

Analyse af gener relevante ved mistanke om nyresvigt og nefrotisk syndrom

Panelbeskrivelse

Navn: Analyse af gener relevante ved mistanke om nyresvigt og nefrotisk syndrom

Antal gener: 424

version: 2.0

Ibrugtagningsdato: 2024-01-17

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. Der undersøges endvidere for deletionerne 11p13del, 16p11.2del, 17q12del, 22q11del, og 2q13del. 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 [%]
ACE	50.09 +/- 8.03	99.97 +/- 0.18	99.59 +/- 1.11	96.4 +/- 7.61
ACTB	52.67 +/- 9.59	100.0 +/- 0.0	99.97 +/- 0.13	98.33 +/- 4.59
ACTG1	50.51 +/- 9.23	100.0 +/- 0.0	99.86 +/- 0.67	97.56 +/- 4.62
ACTG2	52.28 +/- 8.33	100.0 +/- 0.0	100.0 +/- 0.01	98.75 +/- 4.05
ACTN4	49.98 +/- 8.57	99.99 +/- 0.04	99.77 +/- 0.63	96.49 +/- 6.23
ADAMTS13	50.69 +/- 8.84	100.0 +/- 0.0	99.78 +/- 0.98	96.8 +/- 6.62
ADAMTS9	53.08 +/- 7.58	100.0 +/- 0.0	99.91 +/- 0.36	98.31 +/- 2.94
AGT	53.59 +/- 8.91	100.0 +/- 0.0	100.0 +/- 0.0	98.8 +/- 4.45
AGTR1	55.28 +/- 7.14	100.0 +/- 0.0	99.99 +/- 0.07	99.41 +/- 2.17
AGXT	52.74 +/- 8.2	100.0 +/- 0.0	99.94 +/- 0.21	98.69 +/- 4.4
AHI1	55.19 +/- 7.32	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 0.81
ALDOB	54.32 +/- 7.32	100.0 +/- 0.0	100.0 +/- 0.0	99.57 +/- 0.98
ALG1	52.41 +/- 8.45	99.96 +/- 0.05	99.95 +/- 0.06	99.05 +/- 4.0
ALG5	54.21 +/- 6.74	100.0 +/- 0.0	99.96 +/- 0.28	99.35 +/- 2.12
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
ALMS1	53.24 +/- 7.52	100.0 +/- 0.0	99.98 +/- 0.06	99.18 +/- 1.66
ALPL	50.34 +/- 8.5	100.0 +/- 0.0	99.81 +/- 0.62	97.83 +/- 4.5
AMER1	40.4 +/- 15.38	99.68 +/- 1.03	91.0 +/- 15.47	65.97 +/- 37.88
AMN	50.24 +/- 9.42	100.0 +/- 0.0	99.87 +/- 0.55	96.01 +/- 10.85

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
ANKFY1	55.51 +/- 7.8	100.0 +/- 0.0	100.0 +/- 0.03	99.53 +/- 2.0
ANKS6	51.44 +/- 8.39	100.0 +/- 0.0	99.91 +/- 0.48	98.12 +/- 5.44
ANLN	55.79 +/- 7.69	100.0 +/- 0.0	99.99 +/- 0.06	99.57 +/- 1.24
ANOS1	42.27 +/- 15.56	99.91 +/- 0.27	94.09 +/- 9.75	69.44 +/- 35.52
AP2S1	46.71 +/- 8.82	100.0 +/- 0.0	99.72 +/- 0.73	94.83 +/- 9.39
APOA1	54.63 +/- 8.92	100.0 +/- 0.0	99.98 +/- 0.12	99.12 +/- 4.13
APOA2	51.57 +/- 9.17	100.0 +/- 0.0	99.82 +/- 1.08	97.98 +/- 8.0
APOC2	46.55 +/- 7.94	100.0 +/- 0.0	99.91 +/- 0.61	96.54 +/- 7.5
APOC3	47.71 +/- 8.23	100.0 +/- 0.0	100.0 +/- 0.0	97.79 +/- 5.88
APOE	55.72 +/- 9.88	100.0 +/- 0.0	99.98 +/- 0.13	98.39 +/- 4.98
APOL1	49.26 +/- 8.43	100.0 +/- 0.01	99.99 +/- 0.03	98.44 +/- 3.53
APRT	53.87 +/- 9.05	100.0 +/- 0.0	99.83 +/- 1.13	98.12 +/- 5.46
AQP11	53.51 +/- 8.18	100.0 +/- 0.0	100.0 +/- 0.0	99.18 +/- 2.29
AQP2	52.03 +/- 8.63	99.98 +/- 0.04	99.96 +/- 0.08	98.61 +/- 4.25
ARHGAP24	51.15 +/- 7.12	100.0 +/- 0.0	99.75 +/- 0.6	97.34 +/- 3.56
ARHGDIA	46.78 +/- 8.32	100.0 +/- 0.0	99.5 +/- 1.47	94.68 +/- 11.17
ARID1B	50.67 +/- 7.13	99.97 +/- 0.12	98.93 +/- 2.58	93.58 +/- 5.86
ARL13B	55.42 +/- 6.97	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 0.82
ARL6	56.43 +/- 7.8	100.0 +/- 0.0	100.0 +/- 0.01	99.46 +/- 1.64
ARMC9	52.98 +/- 8.18	100.0 +/- 0.01	99.98 +/- 0.09	98.97 +/- 3.48
ATP2A2	52.6 +/- 8.16	100.0 +/- 0.0	99.79 +/- 0.79	98.3 +/- 3.4
ATP6V0A4	52.12 +/- 7.62	100.0 +/- 0.0	99.99 +/- 0.07	99.12 +/- 2.0
ATP6V1B1	49.6 +/- 8.58	100.0 +/- 0.0	99.96 +/- 0.26	97.15 +/- 6.98
ATP6V1C2	53.69 +/- 8.89	100.0 +/- 0.0	99.96 +/- 0.27	98.93 +/- 2.54
ATP7B	53.21 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.03	99.16 +/- 2.06
AVIL	53.34 +/- 8.31	100.0 +/- 0.0	100.0 +/- 0.0	98.96 +/- 4.28
AVP	47.78 +/- 9.54	100.0 +/- 0.0	99.49 +/- 2.41	93.35 +/- 13.23
AVPR2	41.86 +/- 16.72	99.92 +/- 0.49	91.14 +/- 15.73	66.73 +/- 38.65
B2M	53.68 +/- 8.02	100.0 +/- 0.0	100.0 +/- 0.0	99.68 +/- 0.86
B3GLCT	54.52 +/- 7.58	99.86 +/- 0.09	99.86 +/- 0.09	99.11 +/- 1.48
B9D1	51.09 +/- 8.08	100.0 +/- 0.0	100.0 +/- 0.0	98.7 +/- 5.02
B9D2	51.21 +/- 8.93	100.0 +/- 0.0	100.0 +/- 0.0	98.53 +/- 5.79
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
BCOR	41.48 +/- 15.61	99.57 +/- 1.64	91.31 +/- 13.69	67.34 +/- 35.47
BCS1L	52.14 +/- 8.4	100.0 +/- 0.0	100.0 +/- 0.0	98.85 +/- 5.02
BICC1	54.52 +/- 7.28	100.0 +/- 0.0	99.99 +/- 0.05	99.06 +/- 1.71
BMP4	54.45 +/- 8.03	100.0 +/- 0.0	100.0 +/- 0.0	98.78 +/- 3.56
BMP7	52.34 +/- 8.48	100.0 +/- 0.0	99.9 +/- 0.67	97.97 +/- 6.81

