

Analyse af gener relevante ved mistanke om vækst og fedme syndromer

Panelbeskrivelse

Navn: Analyse af gener relevante ved mistanke om vækst og fedme syndromer

Antal gener: 223

version: 1.1

Ibrugtagningsdato: 2023-10-31

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 minimumsandelens 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 [%]
<i>ACAN</i> ²	44.64 +/- 7.05	89.15 +/- 1.15	87.01 +/- 0.85	84.21 +/- 4.62
<i>ACTB</i>	52.67 +/- 9.59	100.0 +/- 0.0	99.97 +/- 0.13	98.33 +/- 4.59
<i>ACTG1</i>	50.51 +/- 9.23	100.0 +/- 0.0	99.86 +/- 0.67	97.56 +/- 4.62
<i>AKR1C2</i>	49.29 +/- 6.81	99.96 +/- 0.25	99.26 +/- 1.43	94.51 +/- 4.4
<i>ALMS1</i>	53.24 +/- 7.52	100.0 +/- 0.0	99.98 +/- 0.06	99.18 +/- 1.66
<i>AMMECR1</i>	42.61 +/- 15.82	99.89 +/- 0.42	93.93 +/- 10.1	69.38 +/- 35.09
<i>ANKRD11</i>	51.64 +/- 7.84	99.96 +/- 0.25	99.12 +/- 1.83	95.6 +/- 5.63
<i>ARCN1</i>	54.17 +/- 7.92	99.93 +/- 0.04	99.9 +/- 0.08	98.84 +/- 2.22
<i>ARL6</i>	56.43 +/- 7.8	100.0 +/- 0.0	100.0 +/- 0.01	99.46 +/- 1.64
<i>ATR</i>	55.58 +/- 7.32	100.0 +/- 0.0	100.0 +/- 0.02	99.47 +/- 1.1
<i>ATRIP</i>	51.38 +/- 8.4	100.0 +/- 0.0	99.82 +/- 0.52	97.53 +/- 4.96
<i>ATRX</i>	41.93 +/- 15.41	99.92 +/- 0.24	94.06 +/- 9.46	68.88 +/- 35.99
<i>B3GAT3</i>	50.4 +/- 8.02	100.0 +/- 0.0	99.96 +/- 0.24	98.49 +/- 3.89
<i>BBS1</i>	52.34 +/- 8.31	100.0 +/- 0.0	100.0 +/- 0.03	98.98 +/- 3.48
<i>BBS10</i>	55.8 +/- 7.93	100.0 +/- 0.0	99.96 +/- 0.27	99.53 +/- 1.25
<i>BBS12</i>	56.62 +/- 8.74	100.0 +/- 0.0	99.99 +/- 0.07	99.45 +/- 1.89
<i>BBS2</i>	54.23 +/- 7.64	100.0 +/- 0.0	100.0 +/- 0.0	99.72 +/- 0.61
<i>BBS4</i>	53.66 +/- 8.02	100.0 +/- 0.0	99.99 +/- 0.09	99.52 +/- 1.37
<i>BBS5</i>	52.67 +/- 7.75	99.98 +/- 0.12	99.54 +/- 1.04	96.14 +/- 5.26
<i>BBS7</i>	56.19 +/- 7.17	100.0 +/- 0.0	99.99 +/- 0.06	99.64 +/- 0.95

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>BBS9</i>	54.64 +/- 7.47	100.0 +/- 0.0	99.99 +/- 0.03	99.15 +/- 1.85
<i>BCS1L</i>	52.14 +/- 8.4	100.0 +/- 0.0	100.0 +/- 0.0	98.85 +/- 5.02
