AALBORG UNIVERSITETSHOSPITAL
– i gode hænder

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Uafklaret cytopeni & Uafklaret hæmolytisk anæmi

Panelbeskrivelse

Navn: Uafklaret cytopeni & Uafklaret hæmolytisk anæmi **version:** 1.0 **Ibrugtagningsdato:** 09/12-2022

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 [%]
ABCC4	54.72 +/- 7.84	100.0 +/- 0.0	100.0 +/- 0.02	99.42 +/- 1.78
ABCG5	52.38 +/- 7.41	100.0 +/- 0.0	99.96 +/- 0.21	98.52 +/- 4.73
ABCG8	53.42 +/- 8.21	99.99 +/- 0.03	99.94 +/- 0.33	98.68 +/- 5.02
ACTB	52.7 +/- 9.38	100.0 +/- 0.0	99.97 +/- 0.13	98.35 +/- 4.52
ACTN1	51.27 +/- 8.06	100.0 +/- 0.0	99.74 +/- 1.39	97.31 +/- 6.88
ADAMTS13	50.65 +/- 8.67	100.0 +/- 0.0	99.78 +/- 0.98	96.81 +/- 6.59
ANKRD26	54.36 +/- 7.35	100.0 +/- 0.0	99.99 +/- 0.09	99.36 +/- 1.42
ANO6	54.35 +/- 7.16	100.0 +/- 0.0	99.98 +/- 0.11	99.37 +/- 1.33
AP3B1	55.4 +/- 7.36	100.0 +/- 0.0	100.0 +/- 0.03	99.47 +/- 1.56
AP3D1	54.8 +/- 8.84	100.0 +/- 0.0	99.96 +/- 0.17	98.2 +/- 6.19
ARPC1B	52.45 +/- 8.75	100.0 +/- 0.0	99.86 +/- 0.58	97.79 +/- 8.11
BLOC1S3	48.86 +/- 8.08	100.0 +/- 0.0	99.93 +/- 0.4	97.44 +/- 6.09
BLOC1S6	56.96 +/- 7.12	100.0 +/- 0.0	100.0 +/- 0.0	99.83 +/- 0.43
CDC42	54.71 +/- 7.9	99.89 +/- 0.09	99.77 +/- 0.22	98.97 +/- 2.19
CYCS	56.18 +/- 7.18	100.0 +/- 0.0	99.99 +/- 0.04	99.81 +/- 0.53
DIAPH1	51.35 +/- 8.06	99.96 +/- 0.19	99.39 +/- 1.31	96.37 +/- 4.26
DTNBP1	53.31 +/- 8.19	100.0 +/- 0.0	100.0 +/- 0.0	97.88 +/- 5.83
ETV6	49.91 +/- 7.75	100.0 +/- 0.0	99.85 +/- 0.66	96.75 +/- 5.0
F10	52.31 +/- 8.31	100.0 +/- 0.0	99.82 +/- 1.22	98.51 +/- 5.12
F11	54.29 +/- 7.59	100.0 +/- 0.0	100.0 +/- 0.0	99.37 +/- 1.46
F12	53.05 +/- 8.93	100.0 +/- 0.0	99.92 +/- 0.35	97.86 +/- 8.03
F13A1	54.58 +/- 7.77	100.0 +/- 0.0	100.0 +/- 0.01	99.46 +/- 2.44
F13B	56.32 +/- 7.13	100.0 +/- 0.0	100.0 +/- 0.0	99.88 +/- 0.44
F2	51.5 +/- 8.06	100.0 +/- 0.0	99.99 +/- 0.06	98.42 +/- 6.71
F5	53.31 +/- 7.6	100.0 +/- 0.0	99.97 +/- 0.22	98.82 +/- 2.72
F7	52.39 +/- 8.42	100.0 +/- 0.0	99.97 +/- 0.15	98.71 +/- 3.41
F8	41.99 +/- 15.81	99.94 +/- 0.37	94.09 +/- 10.17	68.25 +/- 36.91
F9	41.38 +/- 15.1	99.96 +/- 0.17	93.5 +/- 12.07	68.78 +/- 35.37
FERMT3	51.79 +/- 8.56	100.0 +/- 0.0	99.84 +/- 0.73	97.08 +/- 6.16
FGA	52.05 +/- 8.18	100.0 +/- 0.0	99.99 +/- 0.06	98.86 +/- 3.04
FGB	54.68 +/- 7.27	100.0 +/- 0.0	100.0 +/- 0.0	99.5 +/- 1.97

