 +/- 0.0	99.97 +/- 0.14	98.13 +/- 6.69
<i>GFPT1</i>	55.31 +/- 7.17	100.0 +/- 0.0	99.99 +/- 0.05	99.52 +/- 1.12
<i>GJA1</i>	53.43 +/- 7.59	100.0 +/- 0.0	99.99 +/- 0.03	99.14 +/- 1.51
<i>GJC2</i>	51.68 +/- 10.29	100.0 +/- 0.0	99.85 +/- 0.35	95.11 +/- 7.27
<i>GLI3</i>	54.82 +/- 7.78	100.0 +/- 0.0	99.99 +/- 0.06	99.24 +/- 2.65
<i>GLRA1</i>	50.56 +/- 8.67	100.0 +/- 0.0	99.99 +/- 0.06	98.39 +/- 4.72
<i>GLRB</i>	55.35 +/- 6.91	100.0 +/- 0.0	99.99 +/- 0.07	99.29 +/- 1.3
<i>GLRX5</i>	51.54 +/- 7.98	100.0 +/- 0.0	99.99 +/- 0.07	99.22 +/- 2.12
<i>GLS</i>	53.73 +/- 6.97	99.9 +/- 0.18	99.84 +/- 0.23	98.72 +/- 2.18
<i>GMPPA</i>	49.73 +/- 8.67	100.0 +/- 0.0	99.86 +/- 0.85	96.92 +/- 7.93
<i>GMPPB</i>	51.47 +/- 8.43	100.0 +/- 0.0	99.9 +/- 0.69	98.18 +/- 6.13
<i>GNE</i>	54.83 +/- 7.68	99.98 +/- 0.04	99.98 +/- 0.04	99.4 +/- 1.23
<i>GORAB</i>	55.03 +/- 7.75	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.39
<i>GOSR2</i>	53.05 +/- 7.86	100.0 +/- 0.0	99.95 +/- 0.36	98.96 +/- 4.05
<i>GPAA1</i>	52.91 +/- 9.18	100.0 +/- 0.0	99.62 +/- 1.89	97.52 +/- 8.82
<i>GPT2</i>	53.52 +/- 7.8	100.0 +/- 0.0	99.97 +/- 0.15	98.74 +/- 4.24
<i>GRID2</i>	54.07 +/- 7.55	100.0 +/- 0.0	100.0 +/- 0.03	99.24 +/- 2.06
<i>GRM1</i>	54.15 +/- 7.82	100.0 +/- 0.0	99.98 +/- 0.12	98.9 +/- 3.2

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>GRN</i>	53.75 +/- 8.59	100.0 +/- 0.0	100.0 +/- 0.0	98.96 +/- 3.52
<i>HACE1</i>	55.4 +/- 7.36	100.0 +/- 0.0	99.92 +/- 0.46	99.24 +/- 2.12
<i>HARS1</i>	52.54 +/- 8.15	100.0 +/- 0.0	99.96 +/- 0.28	98.46 +/- 6.7
<i>HEATR5B</i>	55.45 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.49 +/- 1.42
<i>HEXA</i>	52.08 +/- 7.91	100.0 +/- 0.0	99.96 +/- 0.2	98.58 +/- 3.93
<i>HEXB</i>	53.14 +/- 7.15	99.99 +/- 0.03	99.99 +/- 0.03	98.56 +/- 3.25
<i>HIKESHI</i>	56.17 +/- 7.45	100.0 +/- 0.0	100.0 +/- 0.0	98.83 +/- 3.34
<i>HPDL</i>	53.8 +/- 8.59	100.0 +/- 0.0	99.98 +/- 0.15	98.98 +/- 5.39
<i>HSPD1</i>	54.23 +/- 7.93	100.0 +/- 0.0	99.99 +/- 0.07	99.05 +/- 3.01
<i>HTT</i>	54.07 +/- 7.98	100.0 +/- 0.0	99.97 +/- 0.11	98.95 +/- 2.52
<i>HYLS1</i>	55.02 +/- 8.28	100.0 +/- 0.0	100.0 +/- 0.0	99.51 +/- 1.84
<i>IBA57</i>	52.76 +/- 7.84	99.99 +/- 0.03	99.96 +/- 0.12	98.77 +/- 3.74
<i>IFIH1</i>	54.43 +/- 7.58	100.0 +/- 0.0	99.96 +/- 0.27	99.44 +/- 1.67
<i>INPP5E</i>	50.81 +/- 8.98	100.0 +/- 0.0	99.5 +/- 2.37	95.13 +/- 8.84
<i>IRF2BPL</i>	50.79 +/- 8.4	99.94 +/- 0.23	99.28 +/- 1.37	94.26 +/- 9.05
<i>ITPR1</i>	54.07 +/- 7.96	100.0 +/- 0.0	99.96 +/- 0.2	98.72 +/- 2.75
<i>KATNIP</i>	52.99 +/- 8.06	100.0 +/- 0.0	100.0 +/- 0.03	99.12 +/- 3.37
<i>KCNA1</i>	52.15 +/- 7.78	100.0 +/- 0.0	99.87 +/- 0.71	98.16 +/- 4.67
<i>KCNA2</i>	52.74 +/- 7.88	100.0 +/- 0.0	99.99 +/- 0.05	99.16 +/- 2.6
<i>KCNC3</i>	41.3 +/- 7.4	99.74 +/- 0.58	95.06 +/- 4.63	81.55 +/- 13.55
<i>KCND3</i>	51.86 +/- 8.09	100.0 +/- 0.0	99.88 +/- 0.3	98.01 +/- 4.26
<i>KCNJ10</i>	52.91 +/- 8.64	100.0 +/- 0.0	99.98 +/- 0.08	98.58 +/- 3.18
<i>KCNK18</i>	52.62 +/- 8.32	100.0 +/- 0.0	100.0 +/- 0.0	98.6 +/- 6.57