<i>BLM</i>	55.78 +/- 7.53	100.0 +/- 0.0	100.0 +/- 0.0	99.74 +/- 0.69
<i>BMP2</i>	49.85 +/- 7.33	100.0 +/- 0.0	99.74 +/- 0.79	95.94 +/- 6.54
<i>BRAF</i>	53.15 +/- 6.81	99.99 +/- 0.05	99.51 +/- 1.19	97.12 +/- 2.41
<i>BRCA2</i>	55.39 +/- 7.18	100.0 +/- 0.0	100.0 +/- 0.01	99.5 +/- 1.2
<i>BRIP1</i>	53.94 +/- 7.38	99.98 +/- 0.04	99.98 +/- 0.04	99.13 +/- 1.61
<i>BTK</i>	41.19 +/- 15.32	99.77 +/- 1.38	94.24 +/- 11.58	68.11 +/- 37.01
<i>CBL</i>	53.02 +/- 7.87	100.0 +/- 0.0	99.95 +/- 0.18	98.83 +/- 3.14
<i>CCDC186</i>	55.41 +/- 7.05	100.0 +/- 0.0	99.98 +/- 0.15	99.46 +/- 1.59
<i>CCDC8</i>	48.56 +/- 8.09	99.99 +/- 0.04	99.72 +/- 1.28	97.33 +/- 8.13
<i>CDC45</i>	52.68 +/- 8.17	100.0 +/- 0.0	99.97 +/- 0.15	98.75 +/- 4.76
<i>CDC6</i>	55.81 +/- 8.32	100.0 +/- 0.0	100.0 +/- 0.03	99.67 +/- 0.81
<i>CDKN1C</i>	51.19 +/- 8.43	99.98 +/- 0.1	99.53 +/- 1.54	94.83 +/- 10.57
<i>CDT1</i>	54.7 +/- 8.66	100.0 +/- 0.0	99.82 +/- 0.9	98.12 +/- 5.89
<i>CEP152</i>	54.95 +/- 7.25	100.0 +/- 0.0	99.99 +/- 0.06	99.6 +/- 1.36
<i>CEP19</i>	52.74 +/- 8.12	99.98 +/- 0.04	99.95 +/- 0.11	98.88 +/- 2.94
<i>CEP290</i>	54.71 +/- 7.35	100.0 +/- 0.0	100.0 +/- 0.01	99.48 +/- 1.25
<i>CEP57</i>	55.77 +/- 7.64	100.0 +/- 0.0	99.97 +/- 0.19	99.77 +/- 0.96
<i>CEP63</i>	53.65 +/- 7.66	100.0 +/- 0.0	99.98 +/- 0.16	99.26 +/- 2.09
<i>CHD7</i>	53.94 +/- 7.9	100.0 +/- 0.0	99.89 +/- 0.31	98.68 +/- 2.47
<i>COL1A1</i>	49.25 +/- 8.22	100.0 +/- 0.0	99.92 +/- 0.45	97.55 +/- 7.02
<i>COL27A1</i>	49.84 +/- 7.76	100.0 +/- 0.0	99.74 +/- 1.2	96.55 +/- 7.07
<i>CPAP</i>	54.76 +/- 7.43	99.99 +/- 0.03	99.98 +/- 0.04	99.29 +/- 2.17
<i>CPE</i>	54.63 +/- 8.17	100.0 +/- 0.0	99.94 +/- 0.27	98.9 +/- 2.99
<i>CREBBP</i>	49.4 +/- 7.69	99.77 +/- 0.53	98.17 +/- 2.39	92.72 +/- 5.97
<i>CRIP1</i>	54.17 +/- 7.43	100.0 +/- 0.0	99.97 +/- 0.17	99.31 +/- 2.19
<i>CUL7</i>	50.3 +/- 8.96	100.0 +/- 0.0	99.93 +/- 0.43	97.74 +/- 8.2
<i>DHCR7</i>	53.79 +/- 9.03	99.97 +/- 0.12	99.79 +/- 0.86	98.57 +/- 5.94
<i>DNA2</i>	54.38 +/- 7.02	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.15
