AALBORG UNIVERSITETSHOSPITAL
– i gode hænder

Aalborg Universitetshospital
Nordjyllands Gensekventeringscenter
Reberbansgade 15, 9000 Aalborg

Monogen diabetes

Panelbeskrivelse

Navn: Monogen diabetes **version:** 1.0 **Ibrugtagningsdato:** 31/10-2023

Metodebeskrivelse: Analysen udføres ved helgenomsekventering (WGS) med Illumina PCR free library prep (tagmentation) og Illumina sekventering (Novaseq). Efterfølgende fortages in silico filtrering af data til relevante genregioner baseret på nedenstående tabel. Middelsekventeringsdybden er minimum 30X, og minimumsandelen af de kodede regioner der dækket minimum 10X er 90 %.

Genliste

Gener hvor middelsekventeringsdybden er < 30X er markeret med[1]. Gener, hvor andelen af de kodede regioner der dækket minimum 10X er < 90 % er markeret med[2]. Værdier er angivet med +/- standardafvigelse.

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
ABCC8	51.36 +/- 8.64	100.0 +/- 0.0	99.98 +/- 0.06	98.29 +/- 6.33
AGPAT2	50.39 +/- 9.34	100.0 +/- 0.0	99.9 +/- 0.4	96.14 +/- 7.48
AGPS	54.51 +/- 7.23	100.0 +/- 0.02	99.91 +/- 0.1	99.17 +/- 1.64
AIRE	50.75 +/- 8.1	100.0 +/- 0.0	99.96 +/- 0.3	97.68 +/- 6.97
AKT2	48.67 +/- 8.08	100.0 +/- 0.0	99.66 +/- 1.37	95.97 +/- 8.15
ALMS1	53.21 +/- 7.37	100.0 +/- 0.0	99.98 +/- 0.06	99.19 +/- 1.67
APPL1	55.21 +/- 7.16	100.0 +/- 0.0	99.98 +/- 0.1	99.23 +/- 1.42
BLK	52.94 +/- 8.42	100.0 +/- 0.0	100.0 +/- 0.0	98.79 +/- 4.03
BSCL2	52.37 +/- 8.4	100.0 +/- 0.0	99.99 +/- 0.06	98.79 +/- 6.57
CAV1	54.2 +/- 8.26	100.0 +/- 0.0	99.95 +/- 0.33	98.9 +/- 3.34
CEL	49.96 +/- 9.5	99.68 +/- 1.45	97.36 +/- 4.8	88.97 +/- 10.31
CIDEC	52.09 +/- 8.06	100.0 +/- 0.0	100.0 +/- 0.0	98.81 +/- 5.12
CISD2	56.95 +/- 7.98	100.0 +/- 0.0	99.99 +/- 0.06	99.34 +/- 1.62
CNOT1	54.27 +/- 7.29	100.0 +/- 0.0	100.0 +/- 0.01	99.2 +/- 1.67
COQ2	55.89 +/- 7.91	100.0 +/- 0.0	100.0 +/- 0.0	99.01 +/- 3.42
COQ9	51.71 +/- 8.51	100.0 +/- 0.0	100.0 +/- 0.0	98.97 +/- 2.62
DCAF17	54.66 +/- 7.54	100.0 +/- 0.0	100.0 +/- 0.01	99.18 +/- 1.49
DMXL2	55.63 +/- 7.45	100.0 +/- 0.0	100.0 +/- 0.01	99.54 +/- 1.34
DNAJC3	54.53 +/- 7.34	99.93 +/- 0.15	99.9 +/- 0.17	99.24 +/- 2.07
DYRK1B	47.23 +/- 8.19	100.0 +/- 0.0	99.72 +/- 1.05	93.94 +/- 9.73
EIF2AK3	54.99 +/- 7.71	99.95 +/- 0.05	99.86 +/- 0.42	99.19 +/- 2.11
EIF2B1	51.34 +/- 7.4	99.83 +/- 0.32	99.35 +/- 0.81	96.48 +/- 4.13
EIF2S3	42.42 +/- 15.81	99.9 +/- 0.6	93.21 +/- 13.86	70.79 +/- 34.6
FICD	54.54 +/- 8.39	100.0 +/- 0.0	99.95 +/- 0.33	99.06 +/- 2.95
FOXP3	38.43 +/- 14.73	99.72 +/- 0.89	88.99 +/- 17.07	63.48 +/- 39.93
GATA4	51.2 +/- 7.57	100.0 +/- 0.0	99.98 +/- 0.09	98.1 +/- 4.44
GATA6	53.04 +/- 7.49	100.0 +/- 0.0	99.78 +/- 1.08	96.74 +/- 7.21
GCK	54.14 +/- 9.34	100.0 +/- 0.0	100.0 +/- 0.0	98.72 +/- 5.18
GLIS3	52.93 +/- 7.55	100.0 +/- 0.0	99.96 +/- 0.17	98.64 +/- 2.97
GLUD1	54.16 +/- 7.47	100.0 +/- 0.0	99.8 +/- 0.88	98.04 +/- 3.48
HADH	53.52 +/- 7.74	100.0 +/- 0.0	99.87 +/- 0.89	98.89 +/- 4.3