<i>KCNN2</i>	52.18 +/- 7.9	100.0 +/- 0.0	99.99 +/- 0.06	98.43 +/- 3.67
<i>KCNQ2</i>	52.18 +/- 9.05	99.99 +/- 0.06	99.75 +/- 0.93	96.65 +/- 7.32
<i>KCNQ3</i>	53.58 +/- 7.82	99.98 +/- 0.09	99.6 +/- 0.87	97.9 +/- 3.33
<i>KDM5C</i>	38.92 +/- 15.26	99.17 +/- 1.45	88.4 +/- 15.93	62.4 +/- 39.38
<i>KIAA0586</i>	55.3 +/- 7.5	100.0 +/- 0.0	99.99 +/- 0.03	99.37 +/- 1.55
<i>KIAA0753</i>	55.17 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.0	99.57 +/- 0.99
<i>KIDINS220</i>	55.9 +/- 7.62	100.0 +/- 0.0	100.0 +/- 0.01	99.41 +/- 1.56
<i>KIF14</i>	55.53 +/- 7.13	99.94 +/- 0.1	99.85 +/- 0.12	99.5 +/- 0.94
<i>KIF1A</i>	51.48 +/- 8.49	100.0 +/- 0.0	99.96 +/- 0.19	98.05 +/- 5.34
<i>KIF1C</i>	51.33 +/- 7.68	99.98 +/- 0.1	99.84 +/- 0.57	97.72 +/- 4.89
<i>KIF5A</i>	50.99 +/- 7.72	100.0 +/- 0.0	99.98 +/- 0.11	98.48 +/- 3.42
<i>KIF7</i>	52.39 +/- 8.25	100.0 +/- 0.0	99.88 +/- 0.79	97.97 +/- 4.36
<i>KLC2</i>	49.41 +/- 9.06	100.0 +/- 0.0	99.85 +/- 0.51	96.46 +/- 9.17
<i>KLC4</i>	51.17 +/- 8.64	100.0 +/- 0.0	99.87 +/- 0.62	97.88 +/- 7.1
<i>KPNA3</i>	54.74 +/- 7.02	99.96 +/- 0.3	99.34 +/- 1.11	97.27 +/- 2.66
<i>L1CAM</i>	39.15 +/- 15.32	99.72 +/- 1.05	89.66 +/- 16.26	63.97 +/- 39.82
<i>LAMA1</i>	55.01 +/- 7.48	100.0 +/- 0.0	100.0 +/- 0.0	99.52 +/- 1.26
<i>LARGE1</i>	51.83 +/- 7.71	100.0 +/- 0.0	99.84 +/- 0.6	97.9 +/- 4.14
<i>LARS2</i>	52.82 +/- 8.22	100.0 +/- 0.0	100.0 +/- 0.01	99.02 +/- 2.91
<i>LFNG</i>	49.29 +/- 9.01	99.99 +/- 0.04	98.6 +/- 2.74	92.53 +/- 9.68
<i>LIG3</i>	52.98 +/- 7.94	100.0 +/- 0.0	99.97 +/- 0.09	98.6 +/- 3.19
<i>LNPK</i>	54.49 +/- 8.05	100.0 +/- 0.03	100.0 +/- 0.03	99.26 +/- 1.52
<i>LYST</i>	55.56 +/- 7.43	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.01
<i>MAG</i>	49.42 +/- 8.48	99.99 +/- 0.04	99.64 +/- 0.76	96.19 +/- 5.68

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>MAGT1</i>	43.44 +/- 16.57	99.94 +/- 0.22	94.08 +/- 10.56	71.02 +/- 34.29
<i>MAN1B1</i>	54.96 +/- 9.04	100.0 +/- 0.0	99.93 +/- 0.45	98.81 +/- 5.33
<i>MAPK8IP3</i>	53.68 +/- 9.22	100.0 +/- 0.0	99.92 +/- 0.49	98.25 +/- 6.22
<i>MARS1</i>	52.63 +/- 8.08	100.0 +/- 0.0	100.0 +/- 0.03	99.09 +/- 3.13
<i>MARS2</i>	55.27 +/- 8.13	100.0 +/- 0.0	99.99 +/- 0.09	99.17 +/- 3.59
<i>MFN2</i>	52.59 +/- 8.41	100.0 +/- 0.0	99.96 +/- 0.22	98.5 +/- 3.53
<i>MGAT2</i>	51.76 +/- 7.35	100.0 +/- 0.0	99.99 +/- 0.07	98.65 +/- 2.58
<i>MINPP1</i>	53.97 +/- 7.81	100.0 +/- 0.0	99.99 +/- 0.09	98.98 +/- 3.36
<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>MME</i>	55.27 +/- 7.85	100.0 +/- 0.0	99.99 +/- 0.06	99.55 +/- 1.42
<i>MOGS</i>	52.95 +/- 9.37	100.0 +/- 0.0	99.96 +/- 0.19	98.28 +/- 5.94
<i>MORC2</i>	50.96 +/- 7.72	100.0 +/- 0.0	99.98 +/- 0.09	98.41 +/- 3.48
<i>MPDU1</i>	46.09 +/- 8.79	100.0 +/- 0.0	99.73 +/- 1.27	93.79 +/- 10.9
<i>MPI</i>	54.14 +/- 8.49	100.0 +/- 0.0	99.97 +/- 0.19	98.98 +/- 4.19
<i>MRE11</i>	54.65 +/- 7.47	100.0 +/- 0.0	100.0 +/- 0.0	99.47 +/- 1.44
<i>MSTO1</i>	40.98 +/- 6.05	99.91 +/- 0.26	94.8 +/- 5.32	77.79 +/- 12.42