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>BNC2</i>	54.48 +/- 7.8	99.97 +/- 0.05	99.93 +/- 0.12	99.07 +/- 2.13
<i>BSND</i>	51.96 +/- 8.18	100.0 +/- 0.0	99.94 +/- 0.37	98.67 +/- 4.94
<i>C3</i>	49.54 +/- 8.42	100.0 +/- 0.0	99.82 +/- 0.82	96.81 +/- 6.95
<i>CA2</i>	55.27 +/- 8.18	100.0 +/- 0.0	99.94 +/- 0.4	99.07 +/- 2.18
<i>CACNA1H</i>	52.09 +/- 9.25	99.93 +/- 0.33	98.6 +/- 2.02	94.0 +/- 6.67
<i>CASR</i>	51.5 +/- 7.85	99.99 +/- 0.09	99.66 +/- 0.5	97.56 +/- 3.22
<i>CC2D2A</i>	54.85 +/- 8.01	100.0 +/- 0.0	99.99 +/- 0.09	99.36 +/- 2.46
<i>CCNQ</i>	44.13 +/- 16.36	99.76 +/- 1.03	94.12 +/- 12.84	72.23 +/- 32.78
<i>CD151</i>	51.9 +/- 9.4	99.88 +/- 0.1	99.79 +/- 0.42	97.49 +/- 7.29
<i>CD2AP</i>	53.59 +/- 7.52	100.0 +/- 0.02	99.93 +/- 0.3	98.77 +/- 2.25
<i>CD46</i>	55.75 +/- 7.63	100.0 +/- 0.0	100.0 +/- 0.0	99.37 +/- 1.45
<i>CDC5L</i>	55.67 +/- 7.5	100.0 +/- 0.01	100.0 +/- 0.01	99.58 +/- 0.63
<i>CDKN1C</i>	51.19 +/- 8.43	99.98 +/- 0.1	99.53 +/- 1.54	94.83 +/- 10.57
<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>CEP120</i>	54.91 +/- 7.32	100.0 +/- 0.0	99.97 +/- 0.15	99.12 +/- 2.02
<i>CEP164</i>	49.98 +/- 8.18	100.0 +/- 0.0	99.82 +/- 0.5	96.83 +/- 6.45
<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
<i>CEP55</i>	54.66 +/- 7.57	100.0 +/- 0.0	100.0 +/- 0.03	99.28 +/- 1.9
<i>CEP83</i>	55.79 +/- 7.36	100.0 +/- 0.0	99.96 +/- 0.27	99.45 +/- 1.91
<i>CFB</i>	50.66 +/- 8.33	100.0 +/- 0.0	100.0 +/- 0.0	98.96 +/- 3.26
<i>CFH</i>	58.11 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.0	99.82 +/- 0.52
<i>CFHR1</i>	46.5 +/- 11.32	99.8 +/- 0.94	95.15 +/- 6.47	79.95 +/- 21.83
<i>CFHR2</i>	52.78 +/- 7.31	99.97 +/- 0.15	96.59 +/- 4.63	91.48 +/- 5.83
<i>CFHR3</i>	50.5 +/- 13.03	99.95 +/- 0.33	99.0 +/- 3.35	89.27 +/- 21.77
<i>CFHR4</i>	57.21 +/- 7.7	100.0 +/- 0.0	100.0 +/- 0.0	99.87 +/- 0.33
<i>CFHR5</i>	56.14 +/- 8.21	100.0 +/- 0.0	100.0 +/- 0.0	99.45 +/- 1.65
<i>CFI</i>	54.73 +/- 7.99	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.77
<i>CHD1L</i>	53.49 +/- 7.68	100.0 +/- 0.0	99.97 +/- 0.18	99.29 +/- 1.98
<i>CHD7</i>	53.94 +/- 7.9	100.0 +/- 0.0	99.89 +/- 0.31	98.68 +/- 2.47
<i>CHRM3</i>	53.92 +/- 7.71	100.0 +/- 0.0	99.98 +/- 0.1	99.03 +/- 1.96
<i>CHRNA3</i>	52.07 +/- 7.84	99.96 +/- 0.05	99.89 +/- 0.41	98.48 +/- 3.27
<i>CILK1</i>	54.96 +/- 7.86	100.0 +/- 0.0	99.96 +/- 0.17	99.4 +/- 1.42
<i>CISD2</i>	56.57 +/- 8.37	100.0 +/- 0.0	99.99 +/- 0.09	99.31 +/- 1.73
<i>CLCN5</i>	43.08 +/- 16.41	99.98 +/- 0.09	94.66 +/- 8.04	69.49 +/- 35.71
<i>CLCNKA</i>	45.14 +/- 7.78	99.04 +/- 1.65	97.09 +/- 4.08	88.76 +/- 11.11
<i>CLCNKB</i>	48.77 +/- 9.62	100.0 +/- 0.0	99.84 +/- 0.39	94.16 +/- 9.81
<i>CLDN10</i>	55.12 +/- 7.47	100.0 +/- 0.0	100.0 +/- 0.0	99.43 +/- 1.55
<i>CLDN16</i>	54.68 +/- 7.31	100.0 +/- 0.0	99.96 +/- 0.23	99.34 +/- 1.65
<i>CLDN19</i>	49.61 +/- 9.11	100.0 +/- 0.0	99.84 +/- 1.0	96.86 +/- 9.17
<i>CLVS1</i>	54.47 +/- 7.85	99.99 +/- 0.03	99.99 +/- 0.03	99.46 +/- 1.45
<i>COL4A1</i>	53.49 +/- 7.61	100.0 +/- 0.0	99.99 +/- 0.04	98.83 +/- 3.9
<i>COL4A3</i>	54.18 +/- 7.33	100.0 +/- 0.0	99.99 +/- 0.08	99.1 +/- 1.77
<i>COL4A4</i>	53.64 +/- 7.23	100.0 +/- 0.0	99.98 +/- 0.09	99.07 +/- 1.64
<i>COL4A5</i>	41.78 +/- 15.52	99.93 +/- 0.33	93.31 +/- 10.33	67.99 +/- 36.59