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
FGG	55.31 +/- 8.18	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 1.09
FLI1	52.82 +/- 7.92	98.94 +/- 3.36	98.47 +/- 3.5	96.09 +/- 5.32
FLNA	42.42 +/- 16.9	99.82 +/- 0.82	92.1 +/- 15.58	67.32 +/- 37.82
FYB1	53.76 +/- 7.27	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 1.28
GATA1	37.58 +/- 14.64	99.85 +/- 0.91	88.03 +/- 18.64	61.8 +/- 42.41
GFI1B	52.72 +/- 8.69	100.0 +/- 0.0	99.96 +/- 0.24	98.47 +/- 4.34
GGCX	53.45 +/- 7.74	100.0 +/- 0.0	100.0 +/- 0.03	99.36 +/- 1.89
GNE	54.84 +/- 7.57	99.98 +/- 0.04	99.98 +/- 0.04	99.41 +/- 1.23
GP1BA	48.42 +/- 8.44	99.87 +/- 0.27	98.18 +/- 1.5	93.59 +/- 6.77
GP1BB	54.66 +/- 10.37	100.0 +/- 0.0	99.86 +/- 0.94	97.17 +/- 11.47
GP6	52.78 +/- 8.88	100.0 +/- 0.0	100.0 +/- 0.0	98.71 +/- 4.14
GP9	52.57 +/- 9.02	100.0 +/- 0.0	100.0 +/- 0.0	98.71 +/- 4.22
HOXA11	52.28 +/- 8.64	100.0 +/- 0.0	99.98 +/- 0.15	98.37 +/- 5.12
HPS1	52.48 +/- 8.51	100.0 +/- 0.0	99.95 +/- 0.34	98.62 +/- 4.65
HPS3	54.51 +/- 7.71	100.0 +/- 0.0	99.95 +/- 0.2	98.99 +/- 2.03
HPS4	53.55 +/- 7.82	99.96 +/- 0.06	99.87 +/- 0.12	98.79 +/- 3.14
HPS5	55.41 +/- 7.19	100.0 +/- 0.0	100.0 +/- 0.0	99.69 +/- 0.82
HPS6	50.72 +/- 8.59	100.0 +/- 0.0	99.94 +/- 0.4	97.58 +/- 7.48
HRG	52.52 +/- 8.63	100.0 +/- 0.0	99.88 +/- 0.64	98.1 +/- 5.7
IKZF5	54.11 +/- 7.15	100.0 +/- 0.0	99.99 +/- 0.07	98.99 +/- 2.82
ITGA2B	49.48 +/- 7.95	100.0 +/- 0.0	99.95 +/- 0.36	97.84 +/- 6.39
ITGB3	52.94 +/- 7.61	100.0 +/- 0.0	99.92 +/- 0.4	98.76 +/- 4.03
KDSR	54.89 +/- 7.31	100.0 +/- 0.0	99.92 +/- 0.36	99.22 +/- 1.71
KNG1	54.58 +/- 7.48	100.0 +/- 0.0	99.98 +/- 0.1	99.47 +/- 1.48
LMAN1	55.98 +/- 7.58	100.0 +/- 0.0	99.99 +/- 0.09	99.16 +/- 1.86
LYST	55.53 +/- 7.3	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.01
MCFD2	53.56 +/- 7.43	99.99 +/- 0.06	99.89 +/- 0.35	99.15 +/- 2.69
MECOM	54.63 +/- 7.54	100.0 +/- 0.0	99.94 +/- 0.26	99.11 +/- 1.61
MPIG6B	51.3 +/- 8.07	100.0 +/- 0.0	99.98 +/- 0.16	98.54 +/- 3.84
MPL	52.02 +/- 8.45	100.0 +/- 0.0	99.99 +/- 0.04	98.5 +/- 3.75
MYH9	52.51 +/- 7.98	100.0 +/- 0.0	99.98 +/- 0.14	98.64 +/- 4.25
NBEA	55.43 +/- 7.51	100.0 +/- 0.03	99.83 +/- 0.56	98.67 +/- 2.14
NBEAL2	51.81 +/- 8.79	100.0 +/- 0.0	99.94 +/- 0.36	97.88 +/- 6.27
P2RY12	55.28 +/- 7.88	100.0 +/- 0.0	99.98 +/- 0.07	99.16 +/- 2.4
PIGA	43.55 +/- 15.82	99.98 +/- 0.1	95.37 +/- 9.91	71.86 +/- 34.18
PLA2G4A	54.87 +/- 7.62	100.0 +/- 0.0	100.0 +/- 0.0	99.48 +/- 1.38
PLAU	52.12 +/- 8.57	100.0 +/- 0.0	99.9 +/- 0.67	97.94 +/- 4.83
PLG	54.24 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.01	99.48 +/- 1.66
PROC	53.03 +/- 8.11	100.0 +/- 0.0	100.0 +/- 0.0	98.65 +/- 5.25
PROS1	55.51 +/- 7.49	100.0 +/- 0.0	100.0 +/- 0.01	99.55 +/- 1.17
PTGS1	51.36 +/- 8.16	100.0 +/- 0.0	99.95 +/- 0.24	98.69 +/- 4.0
RASGRP2	48.74 +/- 7.78	100.0 +/- 0.0	99.82 +/- 0.81	96.15 +/- 9.08
RBM8A	53.81 +/- 7.96	100.0 +/- 0.0	100.0 +/- 0.0	99.12 +/- 2.21
RUNX1	52.94 +/- 7.33	99.99 +/- 0.05	99.8 +/- 0.75	98.13 +/- 3.64
SERPINC1	54.25 +/- 7.49	100.0 +/- 0.0	100.0 +/- 0.0	99.8 +/- 0.71
SERPIND1	55.14 +/- 7.87	100.0 +/- 0.0	100.0 +/- 0.0	99.33 +/- 3.18