<i>MTCL1</i>	50.74 +/- 7.72	100.0 +/- 0.01	99.47 +/- 1.76	95.46 +/- 7.49
<i>MTFMT</i>	54.98 +/- 8.22	100.0 +/- 0.0	99.98 +/- 0.13	98.88 +/- 2.91
<i>MTPAP</i>	54.43 +/- 7.45	100.0 +/- 0.0	99.96 +/- 0.17	99.03 +/- 2.33
<i>MTRFR</i>	54.41 +/- 8.59	100.0 +/- 0.0	99.98 +/- 0.12	99.3 +/- 1.67
<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>NAGLU</i>	53.56 +/- 8.47	100.0 +/- 0.0	99.96 +/- 0.3	98.55 +/- 4.21
<i>NAXE</i>	51.04 +/- 8.73	100.0 +/- 0.0	99.96 +/- 0.22	97.5 +/- 6.27
<i>NDUFA12</i>	52.71 +/- 8.99	100.0 +/- 0.0	99.93 +/- 0.48	98.66 +/- 3.86
<i>NFASC</i>	51.97 +/- 8.82	100.0 +/- 0.0	99.94 +/- 0.26	98.62 +/- 4.54
<i>NGLY1</i>	56.28 +/- 7.24	100.0 +/- 0.0	99.98 +/- 0.12	98.99 +/- 2.4
<i>NHLRC1</i>	54.06 +/- 8.27	100.0 +/- 0.0	99.99 +/- 0.09	99.43 +/- 2.81
<i>NIPA1</i>	54.12 +/- 7.69	99.94 +/- 0.22	99.72 +/- 0.73	98.68 +/- 2.79
<i>NKX2-1</i>	51.81 +/- 7.67	99.99 +/- 0.04	99.97 +/- 0.07	98.01 +/- 5.4
<i>NKX6-2</i>	49.95 +/- 8.84	99.72 +/- 0.87	96.98 +/- 4.2	89.96 +/- 9.52
<i>NMNAT2</i>	50.62 +/- 8.07	99.99 +/- 0.03	99.94 +/- 0.23	98.03 +/- 5.23
<i>NOP56</i>	51.09 +/- 8.25	100.0 +/- 0.0	100.0 +/- 0.01	98.4 +/- 4.18
<i>NPC1</i>	53.64 +/- 7.59	100.0 +/- 0.0	99.95 +/- 0.27	98.96 +/- 3.09
<i>NPC2</i>	51.22 +/- 8.83	100.0 +/- 0.0	99.95 +/- 0.36	98.58 +/- 4.1
<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>NPTX1</i>	52.29 +/- 8.32	100.0 +/- 0.0	99.82 +/- 0.59	97.28 +/- 4.96
<i>NRCAM</i>	56.14 +/- 7.64	100.0 +/- 0.0	99.99 +/- 0.03	99.41 +/- 1.15
<i>NSRP1</i>	52.94 +/- 7.9	100.0 +/- 0.0	100.0 +/- 0.0	99.41 +/- 1.28
<i>NT5C2</i>	55.17 +/- 8.33	100.0 +/- 0.0	99.99 +/- 0.08	99.24 +/- 2.4
<i>NUS1</i>	51.67 +/- 7.28	100.0 +/- 0.0	99.4 +/- 1.16	94.35 +/- 6.2
<i>OFD1</i>	42.18 +/- 15.83	99.93 +/- 0.36	93.98 +/- 9.99	68.58 +/- 36.59
<i>OGDHL</i>	51.22 +/- 8.34	100.0 +/- 0.0	99.88 +/- 0.71	97.89 +/- 6.21
<i>OPA1</i>	56.34 +/- 7.32	100.0 +/- 0.0	100.0 +/- 0.0	99.76 +/- 0.6
<i>OPA3</i>	51.74 +/- 8.39	99.97 +/- 0.04	99.96 +/- 0.06	98.64 +/- 3.21

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>OPHN1</i>	40.76 +/- 15.53	98.71 +/- 1.63	90.34 +/- 11.02	64.67 +/- 35.52
<i>OSTC</i>	56.56 +/- 8.81	100.0 +/- 0.0	99.92 +/- 0.52	99.69 +/- 1.2
<i>PACS2</i>	51.75 +/- 9.07	99.99 +/- 0.04	99.64 +/- 0.96	96.54 +/- 5.95
<i>PAX2</i>	45.55 +/- 7.82	100.0 +/- 0.01	99.64 +/- 1.68	93.13 +/- 12.12
<i>PAX6</i>	52.35 +/- 7.12	100.0 +/- 0.0	99.97 +/- 0.13	98.35 +/- 3.15
<i>PCDH12</i>	52.52 +/- 8.25	100.0 +/- 0.0	99.98 +/- 0.09	98.67 +/- 4.88
<i>PCLO</i>	52.5 +/- 7.17	100.0 +/- 0.02	99.74 +/- 0.29	98.3 +/- 2.44
<i>PCYT2</i>	53.5 +/- 8.87	100.0 +/- 0.0	99.88 +/- 0.44	97.99 +/- 4.94
<i>PDE6D</i>	51.83 +/- 8.41	100.0 +/- 0.0	99.97 +/- 0.18	98.2 +/- 4.37
<i>PDYN</i>	51.47 +/- 8.85	100.0 +/- 0.0	100.0 +/- 0.0	98.61 +/- 4.33
<i>PEX16</i>	51.69 +/- 9.17	100.0 +/- 0.0	99.96 +/- 0.3	98.06 +/- 4.43
<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>PGAP1</i>	55.75 +/- 7.66	100.0 +/- 0.0	99.98 +/- 0.09	99.5 +/- 1.0
<i>PGAP2</i>	50.81 +/- 9.5	100.0 +/- 0.03	99.98 +/- 0.09	98.07 +/- 5.92