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>COL4A6</i>	40.23 +/- 15.44	99.82 +/- 0.84	91.02 +/- 15.09	65.75 +/- 39.13
<i>COQ2</i>	55.89 +/- 7.92	100.0 +/- 0.0	100.0 +/- 0.0	99.01 +/- 3.42
<i>COQ4</i>	51.78 +/- 8.16	100.0 +/- 0.0	99.65 +/- 2.12	97.65 +/- 8.57
<i>COQ6</i>	53.85 +/- 8.14	100.0 +/- 0.0	99.97 +/- 0.22	99.15 +/- 3.1
<i>COQ8A</i>	55.02 +/- 9.11	100.0 +/- 0.0	100.0 +/- 0.0	99.28 +/- 2.55
<i>COQ8B</i>	48.97 +/- 8.25	100.0 +/- 0.0	99.83 +/- 0.73	97.05 +/- 7.99
<i>COQ9</i>	51.79 +/- 8.9	100.0 +/- 0.0	100.0 +/- 0.0	98.97 +/- 2.62
<i>COX10</i>	55.39 +/- 8.65	100.0 +/- 0.0	99.96 +/- 0.28	98.27 +/- 6.06
<i>CPLANE1</i>	55.19 +/- 7.49	100.0 +/- 0.0	99.98 +/- 0.08	99.5 +/- 0.87
<i>CPT2</i>	52.9 +/- 8.76	100.0 +/- 0.0	99.99 +/- 0.04	98.76 +/- 4.32
<i>CRB2</i>	51.5 +/- 8.23	99.95 +/- 0.05	99.88 +/- 0.24	97.72 +/- 5.88
<i>CRKL</i>	54.11 +/- 8.01	99.98 +/- 0.04	99.92 +/- 0.13	98.86 +/- 2.82
<i>CSPP1</i>	54.76 +/- 7.34	100.0 +/- 0.0	100.0 +/- 0.0	99.43 +/- 0.86
<i>CTNS</i>	54.92 +/- 9.25	100.0 +/- 0.0	99.92 +/- 0.33	98.16 +/- 3.76
<i>CTU2</i>	51.9 +/- 8.92	100.0 +/- 0.01	99.94 +/- 0.13	97.38 +/- 7.24
<i>CUBN</i>	55.22 +/- 7.52	100.0 +/- 0.0	99.99 +/- 0.04	99.5 +/- 1.27
<i>CUL3</i>	56.04 +/- 7.63	99.98 +/- 0.04	99.84 +/- 0.47	99.0 +/- 1.71
<i>CYP11B1</i>	50.86 +/- 8.15	100.0 +/- 0.0	100.0 +/- 0.03	98.78 +/- 2.82
<i>CYP11B2</i>	49.04 +/- 8.54	100.0 +/- 0.0	99.95 +/- 0.28	97.31 +/- 6.94
<i>CYP17A1</i>	50.96 +/- 8.04	100.0 +/- 0.0	99.91 +/- 0.61	98.06 +/- 6.79
<i>CYP24A1</i>	53.09 +/- 7.95	100.0 +/- 0.0	99.97 +/- 0.11	98.25 +/- 3.45
<i>DAAM2</i>	50.59 +/- 8.59	100.0 +/- 0.0	99.91 +/- 0.54	98.02 +/- 6.49
<i>DACT1</i>	54.66 +/- 7.93	100.0 +/- 0.0	100.0 +/- 0.0	98.85 +/- 3.27
<i>DCDC2</i>	55.83 +/- 7.63	100.0 +/- 0.0	100.0 +/- 0.01	99.62 +/- 1.06
<i>DDX59</i>	52.36 +/- 7.29	100.0 +/- 0.0	99.84 +/- 0.68	98.54 +/- 2.78
<i>DGKE</i>	56.37 +/- 8.04	100.0 +/- 0.0	99.99 +/- 0.06	99.58 +/- 0.86
<i>DHCR7</i>	53.79 +/- 9.03	99.97 +/- 0.12	99.79 +/- 0.86	98.57 +/- 5.94
<i>DLC1</i>	54.39 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.03	99.45 +/- 1.28
<i>DLG5</i>	52.29 +/- 8.09	100.0 +/- 0.0	99.89 +/- 0.35	97.97 +/- 4.71
<i>DMP1</i>	54.33 +/- 8.19	99.99 +/- 0.03	99.97 +/- 0.07	99.36 +/- 1.8
<i>DNAJB11</i>	53.16 +/- 7.34	100.0 +/- 0.0	99.98 +/- 0.1	98.1 +/- 3.27
<i>DSTYK</i>	52.8 +/- 7.84	100.0 +/- 0.0	100.0 +/- 0.03	98.96 +/- 3.66
<i>DYNC2H1</i>	55.74 +/- 7.17	100.0 +/- 0.0	100.0 +/- 0.0	99.64 +/- 0.76
<i>DYNC2I1</i>	54.59 +/- 7.83	99.99 +/- 0.05	99.87 +/- 0.76	99.25 +/- 1.94
<i>DYNC2I2</i>	53.09 +/- 8.9	100.0 +/- 0.0	99.78 +/- 1.4	98.46 +/- 5.13
<i>DZIP1L</i>	53.1 +/- 8.22	100.0 +/- 0.0	99.97 +/- 0.18	98.65 +/- 4.92
<i>EGF</i>	54.69 +/- 7.27	100.0 +/- 0.0	100.0 +/- 0.0	99.61 +/- 0.92
<i>EHHADH</i>	54.34 +/- 7.83	100.0 +/- 0.0	100.0 +/- 0.0	99.46 +/- 1.91
<i>EIF2AK3</i>	55.03 +/- 7.92	99.95 +/- 0.05	99.86 +/- 0.42	99.19 +/- 2.11
<i>EMP2</i>	53.73 +/- 8.08	99.98 +/- 0.04	99.89 +/- 0.1	98.96 +/- 2.35
<i>ENPP1</i>	54.21 +/- 7.85	100.0 +/- 0.0	99.95 +/- 0.13	98.77 +/- 2.71
<i>ERCC8</i>	54.54 +/- 7.59	100.0 +/- 0.0	100.0 +/- 0.03	99.42 +/- 1.25
<i>EYA1</i>	55.91 +/- 7.53	100.0 +/- 0.0	100.0 +/- 0.0	99.62 +/- 0.99
<i>FAH</i>	51.3 +/- 8.58	100.0 +/- 0.0	99.76 +/- 0.84	97.67 +/- 6.81
<i>FAM20A</i>	51.18 +/- 7.55	100.0 +/- 0.0	99.81 +/- 0.98	97.28 +/- 6.24
<i>FAN1</i>	54.21 +/- 7.44	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.4
<i>FANCA</i>	52.83 +/- 7.93	100.0 +/- 0.0	99.94 +/- 0.4	98.6 +/- 4.06