<i>DOK7</i>	53.65 +/- 9.47	100.0 +/- 0.0	99.57 +/- 1.29	96.09 +/- 7.47
<i>DONSON</i>	54.86 +/- 7.45	99.99 +/- 0.03	99.82 +/- 0.6	97.96 +/- 3.59
<i>EP300</i>	51.74 +/- 7.58	100.0 +/- 0.0	99.9 +/- 0.34	98.16 +/- 4.84
<i>EPHX1</i>	50.42 +/- 9.17	100.0 +/- 0.0	99.88 +/- 0.83	97.02 +/- 8.43
<i>ERCC4</i>	53.69 +/- 7.57	100.0 +/- 0.0	99.99 +/- 0.04	99.22 +/- 1.61
<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>FANCA</i>	52.83 +/- 7.93	100.0 +/- 0.0	99.94 +/- 0.4	98.6 +/- 4.06
<i>FANCB</i>	42.87 +/- 15.58	99.92 +/- 0.37	95.2 +/- 7.95	70.58 +/- 35.02
<i>FANCC</i>	53.44 +/- 7.77	100.0 +/- 0.0	100.0 +/- 0.0	99.08 +/- 3.14
<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>FANCG</i>	49.71 +/- 8.23	100.0 +/- 0.0	99.73 +/- 0.96	96.28 +/- 7.14
<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

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>FANCM</i>	54.19 +/- 7.34	100.0 +/- 0.0	99.99 +/- 0.05	99.51 +/- 1.33
<i>FGD1</i>	37.94 +/- 14.87	99.39 +/- 2.32	86.57 +/- 18.41	60.73 +/- 40.41
<i>FGF8</i>	48.03 +/- 8.31	99.95 +/- 0.31	97.89 +/- 4.61	88.67 +/- 10.01
<i>FGFR1</i>	52.48 +/- 8.64	100.0 +/- 0.0	99.9 +/- 0.47	97.99 +/- 6.49
<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>FOXP4</i>	49.07 +/- 8.12	100.0 +/- 0.0	99.65 +/- 1.21	95.16 +/- 8.73
<i>GGPS1</i>	53.42 +/- 7.76	100.0 +/- 0.0	99.99 +/- 0.04	99.03 +/- 2.33
<i>GH1</i>	52.76 +/- 8.45	100.0 +/- 0.0	100.0 +/- 0.0	98.86 +/- 6.48
<i>GHR</i>	55.05 +/- 7.73	99.79 +/- 0.43	99.68 +/- 0.45	98.59 +/- 2.88
<i>GHRHR</i>	50.29 +/- 9.05	100.0 +/- 0.0	99.91 +/- 0.58	97.98 +/- 6.32
<i>GHSR</i>	53.26 +/- 7.22	99.99 +/- 0.06	99.98 +/- 0.08	99.16 +/- 2.01
<i>GLI2</i>	54.49 +/- 8.68	100.0 +/- 0.0	99.94 +/- 0.29	98.24 +/- 5.05
<i>GLI3</i>	54.82 +/- 7.78	100.0 +/- 0.0	99.99 +/- 0.06	99.24 +/- 2.65
<i>GNAS</i>	47.03 +/- 7.06	99.83 +/- 0.62	94.78 +/- 6.99	83.4 +/- 8.94
<i>GPR161</i>	52.53 +/- 8.34	99.99 +/- 0.03	99.82 +/- 0.54	98.65 +/- 3.11
<i>H19^{1,2}</i>	0.05 +/- 0.07	0.0 +/- 0.0	0.0 +/- 0.0	0.0 +/- 0.0
<i>HDAC8</i>	40.43 +/- 15.06	99.86 +/- 0.74	92.95 +/- 12.03	66.34 +/- 37.72