<i>PGAP3</i>	50.71 +/- 8.17	100.0 +/- 0.0	99.93 +/- 0.33	98.17 +/- 6.7
<i>PGM1</i>	51.79 +/- 7.83	100.0 +/- 0.0	99.97 +/- 0.14	98.28 +/- 6.06
<i>PGM3</i>	54.72 +/- 7.6	100.0 +/- 0.0	99.98 +/- 0.12	99.51 +/- 0.98
<i>PHGDH</i>	52.27 +/- 8.44	100.0 +/- 0.0	100.0 +/- 0.0	98.5 +/- 6.02
<i>PI4KA</i>	52.04 +/- 7.84	99.98 +/- 0.13	99.54 +/- 1.23	97.5 +/- 4.14
<i>PIBF1</i>	55.04 +/- 7.25	100.0 +/- 0.0	99.96 +/- 0.22	99.29 +/- 1.59
<i>PIGA</i>	43.57 +/- 15.89	99.98 +/- 0.1	95.37 +/- 9.92	71.86 +/- 34.18
<i>PIGL</i>	53.93 +/- 7.71	100.0 +/- 0.0	100.0 +/- 0.0	99.23 +/- 3.17
<i>PIGM</i>	53.39 +/- 8.32	100.0 +/- 0.0	99.96 +/- 0.22	98.92 +/- 2.96
<i>PIGN</i>	54.9 +/- 7.58	100.0 +/- 0.0	99.97 +/- 0.15	99.19 +/- 1.65
<i>PIGO</i>	53.27 +/- 8.19	100.0 +/- 0.0	100.0 +/- 0.0	98.94 +/- 3.16
<i>PIGS</i>	52.29 +/- 7.9	100.0 +/- 0.0	100.0 +/- 0.0	99.12 +/- 2.56
<i>PIGT</i>	50.65 +/- 8.58	100.0 +/- 0.0	99.96 +/- 0.27	97.76 +/- 5.86
<i>PIGV</i>	53.84 +/- 7.7	100.0 +/- 0.0	99.95 +/- 0.29	99.36 +/- 2.0
<i>PIGW</i>	55.02 +/- 7.58	100.0 +/- 0.0	99.99 +/- 0.06	99.38 +/- 2.64
<i>PIK3R5</i>	52.01 +/- 8.17	100.0 +/- 0.0	99.96 +/- 0.24	98.46 +/- 5.13
<i>PITRM1</i>	55.92 +/- 7.77	100.0 +/- 0.0	100.0 +/- 0.0	99.51 +/- 1.26
<i>PLA2G6</i>	50.91 +/- 9.79	100.0 +/- 0.0	99.94 +/- 0.2	97.08 +/- 5.68
<i>PLP1</i>	43.22 +/- 16.2	99.9 +/- 0.54	94.53 +/- 8.67	70.22 +/- 34.85
<i>PMM2</i>	53.79 +/- 8.67	100.0 +/- 0.0	99.87 +/- 0.85	98.37 +/- 4.98
<i>PMPCA</i>	55.8 +/- 8.6	100.0 +/- 0.0	99.99 +/- 0.07	99.33 +/- 2.73
<i>PMPCB</i>	55.77 +/- 8.01	100.0 +/- 0.01	100.0 +/- 0.01	99.59 +/- 1.06
<i>PNKD</i>	50.76 +/- 8.02	100.0 +/- 0.0	99.94 +/- 0.31	98.03 +/- 6.31
<i>PNKP</i>	51.37 +/- 8.13	100.0 +/- 0.0	100.0 +/- 0.0	98.05 +/- 3.57
<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>POLG</i>	52.88 +/- 8.23	100.0 +/- 0.0	99.94 +/- 0.37	98.88 +/- 3.87
<i>POLG2</i>	52.33 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.0	99.1 +/- 2.14
<i>POLR3A</i>	54.13 +/- 7.99	100.0 +/- 0.0	99.97 +/- 0.19	99.29 +/- 2.62
<i>POLR3B</i>	54.38 +/- 7.41	100.0 +/- 0.0	99.99 +/- 0.09	99.52 +/- 1.92
<i>POLR3K</i>	53.52 +/- 8.88	100.0 +/- 0.01	99.98 +/- 0.06	98.86 +/- 5.07

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<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>POMK</i>	53.3 +/- 8.89	100.0 +/- 0.0	99.99 +/- 0.07	98.67 +/- 5.37
<i>POMT1</i>	53.59 +/- 8.44	100.0 +/- 0.0	99.98 +/- 0.12	99.07 +/- 3.38
<i>POMT2</i>	49.86 +/- 8.39	99.99 +/- 0.07	99.7 +/- 0.83	96.75 +/- 6.31
<i>POU4F1</i>	47.86 +/- 7.25	100.0 +/- 0.0	99.1 +/- 2.55	93.87 +/- 8.09
<i>PPP2R2B</i>	52.78 +/- 7.79	100.0 +/- 0.0	99.99 +/- 0.03	99.1 +/- 2.44
<i>PRDM13</i>	52.75 +/- 7.97	99.98 +/- 0.1	99.51 +/- 2.39	97.3 +/- 6.89
<i>PRDX3</i>	54.87 +/- 7.9	100.0 +/- 0.0	100.0 +/- 0.0	99.7 +/- 1.08
<i>PRICKLE1</i>	54.09 +/- 7.84	100.0 +/- 0.0	99.9 +/- 0.48	98.73 +/- 2.36
<i>PRKCG</i>	45.14 +/- 7.86	100.0 +/- 0.01	99.46 +/- 2.64	93.32 +/- 12.08
<i>PRNP</i>	52.52 +/- 8.33	100.0 +/- 0.0	99.96 +/- 0.19	98.74 +/- 2.14