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>FANCB</i>	42.87 +/- 15.58	99.92 +/- 0.37	95.2 +/- 7.95	70.58 +/- 35.02
<i>FANCD2</i>	56.01 +/- 8.31	100.0 +/- 0.0	100.0 +/- 0.01	99.29 +/- 1.86
<i>FANCE</i>	49.67 +/- 8.49	100.0 +/- 0.0	99.84 +/- 0.67	97.26 +/- 5.96
<i>FANCF</i>	54.19 +/- 7.69	100.0 +/- 0.0	99.99 +/- 0.06	99.16 +/- 2.74
<i>FANCI</i>	54.8 +/- 7.98	100.0 +/- 0.0	100.0 +/- 0.0	99.57 +/- 1.03
<i>FANCL</i>	55.62 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.0	99.71 +/- 0.89
<i>FAT1</i>	55.13 +/- 7.69	100.0 +/- 0.03	99.94 +/- 0.22	99.41 +/- 1.6
<i>FAT4</i>	54.0 +/- 7.7	100.0 +/- 0.0	99.97 +/- 0.16	99.19 +/- 2.41
<i>FBXL4</i>	55.91 +/- 7.3	100.0 +/- 0.0	100.0 +/- 0.03	99.58 +/- 0.98
<i>FCGR2A</i>	58.58 +/- 9.91	100.0 +/- 0.0	100.0 +/- 0.0	99.77 +/- 0.89
<i>FGA</i>	52.11 +/- 8.44	100.0 +/- 0.0	99.99 +/- 0.06	98.86 +/- 3.04
<i>FGF10</i>	53.81 +/- 7.31	100.0 +/- 0.01	99.94 +/- 0.2	98.98 +/- 2.43
<i>FGF20</i>	55.24 +/- 7.1	100.0 +/- 0.0	100.0 +/- 0.0	99.27 +/- 2.08
<i>FGF23</i>	51.67 +/- 7.76	100.0 +/- 0.0	99.98 +/- 0.06	98.56 +/- 3.88
<i>FGFR2</i>	53.88 +/- 7.73	100.0 +/- 0.0	99.83 +/- 1.13	98.64 +/- 4.2
<i>FGFR3</i>	53.79 +/- 8.88	100.0 +/- 0.0	99.78 +/- 0.84	97.9 +/- 4.27
<i>FN1</i>	54.82 +/- 7.91	100.0 +/- 0.0	99.93 +/- 0.43	99.06 +/- 2.17
<i>FOXC2</i>	49.24 +/- 8.27	100.0 +/- 0.01	99.68 +/- 1.66	95.46 +/- 10.36
<i>FOXP1</i>	54.69 +/- 7.36	100.0 +/- 0.03	99.93 +/- 0.29	98.75 +/- 1.97
<i>FOXP3</i>	38.44 +/- 14.75	99.72 +/- 0.89	88.99 +/- 17.07	63.47 +/- 39.91
<i>FRAS1</i>	53.66 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.03	99.32 +/- 1.77
<i>FREM1</i>	54.5 +/- 7.71	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 2.7
<i>FREM2</i>	55.11 +/- 7.91	100.0 +/- 0.0	99.99 +/- 0.06	99.35 +/- 2.4
<i>FXRD</i>	49.6 +/- 8.85	100.0 +/- 0.0	100.0 +/- 0.0	97.99 +/- 6.03
<i>G6PC1</i>	52.27 +/- 8.53	99.89 +/- 0.1	99.86 +/- 0.15	98.9 +/- 2.44
<i>GALNT3</i>	55.35 +/- 7.5	100.0 +/- 0.0	100.0 +/- 0.0	99.36 +/- 1.49
<i>GANAB</i>	50.55 +/- 7.86	100.0 +/- 0.0	100.0 +/- 0.0	98.46 +/- 3.74
<i>GAPVD1</i>	55.01 +/- 7.55	100.0 +/- 0.0	100.0 +/- 0.0	99.39 +/- 1.76
<i>GATA3</i>	52.14 +/- 8.08	100.0 +/- 0.0	99.91 +/- 0.47	97.78 +/- 4.33
<i>GATM</i>	54.64 +/- 8.23	100.0 +/- 0.0	99.87 +/- 0.79	98.83 +/- 2.67
<i>GDF6</i>	51.23 +/- 7.45	100.0 +/- 0.0	99.91 +/- 0.29	97.75 +/- 5.85
<i>GDNF</i>	51.34 +/- 7.36	100.0 +/- 0.0	99.96 +/- 0.21	98.35 +/- 3.32
<i>GFRA1</i>	54.1 +/- 7.94	100.0 +/- 0.0	99.98 +/- 0.07	99.12 +/- 2.42
<i>GLA</i>	43.47 +/- 15.87	99.99 +/- 0.04	95.69 +/- 8.38	73.05 +/- 32.79
<i>GLI3</i>	54.82 +/- 7.78	100.0 +/- 0.0	99.99 +/- 0.06	99.24 +/- 2.65
<i>GLIS2</i>	49.98 +/- 8.23	99.98 +/- 0.08	98.92 +/- 1.66	94.77 +/- 7.06
<i>GLIS3</i>	52.94 +/- 7.63	100.0 +/- 0.0	99.96 +/- 0.17	98.64 +/- 2.97
<i>GNA11</i>	51.7 +/- 8.7	99.97 +/- 0.16	99.48 +/- 1.48	96.01 +/- 7.41
<i>GON7</i>	57.47 +/- 7.44	100.0 +/- 0.0	100.0 +/- 0.0	99.48 +/- 2.27
<i>GPC3</i>	40.46 +/- 15.32	99.98 +/- 0.09	90.13 +/- 14.69	65.62 +/- 39.14
<i>GREB1L</i>	54.2 +/- 8.03	100.0 +/- 0.0	99.97 +/- 0.17	99.13 +/- 2.23
<i>GRHPR</i>	52.04 +/- 8.5	100.0 +/- 0.0	99.96 +/- 0.18	98.72 +/- 3.38
<i>GRIP1</i>	55.19 +/- 7.74	100.0 +/- 0.0	100.0 +/- 0.0	99.68 +/- 1.07
<i>GSN</i>	50.81 +/- 8.49	100.0 +/- 0.0	99.98 +/- 0.13	97.93 +/- 5.86
<i>HAAO</i>	51.38 +/- 8.59	100.0 +/- 0.0	100.0 +/- 0.0	98.04 +/- 7.04
<i>HNF1B</i>	49.91 +/- 8.03	100.0 +/- 0.0	99.66 +/- 1.6	96.96 +/- 6.57
<i>HNF4A</i>	49.73 +/- 8.48	99.99 +/- 0.06	99.81 +/- 0.5	96.56 +/- 7.06