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
HAMP	48.15 +/- 8.85	100.0 +/- 0.0	100.0 +/- 0.0	97.47 +/- 6.47
HFE	53.83 +/- 7.72	100.0 +/- 0.0	100.0 +/- 0.0	99.17 +/- 1.97
HJV	50.52 +/- 8.0	100.0 +/- 0.0	99.98 +/- 0.12	98.33 +/- 4.4
HNF1A	49.75 +/- 7.93	100.0 +/- 0.0	99.82 +/- 1.24	97.61 +/- 7.48
HNF1B	49.89 +/- 7.94	100.0 +/- 0.0	99.66 +/- 1.6	96.98 +/- 6.57
HNF4A	49.72 +/- 8.43	99.99 +/- 0.06	99.81 +/- 0.5	96.56 +/- 7.06
IER3IP1	54.86 +/- 7.74	99.98 +/- 0.04	99.93 +/- 0.26	98.89 +/- 2.17
IL2RA	53.49 +/- 8.2	100.0 +/- 0.0	99.99 +/- 0.09	99.35 +/- 1.9
INS	53.25 +/- 9.81	100.0 +/- 0.0	100.0 +/- 0.03	98.58 +/- 4.94
INSR	51.64 +/- 7.63	99.97 +/- 0.13	99.74 +/- 0.87	97.2 +/- 4.33
KCNJ11	52.82 +/- 8.15	100.0 +/- 0.0	99.92 +/- 0.4	98.46 +/- 4.78
KLF11	52.98 +/- 7.55	100.0 +/- 0.02	99.64 +/- 0.95	97.23 +/- 3.92
LIPC	54.93 +/- 7.72	100.0 +/- 0.0	99.95 +/- 0.31	99.45 +/- 1.62
LMNA	49.81 +/- 8.27	100.0 +/- 0.0	99.96 +/- 0.16	97.29 +/- 6.96
LPL	54.99 +/- 7.64	100.0 +/- 0.0	100.0 +/- 0.0	99.67 +/- 1.12
LRBA	54.76 +/- 7.25	99.99 +/- 0.03	99.99 +/- 0.03	99.49 +/- 0.96
MNX1	45.28 +/- 8.84	99.89 +/- 0.57	98.14 +/- 4.43	88.84 +/- 16.68
MT-TP	55.4 +/- 7.83	100.0 +/- 0.0	99.98 +/- 0.12	99.39 +/- 1.55
NEUROD1	52.4 +/- 6.68	100.0 +/- 0.0	100.0 +/- 0.01	99.0 +/- 2.59
NEUROG3	51.9 +/- 9.17	100.0 +/- 0.0	99.95 +/- 0.24	97.74 +/- 7.63
NKX2-2	48.35 +/- 7.54	100.0 +/- 0.0	99.34 +/- 1.61	94.05 +/- 8.54
ONECUT1	51.41 +/- 7.28	100.0 +/- 0.0	99.9 +/- 0.48	97.3 +/- 5.69
PAX4	53.38 +/- 8.34	100.0 +/- 0.0	99.98 +/- 0.11	98.84 +/- 3.12
PAX6	52.35 +/- 7.1	100.0 +/- 0.0	99.97 +/- 0.13	98.35 +/- 3.15
PCBD1	50.66 +/- 9.25	100.0 +/- 0.0	99.95 +/- 0.23	97.47 +/- 7.81
PDIA6	54.41 +/- 7.78	100.0 +/- 0.0	99.95 +/- 0.31	99.22 +/- 1.94