<i>PRRT2</i>	46.27 +/- 7.34	100.0 +/- 0.03	99.83 +/- 0.87	93.99 +/- 10.57
<i>PSEN1</i>	54.65 +/- 8.13	100.0 +/- 0.0	99.98 +/- 0.13	99.51 +/- 1.56
<i>PTF1A</i>	47.31 +/- 8.54	99.98 +/- 0.06	99.45 +/- 1.68	92.15 +/- 13.06
<i>PTRH2</i>	54.04 +/- 9.61	100.0 +/- 0.0	100.0 +/- 0.0	99.14 +/- 3.67
<i>PUM1</i>	53.23 +/- 7.59	100.0 +/- 0.0	99.99 +/- 0.06	99.06 +/- 1.97
<i>RAB3GAP2</i>	55.12 +/- 6.99	100.0 +/- 0.0	99.99 +/- 0.03	99.4 +/- 1.17
<i>RABL2A</i>	45.58 +/- 7.28	99.94 +/- 0.42	98.93 +/- 1.88	90.23 +/- 7.79
<i>RARS2</i>	55.04 +/- 7.76	100.0 +/- 0.0	100.0 +/- 0.01	99.68 +/- 0.8
<i>REEP1</i>	54.27 +/- 7.85	100.0 +/- 0.0	100.0 +/- 0.02	99.08 +/- 2.55
<i>REEP2</i>	48.19 +/- 9.49	99.99 +/- 0.07	99.43 +/- 2.03	93.82 +/- 11.93
<i>RELN</i>	55.84 +/- 7.53	100.0 +/- 0.0	99.99 +/- 0.06	99.42 +/- 1.58
<i>RFC1</i>	54.36 +/- 7.81	100.0 +/- 0.0	100.0 +/- 0.0	99.17 +/- 1.69
<i>RFT1</i>	52.22 +/- 8.12	100.0 +/- 0.0	99.99 +/- 0.04	98.85 +/- 3.38
<i>RNASEH2B</i>	53.33 +/- 7.21	100.0 +/- 0.0	99.99 +/- 0.06	98.92 +/- 3.59
<i>RNF170</i>	54.14 +/- 7.78	100.0 +/- 0.0	99.96 +/- 0.23	99.3 +/- 2.48
<i>RNF216</i>	54.31 +/- 8.06	100.0 +/- 0.0	99.97 +/- 0.15	99.07 +/- 3.2
<i>RNF220</i>	50.97 +/- 9.18	100.0 +/- 0.0	99.91 +/- 0.38	98.22 +/- 4.89
<i>RNU7-1</i>	55.29 +/- 10.66	100.0 +/- 0.0	100.0 +/- 0.0	100.0 +/- 0.0
<i>ROBO3</i>	49.39 +/- 8.22	100.0 +/- 0.01	99.88 +/- 0.58	96.77 +/- 9.67
<i>RORA</i>	55.52 +/- 7.56	100.0 +/- 0.0	99.94 +/- 0.2	99.28 +/- 0.97
<i>RPGRIP1L</i>	54.65 +/- 7.42	100.0 +/- 0.0	100.0 +/- 0.0	99.66 +/- 0.88
<i>RTN2</i>	49.43 +/- 8.23	99.98 +/- 0.07	99.73 +/- 0.94	96.16 +/- 7.13
<i>RUBCN</i>	52.53 +/- 7.86	100.0 +/- 0.0	99.92 +/- 0.33	98.34 +/- 4.35
<i>RXYLT1</i>	55.97 +/- 7.84	100.0 +/- 0.0	99.94 +/- 0.39	98.8 +/- 2.54
<i>SACS</i>	54.72 +/- 7.62	100.0 +/- 0.0	99.98 +/- 0.12	99.5 +/- 1.27
<i>SAMD9L</i>	54.91 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.01	99.49 +/- 1.07
<i>SAR1B</i>	55.42 +/- 7.96	100.0 +/- 0.0	99.97 +/- 0.12	99.42 +/- 0.98
<i>SARS2</i>	49.15 +/- 8.09	100.0 +/- 0.0	99.95 +/- 0.25	97.5 +/- 6.54
<i>SCLT1</i>	55.31 +/- 7.5	100.0 +/- 0.0	99.99 +/- 0.05	99.48 +/- 1.54
<i>SCN1A</i>	54.78 +/- 7.63	100.0 +/- 0.01	99.99 +/- 0.03	99.48 +/- 1.2
<i>SCN2A</i>	55.66 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.3
<i>SCN8A</i>	53.15 +/- 7.81	99.99 +/- 0.04	99.82 +/- 0.25	98.45 +/- 2.45
<i>SCN9A</i>	53.76 +/- 7.52	99.98 +/- 0.04	99.97 +/- 0.05	98.87 +/- 1.69
<i>SCYL1</i>	50.53 +/- 8.89	100.0 +/- 0.0	99.49 +/- 2.22	95.46 +/- 9.31

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>SEC23B</i>	54.56 +/- 7.57	100.0 +/- 0.0	100.0 +/- 0.0	99.61 +/- 1.12
<i>SEPSECS</i>	56.23 +/- 7.66	100.0 +/- 0.0	100.0 +/- 0.0	99.64 +/- 1.06
<i>SERAC1</i>	54.64 +/- 8.06	100.0 +/- 0.0	99.97 +/- 0.19	99.04 +/- 2.6
<i>SETX</i>	54.66 +/- 7.66	100.0 +/- 0.0	99.98 +/- 0.11	99.36 +/- 1.53
<i>SIL1</i>	51.94 +/- 8.51	100.0 +/- 0.0	99.88 +/- 0.57	98.73 +/- 4.0
<i>SLC16A2</i>	39.8 +/- 14.92	99.81 +/- 1.08	91.57 +/- 13.87	65.85 +/- 38.94
<i>SLC17A5</i>	54.45 +/- 7.94	100.0 +/- 0.0	99.91 +/- 0.41	98.91 +/- 1.97