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>HOGA1</i>	51.62 +/- 7.88	100.0 +/- 0.0	99.98 +/- 0.13	98.69 +/- 4.95
<i>HOXA13</i>	52.43 +/- 7.49	99.92 +/- 0.34	98.79 +/- 2.34	94.44 +/- 4.39
<i>HPRT1</i>	42.96 +/- 16.27	99.84 +/- 1.03	93.35 +/- 10.85	70.23 +/- 35.75
<i>HPSE2</i>	51.85 +/- 8.1	100.0 +/- 0.0	99.94 +/- 0.37	98.13 +/- 6.89
<i>HRAS</i>	49.95 +/- 9.48	99.95 +/- 0.17	99.17 +/- 2.84	94.67 +/- 10.68
<i>HSD11B2</i>	49.95 +/- 8.52	99.94 +/- 0.39	99.63 +/- 1.7	96.36 +/- 7.36
<i>HYLS1</i>	55.02 +/- 8.28	100.0 +/- 0.0	100.0 +/- 0.0	99.51 +/- 1.84
<i>IFT122</i>	52.88 +/- 7.76	100.0 +/- 0.0	99.99 +/- 0.06	99.08 +/- 3.38
<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>IFT43</i>	54.44 +/- 8.7	100.0 +/- 0.0	99.94 +/- 0.37	99.25 +/- 3.66
<i>IFT80</i>	56.56 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.0	99.68 +/- 0.94
<i>INF2</i>	47.8 +/- 8.29	99.14 +/- 0.92	95.63 +/- 3.04	87.46 +/- 8.73
<i>INPP5E</i>	50.81 +/- 8.98	100.0 +/- 0.0	99.5 +/- 2.37	95.13 +/- 8.84
<i>INVS</i>	54.28 +/- 8.09	100.0 +/- 0.0	100.0 +/- 0.03	99.41 +/- 1.59
<i>IQCB1</i>	54.8 +/- 7.29	99.92 +/- 0.1	99.89 +/- 0.1	99.46 +/- 0.92
<i>ITGA3</i>	49.19 +/- 8.16	100.0 +/- 0.0	99.96 +/- 0.24	97.22 +/- 6.6
<i>ITGA8</i>	54.23 +/- 7.88	100.0 +/- 0.0	99.98 +/- 0.05	99.32 +/- 1.71
<i>ITSN1</i>	52.88 +/- 7.76	100.0 +/- 0.02	99.97 +/- 0.12	98.87 +/- 3.08
<i>ITSN2</i>	53.56 +/- 7.24	100.0 +/- 0.0	99.88 +/- 0.23	98.58 +/- 2.63
<i>JAG1</i>	54.24 +/- 7.78	100.0 +/- 0.0	99.94 +/- 0.37	98.84 +/- 2.65
<i>JAM3</i>	55.4 +/- 8.11	100.0 +/- 0.0	100.0 +/- 0.0	99.4 +/- 1.64
<i>KANK1</i>	52.9 +/- 8.1	100.0 +/- 0.0	99.97 +/- 0.18	98.95 +/- 2.74
<i>KANK2</i>	51.35 +/- 7.76	99.98 +/- 0.06	99.67 +/- 0.62	97.49 +/- 5.48
<i>KANK4</i>	51.54 +/- 8.01	100.0 +/- 0.01	99.86 +/- 0.61	98.24 +/- 5.26
<i>KAT6B</i>	53.19 +/- 7.52	100.0 +/- 0.0	99.94 +/- 0.31	98.89 +/- 2.04
<i>KCNA1</i>	52.15 +/- 7.78	100.0 +/- 0.0	99.87 +/- 0.71	98.16 +/- 4.67
<i>KCNJ1</i>	53.88 +/- 8.51	100.0 +/- 0.0	100.0 +/- 0.0	99.38 +/- 1.98
<i>KCNJ5</i>	52.59 +/- 8.61	99.99 +/- 0.04	99.81 +/- 0.34	98.42 +/- 4.17
<i>KCNQ10T1</i>	53.17 +/- 7.72	100.0 +/- 0.01	99.92 +/- 0.16	98.75 +/- 2.64
<i>KCTD1</i>	52.42 +/- 7.93	99.99 +/- 0.03	99.64 +/- 0.84	96.06 +/- 5.0
<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>KIF14</i>	55.53 +/- 7.13	99.94 +/- 0.1	99.85 +/- 0.12	99.5 +/- 0.94
<i>KIF7</i>	52.39 +/- 8.25	100.0 +/- 0.0	99.88 +/- 0.79	97.97 +/- 4.36
<i>KIRREL1</i>	52.11 +/- 8.52	100.0 +/- 0.03	99.96 +/- 0.25	98.78 +/- 3.72
<i>KLHL3</i>	53.72 +/- 7.65	100.0 +/- 0.0	99.99 +/- 0.07	99.5 +/- 1.54
<i>KYNU</i>	54.66 +/- 7.75	99.92 +/- 0.05	99.88 +/- 0.07	99.1 +/- 1.57
<i>LAGE3</i>	43.85 +/- 17.08	99.94 +/- 0.42	93.54 +/- 15.99	69.86 +/- 37.12
<i>LAMA5</i>	53.27 +/- 9.05	100.0 +/- 0.0	99.9 +/- 0.28	97.47 +/- 4.98
<i>LAMB2</i>	53.76 +/- 8.87	100.0 +/- 0.0	99.98 +/- 0.16	98.94 +/- 4.73
<i>LCAT</i>	50.03 +/- 9.55	100.0 +/- 0.0	99.95 +/- 0.34	96.89 +/- 8.91
<i>LIFR</i>	54.58 +/- 7.26	100.0 +/- 0.0	99.88 +/- 0.32	98.69 +/- 1.79
<i>LMNA</i>	49.82 +/- 8.35	100.0 +/- 0.0	99.96 +/- 0.16	97.29 +/- 6.96
<i>LMX1B</i>	48.87 +/- 8.42	99.88 +/- 0.42	97.96 +/- 2.69	93.1 +/- 7.22