<i>HESX1</i>	55.28 +/- 7.79	100.0 +/- 0.0	99.92 +/- 0.54	99.44 +/- 2.26
<i>HMGA2</i>	49.93 +/- 7.16	100.0 +/- 0.0	98.86 +/- 2.49	93.04 +/- 7.64
<i>HRAS</i>	49.95 +/- 9.48	99.95 +/- 0.17	99.17 +/- 2.84	94.67 +/- 10.68
<i>IDUA</i>	50.12 +/- 9.17	100.0 +/- 0.0	99.59 +/- 1.62	95.77 +/- 9.73
<i>IFT172</i>	51.73 +/- 7.98	100.0 +/- 0.0	99.98 +/- 0.09	98.92 +/- 3.1
<i>IGF1</i>	55.24 +/- 7.21	100.0 +/- 0.0	100.0 +/- 0.03	99.47 +/- 1.51
<i>IGF1R</i>	53.14 +/- 7.59	99.99 +/- 0.03	99.7 +/- 0.81	98.02 +/- 3.28
<i>IGF2</i>	42.18 +/- 7.41	99.81 +/- 0.32	95.42 +/- 4.56	81.02 +/- 12.5
<i>IGFALS</i>	59.53 +/- 10.25	100.0 +/- 0.0	99.99 +/- 0.07	98.81 +/- 6.2
<i>IGFBP1</i>	55.02 +/- 8.28	100.0 +/- 0.0	100.0 +/- 0.0	99.71 +/- 1.06
<i>IGFBP3</i>	53.27 +/- 8.11	100.0 +/- 0.0	99.96 +/- 0.25	98.54 +/- 4.4
<i>INPP5E</i>	50.81 +/- 8.98	100.0 +/- 0.0	99.5 +/- 2.37	95.13 +/- 8.84
<i>INSR</i>	51.65 +/- 7.72	99.97 +/- 0.13	99.74 +/- 0.87	97.19 +/- 4.33
<i>IRS1</i>	52.15 +/- 7.6	100.0 +/- 0.03	99.66 +/- 0.91	97.29 +/- 4.83
<i>KANSL1</i>	58.98 +/- 10.17	100.0 +/- 0.0	99.83 +/- 0.79	98.09 +/- 4.41
<i>KDM3B</i>	52.76 +/- 7.55	100.0 +/- 0.01	99.78 +/- 0.42	97.95 +/- 3.51
<i>KDM6A</i>	41.98 +/- 15.75	99.88 +/- 0.7	93.58 +/- 9.68	68.64 +/- 35.84
<i>KMT2D</i>	49.24 +/- 8.18	100.0 +/- 0.01	99.7 +/- 0.9	95.92 +/- 7.53
<i>KRAS</i>	55.79 +/- 7.63	99.98 +/- 0.04	99.92 +/- 0.22	99.08 +/- 2.05
<i>KSR2</i>	51.81 +/- 8.43	99.99 +/- 0.04	99.9 +/- 0.2	98.32 +/- 4.03
<i>LARP7</i>	54.24 +/- 7.46	100.0 +/- 0.0	100.0 +/- 0.0	99.5 +/- 0.98
<i>LEP</i>	50.68 +/- 8.43	100.0 +/- 0.0	99.94 +/- 0.26	97.7 +/- 4.21
<i>LEPR</i>	55.98 +/- 7.55	100.0 +/- 0.0	99.98 +/- 0.09	99.54 +/- 1.12
<i>LFNG</i>	49.29 +/- 9.01	99.99 +/- 0.04	98.6 +/- 2.74	92.53 +/- 9.68
<i>LHX3</i>	51.17 +/- 8.78	100.0 +/- 0.0	99.96 +/- 0.24	97.67 +/- 6.54
<i>LHX4</i>	51.44 +/- 8.61	99.84 +/- 0.06	99.77 +/- 0.39	98.04 +/- 3.37
<i>LIG1</i>	50.82 +/- 7.9	100.0 +/- 0.0	99.9 +/- 0.55	97.5 +/- 5.57
<i>LIG4</i>	54.4 +/- 7.23	100.0 +/- 0.01	99.4 +/- 0.53	98.29 +/- 1.27