<i>SLC1A3</i>	54.45 +/- 7.92	100.0 +/- 0.0	100.0 +/- 0.0	99.62 +/- 1.16
<i>SLC1A4</i>	54.8 +/- 8.22	100.0 +/- 0.0	99.89 +/- 0.59	99.03 +/- 3.3
<i>SLC25A15</i>	54.22 +/- 8.02	100.0 +/- 0.0	99.98 +/- 0.12	99.36 +/- 2.3
<i>SLC25A32</i>	55.61 +/- 7.53	99.99 +/- 0.04	99.98 +/- 0.11	99.57 +/- 1.49
<i>SLC25A46</i>	54.42 +/- 7.38	100.0 +/- 0.0	99.99 +/- 0.07	99.43 +/- 1.22
<i>SLC2A1</i>	51.73 +/- 8.42	100.0 +/- 0.0	100.0 +/- 0.01	98.8 +/- 2.86
<i>SLC33A1</i>	52.9 +/- 7.75	100.0 +/- 0.0	99.94 +/- 0.14	98.0 +/- 3.25
<i>SLC35A1</i>	55.19 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.2
<i>SLC35A2</i>	40.93 +/- 16.01	99.86 +/- 0.82	92.55 +/- 13.29	66.68 +/- 40.05
<i>SLC35A3</i>	54.74 +/- 7.64	100.0 +/- 0.0	99.99 +/- 0.04	99.2 +/- 1.68
<i>SLC35C1</i>	50.94 +/- 8.52	99.98 +/- 0.16	99.51 +/- 1.12	95.7 +/- 8.39
<i>SLC35D1</i>	54.24 +/- 8.12	100.0 +/- 0.0	99.99 +/- 0.06	99.11 +/- 3.06
<i>SLC37A4</i>	52.3 +/- 8.85	100.0 +/- 0.0	99.87 +/- 0.74	98.29 +/- 5.73
<i>SLC39A8</i>	55.07 +/- 8.13	100.0 +/- 0.0	99.96 +/- 0.17	99.0 +/- 2.48
<i>SLC44A1</i>	55.17 +/- 7.63	99.99 +/- 0.03	99.78 +/- 0.48	98.83 +/- 1.74
<i>SLC52A2</i>	53.91 +/- 9.94	99.99 +/- 0.04	99.72 +/- 1.85	97.97 +/- 7.75
<i>SLC6A5</i>	52.73 +/- 7.33	100.0 +/- 0.0	99.97 +/- 0.06	98.98 +/- 2.42
<i>SLC9A1</i>	49.05 +/- 8.09	100.0 +/- 0.0	99.86 +/- 0.91	97.27 +/- 8.44
<i>SLC9A6</i>	43.48 +/- 15.85	100.0 +/- 0.01	95.4 +/- 8.11	71.99 +/- 33.03
<i>SMN1</i> ^{1,2,3}	10.73 +/- 2.39	39.45 +/- 8.02	19.85 +/- 6.67	9.9 +/- 5.05
<i>SMN2</i> ^{1,2,3}	11.91 +/- 4.38	47.75 +/- 14.45	25.07 +/- 15.7	10.18 +/- 9.46
<i>SMPD4</i>	51.71 +/- 8.32	100.0 +/- 0.0	99.88 +/- 0.58	98.0 +/- 4.18
<i>SNAP25</i>	54.6 +/- 7.87	100.0 +/- 0.0	99.94 +/- 0.42	99.24 +/- 2.55
<i>SNX14</i>	55.65 +/- 7.31	100.0 +/- 0.0	100.0 +/- 0.0	99.57 +/- 1.12
<i>SPART</i>	54.81 +/- 7.63	99.99 +/- 0.03	99.91 +/- 0.27	99.09 +/- 1.96
<i>SPAST</i>	53.37 +/- 6.62	100.0 +/- 0.0	99.87 +/- 0.39	98.11 +/- 2.89
<i>SPG11</i>	54.86 +/- 7.79	100.0 +/- 0.0	99.99 +/- 0.05	99.49 +/- 1.56
<i>SPG21</i>	53.43 +/- 7.54	100.0 +/- 0.0	100.0 +/- 0.0	98.45 +/- 2.71
<i>SPG7</i>	52.89 +/- 8.01	100.0 +/- 0.0	99.93 +/- 0.35	98.32 +/- 3.91
<i>SPR</i>	51.36 +/- 8.93	100.0 +/- 0.0	99.99 +/- 0.06	98.32 +/- 4.99
<i>SPTAN1</i>	52.82 +/- 7.92	100.0 +/- 0.0	99.94 +/- 0.21	98.73 +/- 2.99
<i>SPTBN2</i>	52.19 +/- 8.11	100.0 +/- 0.0	99.87 +/- 0.57	97.91 +/- 4.8
<i>SQSTM1</i>	53.33 +/- 8.71	100.0 +/- 0.0	100.0 +/- 0.0	98.74 +/- 3.55
<i>SRD5A3</i>	54.39 +/- 7.94	99.99 +/- 0.04	99.92 +/- 0.42	99.21 +/- 1.59
<i>SSR3</i>	56.47 +/- 8.38	99.99 +/- 0.04	99.99 +/- 0.07	99.28 +/- 1.15
<i>SSR4</i>	40.45 +/- 15.7	99.92 +/- 0.57	89.75 +/- 17.6	67.45 +/- 39.54
<i>ST3GAL3</i>	52.92 +/- 8.78	100.0 +/- 0.0	99.92 +/- 0.55	98.5 +/- 4.52
<i>ST3GAL5</i>	54.53 +/- 7.85	100.0 +/- 0.0	99.93 +/- 0.29	98.82 +/- 2.96
<i>STN1</i>	54.12 +/- 8.47	100.0 +/- 0.0	99.99 +/- 0.04	99.28 +/- 2.46

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>STT3A</i>	54.09 +/- 7.54	100.0 +/- 0.0	100.0 +/- 0.0	99.43 +/- 1.41