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>LRIG2</i>	53.73 +/- 7.61	100.0 +/- 0.0	99.97 +/- 0.06	99.27 +/- 1.95
<i>LRP2</i>	54.31 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.01	99.41 +/- 1.36
<i>LRP4</i>	51.39 +/- 8.21	100.0 +/- 0.0	99.95 +/- 0.26	98.26 +/- 4.32
<i>LYZ</i>	54.7 +/- 7.83	100.0 +/- 0.0	100.0 +/- 0.0	99.29 +/- 1.7
<i>LZTFL1</i>	56.16 +/- 8.07	100.0 +/- 0.0	99.99 +/- 0.03	99.54 +/- 1.34
<i>MAFB</i>	53.89 +/- 8.3	100.0 +/- 0.0	99.98 +/- 0.1	98.51 +/- 5.73
<i>MAGED2</i>	40.78 +/- 15.35	99.93 +/- 0.39	92.54 +/- 13.68	66.78 +/- 37.67
<i>MAGI2</i>	51.11 +/- 7.1	99.98 +/- 0.13	99.22 +/- 1.65	95.08 +/- 5.5
<i>MAPKBP1</i>	51.76 +/- 8.65	99.99 +/- 0.06	99.91 +/- 0.19	97.86 +/- 6.2
<i>MEFV</i>	51.61 +/- 8.18	99.98 +/- 0.04	99.97 +/- 0.04	98.92 +/- 3.72
<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>MMUT</i>	55.42 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.67 +/- 1.04
<i>MT-TF</i>	3330.41 +/- 827.67	100.0 +/- 0.0	100.0 +/- 0.0	100.0 +/- 0.0
<i>MT-TL1</i>	3618.58 +/- 725.81	100.0 +/- 0.0	100.0 +/- 0.0	100.0 +/- 0.0
<i>MTX2</i>	55.35 +/- 7.59	100.0 +/- 0.0	99.99 +/- 0.07	98.57 +/- 3.84
<i>MUC1^{1,2,3}</i>	29.22 +/- 5.83	65.9 +/- 4.48	56.04 +/- 4.31	48.97 +/- 6.43
<i>MYCN</i>	51.6 +/- 8.59	100.0 +/- 0.0	99.9 +/- 0.31	96.18 +/- 6.41
<i>MYH9</i>	52.53 +/- 8.07	100.0 +/- 0.0	99.98 +/- 0.14	98.64 +/- 4.25
<i>MYO1E</i>	53.41 +/- 8.16	99.97 +/- 0.07	99.95 +/- 0.09	99.07 +/- 2.36
<i>MYO9A</i>	54.95 +/- 7.4	100.0 +/- 0.0	99.98 +/- 0.11	99.38 +/- 1.88
<i>MYOCD</i>	52.98 +/- 7.72	100.0 +/- 0.0	99.99 +/- 0.06	99.09 +/- 3.05
<i>NADSYN1</i>	54.24 +/- 8.35	100.0 +/- 0.0	99.94 +/- 0.29	99.02 +/- 2.89
<i>NEK1</i>	54.4 +/- 7.35	100.0 +/- 0.0	99.97 +/- 0.14	99.32 +/- 1.62
<i>NEK8</i>	51.87 +/- 8.25	99.98 +/- 0.08	99.96 +/- 0.17	98.28 +/- 4.31
<i>NEU1</i>	51.09 +/- 8.34	100.0 +/- 0.0	99.98 +/- 0.11	98.09 +/- 5.5
<i>NHERF1</i>	50.25 +/- 8.27	100.0 +/- 0.0	99.97 +/- 0.15	97.4 +/- 7.28
<i>NIPBL</i>	54.63 +/- 6.99	100.0 +/- 0.03	99.82 +/- 0.35	98.43 +/- 1.48
<i>NLRP3</i>	53.25 +/- 8.44	100.0 +/- 0.0	99.93 +/- 0.4	98.69 +/- 5.25
<i>NOTCH2</i>	53.16 +/- 7.9	100.0 +/- 0.0	99.88 +/- 0.23	98.25 +/- 2.58
<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>NPHS1</i>	49.85 +/- 8.4	100.0 +/- 0.0	99.87 +/- 0.72	97.73 +/- 5.74
<i>NPHS2</i>	51.87 +/- 8.03	100.0 +/- 0.0	99.89 +/- 0.48	97.88 +/- 5.98
<i>NR3C1</i>	53.67 +/- 7.45	100.0 +/- 0.0	99.97 +/- 0.21	99.06 +/- 2.41
<i>NR3C2</i>	53.96 +/- 8.02	100.0 +/- 0.0	99.99 +/- 0.05	99.22 +/- 1.84
<i>NRIP1</i>	54.7 +/- 7.79	99.97 +/- 0.05	99.93 +/- 0.07	99.35 +/- 1.43
<i>NUP107</i>	55.62 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.44 +/- 1.37
<i>NUP133</i>	55.13 +/- 7.83	100.0 +/- 0.0	99.98 +/- 0.11	99.45 +/- 1.79
<i>NUP160</i>	54.89 +/- 7.82	100.0 +/- 0.0	100.0 +/- 0.0	99.61 +/- 0.89
<i>NUP205</i>	55.51 +/- 7.28	100.0 +/- 0.0	100.0 +/- 0.0	99.63 +/- 1.01
<i>NUP85</i>	53.33 +/- 8.49	100.0 +/- 0.0	100.0 +/- 0.03	98.98 +/- 2.64
<i>NUP93</i>	53.65 +/- 8.21	99.97 +/- 0.1	99.96 +/- 0.1	99.07 +/- 2.31
<i>NXF5</i>	37.49 +/- 14.12	98.21 +/- 3.27	86.03 +/- 17.51	60.27 +/- 36.3