PDX1	46.09 +/- 7.2	99.95 +/- 0.14	97.98 +/- 3.08	88.72 +/- 9.85
PIK3R1	52.74 +/- 7.63	99.98 +/- 0.09	99.59 +/- 0.57	97.62 +/- 2.31
PLIN1	51.38 +/- 7.43	100.0 +/- 0.0	100.0 +/- 0.01	98.02 +/- 4.5
POLD1	52.38 +/- 8.54	100.0 +/- 0.0	99.9 +/- 0.35	97.4 +/- 5.08
PPARG	54.63 +/- 7.85	100.0 +/- 0.0	100.0 +/- 0.0	99.58 +/- 2.32
PPP1R15B	53.52 +/- 7.42	100.0 +/- 0.0	100.0 +/- 0.01	99.41 +/- 1.95
PTF1A	47.32 +/- 8.53	99.98 +/- 0.06	99.45 +/- 1.67	92.16 +/- 13.06
RFX6	54.6 +/- 7.37	100.0 +/- 0.0	99.92 +/- 0.52	99.3 +/- 1.82
SLC16A1	55.85 +/- 7.13	100.0 +/- 0.0	100.0 +/- 0.0	99.22 +/- 1.72
SLC19A2	54.84 +/- 7.74	100.0 +/- 0.0	100.0 +/- 0.0	99.46 +/- 1.3
SLC29A3	51.62 +/- 8.71	100.0 +/- 0.0	99.97 +/- 0.21	98.33 +/- 3.8
SLC2A2	55.5 +/- 7.45	100.0 +/- 0.0	100.0 +/- 0.01	99.71 +/- 1.18
SLC40A1	55.61 +/- 7.87	100.0 +/- 0.0	99.99 +/- 0.07	99.57 +/- 1.31
STAT1	56.34 +/- 7.27	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.24
STAT3	51.76 +/- 7.87	100.0 +/- 0.0	99.97 +/- 0.21	98.6 +/- 4.03
TFR2	48.57 +/- 7.97	100.0 +/- 0.0	99.76 +/- 1.15	95.88 +/- 9.14
TRMT10A	55.86 +/- 7.36	100.0 +/- 0.0	99.98 +/- 0.15	99.52 +/- 1.36
UCP2	50.29 +/- 8.48	100.0 +/- 0.0	99.99 +/- 0.06	97.44 +/- 7.46
WFS1	54.5 +/- 8.88	100.0 +/- 0.0	99.97 +/- 0.11	98.44 +/- 6.43
ZBTB20	54.47 +/- 7.43	100.0 +/- 0.0	99.99 +/- 0.05	99.31 +/- 1.64

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
ZFP57	50.34 +/- 8.55	100.0 +/- 0.0	100.0 +/- 0.0	98.39 +/- 4.07
ZMPSTE24	55.06 +/- 6.95	100.0 +/- 0.0	100.0 +/- 0.0	99.62 +/- 1.17
ZNF808	56.59 +/- 8.22	100.0 +/- 0.0	100.0 +/- 0.0	99.63 +/- 0.87

For supplerende oplysninger vedrørende analysen kan afdelingen kontaktes på mail: mol-dia@rn.dk. Rapport genereret: 09/09-2024