<i>STT3B</i>	54.0 +/- 7.34	100.0 +/- 0.0	99.91 +/- 0.38	98.41 +/- 2.68
<i>STUB1</i>	54.37 +/- 8.47	100.0 +/- 0.0	99.45 +/- 2.49	96.38 +/- 6.53
<i>SUFU</i>	50.71 +/- 7.85	100.0 +/- 0.0	99.95 +/- 0.28	97.89 +/- 4.64
<i>SVBP</i>	49.23 +/- 7.34	100.0 +/- 0.0	99.91 +/- 0.47	96.33 +/- 7.03
<i>SYNE1</i>	54.65 +/- 7.43	100.0 +/- 0.0	100.0 +/- 0.01	99.5 +/- 1.52
<i>SYNGAP1</i>	45.04 +/- 7.84	99.96 +/- 0.2	98.69 +/- 2.07	89.72 +/- 9.85
<i>SYT14</i>	55.16 +/- 7.2	99.99 +/- 0.03	99.98 +/- 0.07	99.47 +/- 1.01
<i>TAF8</i>	54.69 +/- 8.34	100.0 +/- 0.0	99.98 +/- 0.09	99.29 +/- 2.4
<i>TAPT1</i>	54.5 +/- 7.28	100.0 +/- 0.0	99.88 +/- 0.44	98.78 +/- 2.08
<i>TBC1D23</i>	56.07 +/- 7.9	100.0 +/- 0.0	100.0 +/- 0.0	99.69 +/- 0.96
<i>TBC1D32</i>	55.78 +/- 7.43	100.0 +/- 0.0	100.0 +/- 0.0	99.76 +/- 0.56
<i>TBP</i>	53.72 +/- 7.82	100.0 +/- 0.0	99.99 +/- 0.04	99.03 +/- 2.34
<i>TCTN1</i>	54.38 +/- 7.63	100.0 +/- 0.0	100.0 +/- 0.0	99.28 +/- 1.52
<i>TCTN2</i>	52.87 +/- 7.68	100.0 +/- 0.0	99.96 +/- 0.19	98.81 +/- 2.6
<i>TCTN3</i>	52.45 +/- 8.27	100.0 +/- 0.0	99.99 +/- 0.04	98.78 +/- 3.22
<i>TDP1</i>	55.15 +/- 7.65	100.0 +/- 0.0	99.96 +/- 0.24	99.6 +/- 1.76
<i>TDP2</i>	54.25 +/- 7.57	100.0 +/- 0.0	99.98 +/- 0.12	99.44 +/- 1.86
<i>TECPR2</i>	54.01 +/- 7.69	100.0 +/- 0.0	99.99 +/- 0.04	98.98 +/- 3.97
<i>TERT</i>	57.28 +/- 9.3	100.0 +/- 0.0	100.0 +/- 0.0	99.04 +/- 2.76
<i>TFG</i>	52.99 +/- 7.52	100.0 +/- 0.0	99.92 +/- 0.44	97.85 +/- 3.81
<i>TGM6</i>	50.86 +/- 8.01	100.0 +/- 0.0	99.99 +/- 0.04	98.66 +/- 4.11
<i>THG1L</i>	53.52 +/- 7.41	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.35
<i>TINF2</i>	51.29 +/- 8.06	100.0 +/- 0.0	100.0 +/- 0.01	97.99 +/- 6.43
<i>TMEM106B</i>	55.21 +/- 7.37	99.44 +/- 1.05	99.36 +/- 1.03	98.25 +/- 1.52
<i>TMEM107</i>	53.12 +/- 8.41	100.0 +/- 0.0	99.97 +/- 0.19	98.74 +/- 3.11
<i>TMEM138</i>	54.49 +/- 10.16	100.0 +/- 0.0	99.89 +/- 0.54	98.69 +/- 6.18
<i>TMEM165</i>	54.73 +/- 7.48	99.98 +/- 0.04	99.96 +/- 0.15	98.24 +/- 3.61
<i>TMEM199</i>	53.1 +/- 8.59	100.0 +/- 0.0	100.0 +/- 0.0	99.31 +/- 2.23
<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>TMEM240</i>	43.25 +/- 8.41	99.83 +/- 0.73	97.13 +/- 7.33	84.6 +/- 17.11
<i>TMEM63C</i>	50.59 +/- 8.56	100.0 +/- 0.0	99.86 +/- 0.94	97.81 +/- 6.99
<i>TMEM67</i>	54.94 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.03	99.08 +/- 1.68
<i>TOE1</i>	49.3 +/- 8.19	100.0 +/- 0.0	99.89 +/- 0.72	98.96 +/- 3.73
<i>TPP1</i>	50.74 +/- 8.83	100.0 +/- 0.0	100.0 +/- 0.0	98.62 +/- 3.99
<i>TPR</i>	55.3 +/- 7.6	100.0 +/- 0.0	100.0 +/- 0.03	99.4 +/- 1.28
<i>TRAPPC11</i>	55.17 +/- 7.68	100.0 +/- 0.0	99.98 +/- 0.1	99.21 +/- 1.5
<i>TSEN15</i>	55.58 +/- 7.66	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.18
<i>TSEN2</i>	54.0 +/- 7.66	100.0 +/- 0.0	100.0 +/- 0.0	99.49 +/- 1.16
<i>TSEN34</i>	52.18 +/- 8.58	100.0 +/- 0.0	99.9 +/- 0.58	98.65 +/- 4.88
<i>TSEN54</i>	49.17 +/- 8.28	100.0 +/- 0.0	99.78 +/- 1.19	97.5 +/- 6.82
<i>TTBK2</i>	54.59 +/- 7.39	99.99 +/- 0.06	99.94 +/- 0.25	99.17 +/- 1.87