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>OCRL</i>	40.95 +/- 15.68	99.87 +/- 0.59	91.54 +/- 13.83	66.68 +/- 38.47
<i>OFD1</i>	42.18 +/- 15.83	99.93 +/- 0.36	93.98 +/- 9.99	68.58 +/- 36.59
<i>OSGEP</i>	52.23 +/- 8.21	99.96 +/- 0.05	99.95 +/- 0.05	99.49 +/- 1.5
<i>PAX2</i>	45.55 +/- 7.82	100.0 +/- 0.01	99.64 +/- 1.68	93.13 +/- 12.12
<i>PBX1</i>	51.52 +/- 7.16	100.0 +/- 0.0	99.86 +/- 0.46	97.67 +/- 3.92
<i>PDE6D</i>	51.83 +/- 8.41	100.0 +/- 0.0	99.97 +/- 0.18	98.2 +/- 4.37
<i>PDSS2</i>	54.56 +/- 7.59	100.0 +/- 0.0	100.0 +/- 0.01	99.48 +/- 1.26
<i>PEX12</i>	52.36 +/- 8.05	99.94 +/- 0.09	99.91 +/- 0.19	99.26 +/- 1.91
<i>PEX2</i>	54.88 +/- 8.0	100.0 +/- 0.0	100.0 +/- 0.0	99.74 +/- 0.88
<i>PHEX</i>	42.23 +/- 15.53	99.9 +/- 0.34	94.16 +/- 10.44	68.85 +/- 35.68
<i>PIGN</i>	54.9 +/- 7.58	100.0 +/- 0.0	99.97 +/- 0.15	99.19 +/- 1.65
<i>PIGT</i>	50.65 +/- 8.58	100.0 +/- 0.0	99.96 +/- 0.27	97.76 +/- 5.86
<i>PKD1</i>	55.91 +/- 9.22	100.0 +/- 0.01	99.56 +/- 0.94	97.14 +/- 4.45
<i>PKD2</i>	53.55 +/- 7.31	100.0 +/- 0.0	99.72 +/- 0.98	97.86 +/- 4.21
<i>PKHD1</i>	54.57 +/- 7.68	100.0 +/- 0.0	99.99 +/- 0.06	99.52 +/- 1.3
<i>PLCE1</i>	54.78 +/- 7.56	100.0 +/- 0.01	99.99 +/- 0.06	99.36 +/- 2.17
<i>PMM2</i>	53.79 +/- 8.67	100.0 +/- 0.0	99.87 +/- 0.85	98.37 +/- 4.98
<i>PODXL</i>	50.83 +/- 8.41	100.0 +/- 0.02	99.87 +/- 0.42	97.63 +/- 5.82
<i>PRKCSH</i>	49.56 +/- 8.8	99.99 +/- 0.03	99.8 +/- 1.09	97.39 +/- 7.65
<i>PTPRO</i>	54.62 +/- 7.91	100.0 +/- 0.0	100.0 +/- 0.01	99.37 +/- 1.59
<i>REN</i>	49.79 +/- 8.57	100.0 +/- 0.0	99.97 +/- 0.15	96.95 +/- 7.11
<i>RET</i>	52.13 +/- 8.46	100.0 +/- 0.0	99.98 +/- 0.12	98.51 +/- 3.77
<i>RMND1</i>	54.92 +/- 7.89	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.8
<i>ROBO1</i>	55.1 +/- 6.94	100.0 +/- 0.0	100.0 +/- 0.01	99.61 +/- 0.83
<i>ROBO2</i>	54.73 +/- 7.43	100.0 +/- 0.0	99.98 +/- 0.07	99.32 +/- 1.17
<i>RPGRIP1L</i>	54.65 +/- 7.42	100.0 +/- 0.0	100.0 +/- 0.0	99.66 +/- 0.88
<i>RRAGD</i>	53.32 +/- 7.55	99.99 +/- 0.09	99.64 +/- 1.25	97.93 +/- 2.9
<i>RRM2B</i>	55.03 +/- 7.65	100.0 +/- 0.0	99.99 +/- 0.06	99.53 +/- 1.04
<i>SALL1</i>	52.18 +/- 8.0	100.0 +/- 0.0	99.9 +/- 0.29	98.14 +/- 4.76
<i>SALL4</i>	53.1 +/- 8.04	100.0 +/- 0.0	99.93 +/- 0.33	98.85 +/- 3.95
<i>SARS2</i>	49.15 +/- 8.09	100.0 +/- 0.0	99.95 +/- 0.25	97.5 +/- 6.54
<i>SCARB2</i>	54.77 +/- 8.08	100.0 +/- 0.0	99.97 +/- 0.18	99.32 +/- 2.51
<i>SCNN1A</i>	49.4 +/- 8.14	100.0 +/- 0.0	99.85 +/- 0.68	97.49 +/- 6.95
<i>SCNN1B</i>	50.53 +/- 8.34	100.0 +/- 0.0	100.0 +/- 0.03	98.34 +/- 4.77
<i>SCNN1G</i>	52.43 +/- 8.35	100.0 +/- 0.0	100.0 +/- 0.0	98.83 +/- 4.04
<i>SDCCAG8</i>	55.16 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.53 +/- 1.56
<i>SEC61A1</i>	54.11 +/- 8.24	100.0 +/- 0.0	99.93 +/- 0.34	98.56 +/- 5.08
<i>SEC63</i>	53.98 +/- 7.26	99.88 +/- 0.08	99.84 +/- 0.08	98.78 +/- 1.99
<i>SGPL1</i>	53.7 +/- 7.8	100.0 +/- 0.0	99.86 +/- 0.55	98.4 +/- 3.17
<i>SIX1</i>	52.44 +/- 7.62	100.0 +/- 0.0	99.95 +/- 0.31	98.61 +/- 4.0
<i>SIX2</i>	51.34 +/- 8.32	100.0 +/- 0.0	99.9 +/- 0.69	97.44 +/- 6.96
<i>SIX5</i>	48.22 +/- 9.01	99.97 +/- 0.19	99.18 +/- 2.09	93.15 +/- 11.54
<i>SLC12A1</i>	54.69 +/- 7.53	100.0 +/- 0.0	100.0 +/- 0.01	99.55 +/- 1.24
<i>SLC12A3</i>	51.38 +/- 7.83	100.0 +/- 0.0	99.95 +/- 0.13	97.57 +/- 5.45
<i>SLC22A12</i>	49.79 +/- 8.55	99.93 +/- 0.34	99.43 +/- 1.25	95.28 +/- 7.12
<i>SLC26A1</i>	59.15 +/- 10.19	100.0 +/- 0.0	99.9 +/- 0.67	99.0 +/- 4.49
<i>SLC2A2</i>	55.54 +/- 7.59	100.0 +/- 0.0	100.0 +/- 0.01	99.71 +/- 1.18

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>SLC2A9</i>	53.6 +/- 8.25	100.0 +/- 0.0	99.96 +/- 0.3	98.9 +/- 4.28
<i>SLC34A1</i>	50.05 +/- 9.04	100.0 +/- 0.0	99.92 +/- 0.29	97.59 +/- 6.37
<i>SLC34A3</i>	48.19 +/- 8.4	100.0 +/- 0.0	99.69 +/- 0.89	95.3 +/- 8.21
<i>SLC37A4</i>	52.3 +/- 8.85	100.0 +/- 0.0	99.87 +/- 0.74	98.29 +/- 5.73
<i>SLC3A1</i>	56.93 +/- 7.63	100.0 +/- 0.0	100.0 +/- 0.01	99.5 +/- 2.33
<i>SLC41A1</i>	51.25 +/- 7.9	100.0 +/- 0.0	99.91 +/- 0.39	98.18 +/- 4.38
<i>SLC4A1</i>	48.3 +/- 8.35	100.0 +/- 0.0	99.6 +/- 1.75	95.85 +/- 8.36
<i>SLC4A4</i>	54.85 +/- 7.79	99.94 +/- 0.13	99.91 +/- 0.16	99.29 +/- 0.96
<i>SLC5A2</i>	52.65 +/- 8.95	100.0 +/- 0.0	99.99 +/- 0.09	97.61 +/- 6.87
<i>SLC6A20</i>	52.73 +/- 7.81	100.0 +/- 0.01	99.97 +/- 0.14	98.82 +/- 2.66
<i>SLC7A7</i>	52.7 +/- 8.37	100.0 +/- 0.0	100.0 +/- 0.0	99.13 +/- 2.96
<i>SLC7A9</i>	52.39 +/- 8.05	100.0 +/- 0.0	100.0 +/- 0.0	98.81 +/- 4.44
<i>SLIT2</i>	53.97 +/- 7.54	100.0 +/- 0.0	99.99 +/- 0.03	99.25 +/- 1.58
<i>SMARCAL1</i>	52.78 +/- 7.67	100.0 +/- 0.0	99.97 +/- 0.21	98.85 +/- 3.15
<i>SRGAP1</i>	52.84 +/- 7.63	99.99 +/- 0.03	99.88 +/- 0.28	98.27 +/- 3.01
<i>STRA6</i>	49.84 +/- 8.46	100.0 +/- 0.0	99.91 +/- 0.57	97.75 +/- 8.12
<i>SYNPO</i>	48.57 +/- 8.11	99.98 +/- 0.04	99.77 +/- 0.64	95.88 +/- 8.84
<i>SYNPO2</i>	51.96 +/- 7.68	99.99 +/- 0.03	99.95 +/- 0.18	98.44 +/- 3.53
<i>TBC1D1</i>	53.54 +/- 7.5	100.0 +/- 0.0	99.89 +/- 0.43	98.79 +/- 2.7
<i>TBC1D24</i>	54.38 +/- 8.59	100.0 +/- 0.0	99.93 +/- 0.29	98.65 +/- 3.4
<i>TBC1D8B</i>	43.34 +/- 16.34	100.0 +/- 0.0	94.81 +/- 9.45	71.18 +/- 34.41
<i>TBX1</i>	44.42 +/- 8.42	100.0 +/- 0.0	97.39 +/- 7.75	87.45 +/- 18.6
<i>TBX18</i>	54.43 +/- 7.55	100.0 +/- 0.0	99.98 +/- 0.1	99.16 +/- 2.07
<i>TBX6</i>	48.44 +/- 8.76	100.0 +/- 0.0	99.4 +/- 1.24	93.62 +/- 10.93
<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>TFAP2A</i>	49.35 +/- 7.05	100.0 +/- 0.0	99.05 +/- 1.38	93.9 +/- 4.73
<i>THBD</i>	54.08 +/- 8.01	100.0 +/- 0.0	99.9 +/- 0.37	98.18 +/- 5.04
<i>TMEM107</i>	53.12 +/- 8.41	100.0 +/- 0.0	99.97 +/- 0.19	98.74 +/- 3.11
<i>TMEM126B</i>	56.97 +/- 8.13	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.59
<i>TMEM138</i>	54.49 +/- 10.16	100.0 +/- 0.0	99.89 +/- 0.54	98.69 +/- 6.18
<i>TMEM216</i>	51.73 +/- 9.05	100.0 +/- 0.0	100.0 +/- 0.0	98.86 +/- 2.76
<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>TMEM67</i>	54.94 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.03	99.08 +/- 1.68
<i>TNS2</i>	51.21 +/- 8.71	100.0 +/- 0.0	99.85 +/- 0.8	97.77 +/- 6.39
<i>TNXB</i>	47.58 +/- 7.81	97.64 +/- 1.41	92.13 +/- 2.3	87.42 +/- 5.39
<i>TP53RK</i>	56.03 +/- 7.95	100.0 +/- 0.0	99.99 +/- 0.09	99.52 +/- 1.77
<i>TP63</i>	55.97 +/- 7.98	100.0 +/- 0.0	100.0 +/- 0.0	99.66 +/- 1.02
<i>TPRKB</i>	54.1 +/- 7.82	100.0 +/- 0.0	100.0 +/- 0.0	99.44 +/- 1.7
<i>TRAF3IP1</i>	54.04 +/- 8.11	100.0 +/- 0.0	99.91 +/- 0.5	98.43 +/- 3.04
<i>TRAP1</i>	51.94 +/- 8.32	100.0 +/- 0.0	99.98 +/- 0.09	98.16 +/- 4.79
<i>TRIM32</i>	55.0 +/- 8.0	100.0 +/- 0.0	99.83 +/- 0.45	98.45 +/- 2.54
<i>TRIM8</i>	51.69 +/- 8.67	100.0 +/- 0.0	99.87 +/- 0.59	97.74 +/- 5.8
<i>TRPC6</i>	54.94 +/- 7.36	100.0 +/- 0.0	100.0 +/- 0.03	99.52 +/- 1.23
<i>TSC1</i>	54.3 +/- 7.8	100.0 +/- 0.0	99.99 +/- 0.03	99.26 +/- 2.15