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>LZTR1</i>	51.86 +/- 8.39	100.0 +/- 0.0	99.95 +/- 0.34	98.54 +/- 6.12
<i>MAGEL2</i>	53.34 +/- 8.42	100.0 +/- 0.0	99.95 +/- 0.23	97.97 +/- 4.46
<i>MAP2K1</i>	51.3 +/- 8.2	100.0 +/- 0.0	99.8 +/- 0.72	97.54 +/- 4.52
<i>MAP2K2</i>	50.63 +/- 8.38	100.0 +/- 0.0	99.73 +/- 1.15	95.96 +/- 8.4
<i>MAPK1</i>	54.98 +/- 7.88	100.0 +/- 0.0	99.91 +/- 0.39	98.8 +/- 2.28
<i>MC4R</i>	54.34 +/- 8.69	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.55
<i>MCM5</i>	51.58 +/- 8.29	100.0 +/- 0.0	99.92 +/- 0.49	98.24 +/- 5.13
<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>MRAP2</i>	53.19 +/- 8.23	100.0 +/- 0.0	100.0 +/- 0.0	98.97 +/- 2.9
<i>MRAS</i>	52.12 +/- 7.74	100.0 +/- 0.0	99.84 +/- 0.85	98.34 +/- 4.13
<i>MYT1L</i>	55.87 +/- 8.0	100.0 +/- 0.0	99.97 +/- 0.16	99.31 +/- 2.13
<i>NBN</i>	55.05 +/- 7.63	100.0 +/- 0.0	99.99 +/- 0.07	99.44 +/- 1.07
<i>NIPBL</i>	54.63 +/- 6.99	100.0 +/- 0.03	99.82 +/- 0.35	98.43 +/- 1.48
<i>NLRP2</i>	53.02 +/- 8.4	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 3.01
<i>NLRP5</i>	53.43 +/- 7.87	100.0 +/- 0.0	99.98 +/- 0.12	99.3 +/- 2.13
<i>NLRP7</i>	52.98 +/- 8.28	100.0 +/- 0.0	99.98 +/- 0.15	99.18 +/- 3.73
<i>NOTCH2</i>	53.16 +/- 7.9	100.0 +/- 0.0	99.88 +/- 0.23	98.25 +/- 2.58
<i>NR0B2</i>	49.06 +/- 8.11	100.0 +/- 0.0	100.0 +/- 0.0	98.28 +/- 5.43
<i>NRAS</i>	55.99 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 0.83
<i>NTRK2</i>	55.32 +/- 7.63	100.0 +/- 0.0	99.97 +/- 0.19	99.56 +/- 0.99
<i>OBSL1</i>	52.21 +/- 8.76	100.0 +/- 0.0	99.91 +/- 0.57	97.56 +/- 7.48
<i>ORC1</i>	53.6 +/- 7.96	100.0 +/- 0.0	100.0 +/- 0.0	99.49 +/- 1.9
<i>ORC4</i>	54.8 +/- 7.74	100.0 +/- 0.0	99.96 +/- 0.19	99.03 +/- 1.62
<i>ORC6</i>	55.41 +/- 8.38	100.0 +/- 0.0	100.0 +/- 0.03	99.27 +/- 2.38
<i>OSGEP</i>	52.23 +/- 8.21	99.96 +/- 0.05	99.95 +/- 0.05	99.49 +/- 1.5
<i>OTX2</i>	52.15 +/- 8.58	100.0 +/- 0.0	100.0 +/- 0.0	98.91 +/- 2.84
<i>PADI6</i>	51.01 +/- 8.05	99.93 +/- 0.28	99.19 +/- 1.25	95.97 +/- 4.36