<i>TTC19</i>	53.28 +/- 7.76	100.0 +/- 0.0	99.96 +/- 0.16	98.06 +/- 5.06
<i>TTPA</i>	54.78 +/- 7.23	99.98 +/- 0.04	99.97 +/- 0.05	99.0 +/- 3.25

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
TUBA1A	51.74 +/- 8.54	100.0 +/- 0.0	100.0 +/- 0.0	99.01 +/- 3.02
TUBA8	51.29 +/- 8.34	100.0 +/- 0.0	100.0 +/- 0.0	98.74 +/- 4.6
TUBB	53.39 +/- 8.13	100.0 +/- 0.0	100.0 +/- 0.0	98.79 +/- 4.72
TUBB2A	40.29 +/- 6.55	99.02 +/- 3.04	88.44 +/- 10.64	69.93 +/- 11.67
TUBB2B	43.53 +/- 6.76	99.38 +/- 2.41	92.79 +/- 8.02	76.52 +/- 9.9
TUBB3	56.08 +/- 8.71	100.0 +/- 0.0	99.91 +/- 0.52	98.6 +/- 3.98
TUBB4A	50.1 +/- 8.11	100.0 +/- 0.0	99.51 +/- 2.08	94.53 +/- 10.01
TUSC3	54.81 +/- 6.97	100.0 +/- 0.0	100.0 +/- 0.0	99.75 +/- 0.86
TWNK	52.49 +/- 9.07	100.0 +/- 0.0	99.91 +/- 0.44	98.51 +/- 5.9
TXNDC15	53.6 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.0	99.2 +/- 2.54
UBA5	51.82 +/- 6.93	100.0 +/- 0.0	99.91 +/- 0.4	98.21 +/- 2.77
UBAP1	50.17 +/- 8.37	99.95 +/- 0.05	99.73 +/- 0.95	97.13 +/- 6.19
UBR4	53.99 +/- 8.04	100.0 +/- 0.0	99.97 +/- 0.12	99.08 +/- 2.68
UBTF	49.73 +/- 8.42	99.99 +/- 0.03	99.68 +/- 0.78	96.44 +/- 6.6
UCHL1	53.36 +/- 7.63	100.0 +/- 0.0	99.9 +/- 0.64	98.9 +/- 3.41
USP8	55.01 +/- 7.88	100.0 +/- 0.0	99.99 +/- 0.04	99.39 +/- 1.49
VAMP1	51.89 +/- 8.12	100.0 +/- 0.0	100.0 +/- 0.0	98.54 +/- 4.68
VLDLR	54.03 +/- 7.8	99.97 +/- 0.05	99.93 +/- 0.16	98.88 +/- 1.99
VMA22	53.73 +/- 8.2	100.0 +/- 0.0	99.94 +/- 0.39	98.73 +/- 4.42
VPS13B	55.06 +/- 7.31	100.0 +/- 0.0	100.0 +/- 0.01	99.62 +/- 1.05
VPS13D	53.76 +/- 7.65	100.0 +/- 0.0	99.99 +/- 0.04	99.18 +/- 1.94
VPS37A	54.49 +/- 7.57	100.0 +/- 0.0	99.85 +/- 0.42	98.74 +/- 1.82
VPS41	54.93 +/- 7.14	100.0 +/- 0.0	100.0 +/- 0.0	99.68 +/- 0.74
VPS53	50.14 +/- 7.75	98.06 +/- 0.44	96.48 +/- 1.16	92.89 +/- 4.32
VRK1	56.03 +/- 7.99	100.0 +/- 0.0	99.99 +/- 0.04	99.47 +/- 1.23
WASHC5	53.52 +/- 7.48	100.0 +/- 0.0	99.99 +/- 0.04	99.38 +/- 2.07
WDR45B	52.49 +/- 8.01	100.0 +/- 0.0	99.88 +/- 0.68	97.99 +/- 5.17
WDR48	55.69 +/- 7.83	100.0 +/- 0.0	99.99 +/- 0.03	99.65 +/- 0.86
WDR73	51.07 +/- 8.43	99.98 +/- 0.07	99.92 +/- 0.13	98.83 +/- 3.16
WDR81	53.85 +/- 8.82	100.0 +/- 0.0	99.93 +/- 0.49	98.84 +/- 4.16
WFS1	54.56 +/- 9.12	100.0 +/- 0.0	99.97 +/- 0.11	98.44 +/- 6.43
WWOX	55.25 +/- 7.83	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.74
XRCC1	46.93 +/- 7.82	100.0 +/- 0.03	99.68 +/- 1.3	95.09 +/- 10.31
XYLT1	51.32 +/- 8.06	99.98 +/- 0.09	99.49 +/- 1.2	96.92 +/- 5.6
XYLT2	52.98 +/- 8.02	100.0 +/- 0.0	99.81 +/- 0.73	98.4 +/- 5.72
ZEB2	53.74 +/- 7.37	100.0 +/- 0.0	99.96 +/- 0.14	98.82 +/- 2.32
ZFYVE26	52.88 +/- 7.91	100.0 +/- 0.0	99.94 +/- 0.29	98.83 +/- 3.35
ZFYVE27	51.28 +/- 8.49	100.0 +/- 0.0	99.87 +/- 0.78	98.2 +/- 5.31
ZNF423	52.79 +/- 8.22	99.99 +/- 0.08	99.68 +/- 0.81	97.53 +/- 3.64
ZNF592	52.28 +/- 7.9	100.0 +/- 0.0	99.95 +/- 0.2	98.47 +/- 4.21
ZSWIM6	51.36 +/- 7.39	99.43 +/- 0.48	97.42 +/- 1.64	92.62 +/- 4.02

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