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
TSC2	54.73 +/- 8.8	100.0 +/- 0.0	99.98 +/- 0.09	98.65 +/- 5.08
TTC21B	56.5 +/- 7.06	100.0 +/- 0.0	99.98 +/- 0.12	99.64 +/- 0.8
TTC8	55.08 +/- 7.81	100.0 +/- 0.0	99.87 +/- 0.7	99.14 +/- 2.09
TTR	55.5 +/- 8.45	100.0 +/- 0.0	100.0 +/- 0.0	99.52 +/- 1.77
TULP3	54.24 +/- 8.14	99.99 +/- 0.03	99.99 +/- 0.03	99.36 +/- 2.72
TXNDC15	53.6 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.0	99.2 +/- 2.54
UMOD	54.59 +/- 8.85	100.0 +/- 0.0	99.88 +/- 0.63	98.76 +/- 2.69
UPK3A	49.52 +/- 9.1	100.0 +/- 0.0	100.0 +/- 0.03	97.46 +/- 7.83
VHL	51.91 +/- 7.58	100.0 +/- 0.01	99.92 +/- 0.15	97.75 +/- 3.21
VIPAS39	53.3 +/- 8.48	100.0 +/- 0.0	99.95 +/- 0.31	98.66 +/- 3.88
VPS33B	52.77 +/- 8.38	100.0 +/- 0.0	99.94 +/- 0.35	99.06 +/- 2.64
WDPCP	55.08 +/- 7.68	100.0 +/- 0.0	99.96 +/- 0.16	99.43 +/- 1.45
WDR19	55.29 +/- 7.59	100.0 +/- 0.0	99.98 +/- 0.13	99.6 +/- 1.17
WDR35	56.0 +/- 7.39	100.0 +/- 0.0	99.99 +/- 0.06	99.79 +/- 0.88
WDR4	52.92 +/- 8.39	100.0 +/- 0.0	99.99 +/- 0.04	98.89 +/- 3.17
WDR72	55.31 +/- 7.55	100.0 +/- 0.0	100.0 +/- 0.0	99.61 +/- 0.88
WDR73	51.07 +/- 8.43	99.98 +/- 0.07	99.92 +/- 0.13	98.83 +/- 3.16
WNK1	51.73 +/- 7.56	99.99 +/- 0.04	99.78 +/- 0.51	97.83 +/- 3.27
WNK4	47.09 +/- 7.86	100.0 +/- 0.0	99.71 +/- 1.05	95.06 +/- 8.78
WNT3	51.6 +/- 8.02	99.99 +/- 0.04	99.82 +/- 0.43	97.61 +/- 5.03
WNT4	50.97 +/- 8.17	99.94 +/- 0.07	99.11 +/- 1.69	95.51 +/- 5.81
WNT5A	55.15 +/- 7.6	100.0 +/- 0.0	99.95 +/- 0.24	98.96 +/- 2.18
WNT9B	53.13 +/- 8.86	100.0 +/- 0.0	99.86 +/- 0.74	98.58 +/- 3.54
WT1	51.71 +/- 7.71	99.98 +/- 0.13	99.27 +/- 1.56	95.89 +/- 5.6
XDH	52.28 +/- 8.12	100.0 +/- 0.0	99.98 +/- 0.13	98.77 +/- 3.51
XPNPEP3	54.73 +/- 7.96	100.0 +/- 0.0	99.99 +/- 0.08	99.31 +/- 1.65
XPO5	54.68 +/- 7.95	100.0 +/- 0.0	100.0 +/- 0.0	99.27 +/- 2.68
YRDC	52.88 +/- 8.5	100.0 +/- 0.0	99.84 +/- 0.54	97.43 +/- 5.4
ZIC3	42.58 +/- 16.07	99.76 +/- 0.73	92.74 +/- 11.66	69.4 +/- 35.52
ZMPSTE24	55.06 +/- 6.99	100.0 +/- 0.0	100.0 +/- 0.0	99.61 +/- 1.18
ZMYM2	55.44 +/- 7.4	100.0 +/- 0.0	99.94 +/- 0.18	99.15 +/- 1.41
ZNF423	52.79 +/- 8.22	99.99 +/- 0.08	99.68 +/- 0.81	97.53 +/- 3.64

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