<i>PALB2</i>	53.71 +/- 7.7	100.0 +/- 0.0	100.0 +/- 0.03	98.7 +/- 2.37
<i>PAPPA2</i>	52.91 +/- 7.73	100.0 +/- 0.0	99.99 +/- 0.09	99.14 +/- 2.74
<i>PCNT</i>	55.98 +/- 8.01	100.0 +/- 0.0	99.98 +/- 0.16	99.27 +/- 2.67
<i>PCSK1</i>	54.54 +/- 7.7	100.0 +/- 0.0	100.0 +/- 0.0	99.41 +/- 2.0
<i>PHF6</i>	44.03 +/- 16.05	99.98 +/- 0.08	95.48 +/- 8.69	72.36 +/- 32.74
<i>PIK3R1</i>	52.65 +/- 7.75	99.97 +/- 0.1	99.48 +/- 0.65	97.31 +/- 2.35
<i>PISD</i>	55.09 +/- 8.55	100.0 +/- 0.0	99.99 +/- 0.07	99.27 +/- 3.38
<i>PITX2</i>	52.06 +/- 7.09	100.0 +/- 0.0	99.92 +/- 0.52	98.78 +/- 4.6
<i>PLAG1</i>	56.16 +/- 8.71	100.0 +/- 0.0	99.99 +/- 0.03	99.4 +/- 1.26
<i>PNPLA6</i>	52.2 +/- 8.63	100.0 +/- 0.0	99.94 +/- 0.24	97.88 +/- 5.9
<i>POC1A</i>	53.79 +/- 8.86	100.0 +/- 0.0	100.0 +/- 0.01	98.95 +/- 4.26
<i>POMC</i>	56.16 +/- 9.67	100.0 +/- 0.0	100.0 +/- 0.0	99.19 +/- 2.65
<i>POP1</i>	53.52 +/- 7.6	100.0 +/- 0.0	100.0 +/- 0.0	99.23 +/- 1.82
<i>POU1F1</i>	55.33 +/- 7.88	100.0 +/- 0.0	99.99 +/- 0.03	99.19 +/- 2.4
<i>PPARG</i>	54.63 +/- 7.88	100.0 +/- 0.0	100.0 +/- 0.0	99.58 +/- 2.32
<i>PPP1CB</i>	55.13 +/- 7.65	100.0 +/- 0.03	99.76 +/- 0.67	97.96 +/- 2.01
<i>PPP3CA</i>	54.53 +/- 7.45	100.0 +/- 0.0	99.94 +/- 0.32	98.17 +/- 3.89
<i>PRMT7</i>	54.41 +/- 7.96	100.0 +/- 0.0	99.99 +/- 0.06	98.92 +/- 3.91
<i>PROKR2</i>	53.08 +/- 8.32	100.0 +/- 0.0	100.0 +/- 0.0	99.07 +/- 2.92

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>PROP1</i>	46.73 +/- 7.13	100.0 +/- 0.0	99.7 +/- 0.91	94.59 +/- 6.62
<i>PTPN11</i>	55.1 +/- 7.94	100.0 +/- 0.0	99.96 +/- 0.18	99.28 +/- 1.99
<i>PUF60</i>	48.41 +/- 8.66	100.0 +/- 0.0	99.9 +/- 0.43	96.0 +/- 6.95
<i>RAD21</i>	54.67 +/- 7.16	100.0 +/- 0.01	99.66 +/- 0.66	97.88 +/- 1.66
<i>RAD51</i>	53.55 +/- 7.77	100.0 +/- 0.0	99.94 +/- 0.37	99.15 +/- 2.44
<i>RAF1</i>	54.46 +/- 7.88	100.0 +/- 0.01	99.97 +/- 0.19	98.64 +/- 4.74
<i>RALA</i>	54.19 +/- 7.51	100.0 +/- 0.0	99.96 +/- 0.19	98.34 +/- 2.92
<i>RAP1B</i>	53.55 +/- 7.68	100.0 +/- 0.0	99.8 +/- 0.55	98.16 +/- 2.22
<i>RAPSN</i>	51.01 +/- 7.7	100.0 +/- 0.0	100.0 +/- 0.0	98.67 +/- 3.1