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
SERPINE1	50.74 +/- 7.92	100.0 +/- 0.0	99.9 +/- 0.67	98.1 +/- 6.63
SERPINF2	50.2 +/- 9.3	100.0 +/- 0.0	99.89 +/- 0.36	96.02 +/- 7.39
SLFN14	54.62 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.01	99.25 +/- 2.18
SRC	50.77 +/- 8.08	99.99 +/- 0.04	99.7 +/- 1.12	97.34 +/- 6.53
STIM1	49.63 +/- 7.38	99.99 +/- 0.03	99.91 +/- 0.3	97.61 +/- 5.11
STXBP2	51.74 +/- 9.08	100.0 +/- 0.0	99.83 +/- 0.82	97.49 +/- 5.75
TBXA2R	53.93 +/- 9.17	100.0 +/- 0.0	99.98 +/- 0.11	97.94 +/- 6.72
TBXAS1	52.3 +/- 8.21	100.0 +/- 0.0	99.98 +/- 0.12	98.59 +/- 5.7
THBD	54.08 +/- 7.98	100.0 +/- 0.0	99.9 +/- 0.37	98.18 +/- 5.04
THPO	48.8 +/- 8.23	100.0 +/- 0.0	99.98 +/- 0.07	97.43 +/- 5.75
TPM4	52.79 +/- 7.43	100.0 +/- 0.0	99.99 +/- 0.05	98.61 +/- 4.53
TUBB1	55.0 +/- 7.3	100.0 +/- 0.0	100.0 +/- 0.0	99.66 +/- 0.95
VIPAS39	53.27 +/- 8.33	100.0 +/- 0.0	99.95 +/- 0.31	98.67 +/- 3.87
VKORC1	50.72 +/- 8.8	100.0 +/- 0.0	99.97 +/- 0.21	97.48 +/- 9.27
VPS33B	52.71 +/- 8.11	100.0 +/- 0.0	99.94 +/- 0.35	99.06 +/- 2.64
VWF	52.06 +/- 7.8	100.0 +/- 0.0	99.99 +/- 0.04	98.7 +/- 3.84
WAS	36.39 +/- 14.17	99.35 +/- 2.48	85.76 +/- 20.61	59.56 +/- 41.46

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