<i>RASA2</i>	54.15 +/- 7.33	100.0 +/- 0.0	99.93 +/- 0.25	99.15 +/- 1.33
<i>RBBP8</i>	54.25 +/- 7.39	100.0 +/- 0.0	99.96 +/- 0.14	99.0 +/- 2.1
<i>RIT1</i>	53.38 +/- 7.23	100.0 +/- 0.0	99.97 +/- 0.09	99.0 +/- 2.2
<i>RNPC3</i>	54.08 +/- 7.18	100.0 +/- 0.0	100.0 +/- 0.02	98.9 +/- 2.52
<i>RNU4ATAC</i>	58.08 +/- 10.68	100.0 +/- 0.0	100.0 +/- 0.0	99.88 +/- 0.8
<i>ROR2</i>	53.88 +/- 7.85	99.97 +/- 0.05	99.93 +/- 0.19	98.7 +/- 4.19
<i>RPL10</i>	42.12 +/- 16.4	99.89 +/- 0.5	92.79 +/- 13.45	68.91 +/- 37.15
<i>RPS6KA3</i>	43.46 +/- 16.35	99.83 +/- 0.37	94.54 +/- 8.88	70.62 +/- 33.96
<i>RRAS</i>	46.2 +/- 7.88	100.0 +/- 0.0	99.3 +/- 2.07	93.15 +/- 12.2
<i>RRAS2</i>	53.56 +/- 7.43	100.0 +/- 0.0	99.78 +/- 1.01	98.47 +/- 4.02
<i>RTTN</i>	55.51 +/- 7.61	100.0 +/- 0.0	99.99 +/- 0.06	99.29 +/- 1.69
<i>SAMD9</i>	54.36 +/- 7.32	100.0 +/- 0.0	100.0 +/- 0.0	99.4 +/- 1.64
<i>SDCCAG8</i>	55.16 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.53 +/- 1.56
<i>SETD5</i>	52.26 +/- 7.48	99.98 +/- 0.12	99.55 +/- 0.57	96.94 +/- 2.95
<i>SGMS2</i>	55.03 +/- 7.45	100.0 +/- 0.0	99.96 +/- 0.19	99.1 +/- 1.73
<i>SH2B1</i>	48.75 +/- 8.43	99.95 +/- 0.08	99.73 +/- 1.25	96.28 +/- 8.0
<i>SHOC2</i>	54.91 +/- 7.5	100.0 +/- 0.0	99.92 +/- 0.29	99.21 +/- 0.87
<i>SHOX²</i>	48.04 +/- 7.12	93.36 +/- 2.12	89.11 +/- 1.07	86.51 +/- 5.23
<i>SHOX2</i>	49.97 +/- 6.97	100.0 +/- 0.0	99.78 +/- 0.66	96.58 +/- 6.3
<i>SIM1</i>	55.25 +/- 7.69	100.0 +/- 0.0	100.0 +/- 0.03	99.48 +/- 1.86
<i>SLX4</i>	52.78 +/- 7.94	100.0 +/- 0.0	99.98 +/- 0.1	98.82 +/- 4.37
<i>SMARCA2</i>	54.21 +/- 8.21	100.0 +/- 0.0	99.97 +/- 0.2	99.15 +/- 2.54
<i>SMARCA11</i>	52.78 +/- 7.67	100.0 +/- 0.0	99.97 +/- 0.21	98.85 +/- 3.15
<i>SMARCE1</i>	55.14 +/- 7.79	100.0 +/- 0.0	100.0 +/- 0.0	99.5 +/- 1.36
<i>SMC1A</i>	41.05 +/- 15.95	99.84 +/- 0.83	92.32 +/- 13.46	66.34 +/- 38.2
<i>SMC3</i>	55.91 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.72 +/- 0.77
<i>SOS1</i>	53.84 +/- 7.54	100.0 +/- 0.0	99.69 +/- 0.98	97.96 +/- 3.04
<i>SOS2</i>	54.73 +/- 7.38	99.99 +/- 0.04	99.78 +/- 0.76	98.52 +/- 2.17
<i>SOX11</i>	50.59 +/- 7.32	99.98 +/- 0.04	99.47 +/- 1.41	94.82 +/- 6.56
<i>SOX2</i>	48.55 +/- 6.94	100.0 +/- 0.0	99.81 +/- 0.54	95.32 +/- 7.31
<i>SOX3</i>	37.79 +/- 14.98	98.82 +/- 5.4	85.66 +/- 21.15	60.83 +/- 40.37
<i>SPRED1</i>	54.98 +/- 7.33	100.0 +/- 0.0	99.95 +/- 0.23	99.04 +/- 1.38
<i>SPRED2</i>	53.17 +/- 7.71	99.99 +/- 0.09	99.81 +/- 0.74	97.61 +/- 4.14
<i>SRCAP</i>	50.57 +/- 8.06	100.0 +/- 0.0	99.95 +/- 0.31	97.99 +/- 5.68
<i>STAT5B</i>	51.92 +/- 7.21	99.98 +/- 0.12	99.83 +/- 0.65	97.81 +/- 3.19
<i>TALDO1</i>	51.31 +/- 9.35	100.0 +/- 0.0	99.73 +/- 0.81	95.51 +/- 7.1
<i>TBCE</i>	55.02 +/- 8.35	100.0 +/- 0.0	100.0 +/- 0.0	99.46 +/- 1.54

Gen	Middelsekventeringsdybde [x]	Andel >= 15x [%]	Andel >= 20x [%]	Andel >= 30x [%]
TBX19	53.67 +/- 8.58	100.0 +/- 0.02	99.86 +/- 0.83	98.93 +/- 4.4
TBX2	49.08 +/- 8.67	99.96 +/- 0.28	99.15 +/- 2.02	94.3 +/- 9.2
TBX3	52.11 +/- 7.31	100.0 +/- 0.0	99.98 +/- 0.13	98.93 +/- 4.06
THRB	54.02 +/- 7.6	100.0 +/- 0.0	99.99 +/- 0.06	99.09 +/- 2.66
TOP3A	52.87 +/- 8.09	100.0 +/- 0.0	99.98 +/- 0.12	98.96 +/- 2.87
TRIM32	55.0 +/- 8.0	100.0 +/- 0.0	99.83 +/- 0.45	98.45 +/- 2.54
TRIM37	56.1 +/- 7.64	100.0 +/- 0.0	99.97 +/- 0.21	99.52 +/- 1.68
TRMT10A	55.83 +/- 7.39	100.0 +/- 0.0	99.98 +/- 0.15	99.52 +/- 1.36
TTC8	55.08 +/- 7.81	100.0 +/- 0.0	99.87 +/- 0.7	99.14 +/- 2.09
TUB	52.68 +/- 7.98	100.0 +/- 0.0	99.8 +/- 0.74	98.28 +/- 4.17
UBE2T	54.68 +/- 8.44	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.7
VPS13B	55.06 +/- 7.31	100.0 +/- 0.0	100.0 +/- 0.01	99.62 +/- 1.05
VPS50	54.51 +/- 7.61	100.0 +/- 0.0	99.96 +/- 0.14	99.25 +/- 1.99
WDPCP	55.08 +/- 7.68	100.0 +/- 0.0	99.96 +/- 0.16	99.43 +/- 1.45
WRN	54.75 +/- 7.13	99.98 +/- 0.07	99.8 +/- 0.25	98.66 +/- 1.68
XRCC4	56.31 +/- 7.21	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.17
ZFP57	50.37 +/- 8.67	100.0 +/- 0.0	100.0 +/- 0.0	98.39 +/- 4.07
ZNF668	56.65 +/- 10.34	100.0 +/- 0.0	99.86 +/- 0.58	98.06 +/- 6.14

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