AALBORG UNIVERSITETSHOSPITAL
– i gode hænder

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Medfødt Iktyosis

Panelbeskrivelse

Navn: Medfødt Iktyosis **version:** 1.0 **Ibrugtagningsdato:** 30/05/2023

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 [%]
ABCA12	55.54 +/- 7.57	100.0 +/- 0.0	99.99 +/- 0.04	99.51 +/- 1.24
ABHD5	55.92 +/- 7.62	100.0 +/- 0.0	99.99 +/- 0.04	99.61 +/- 1.0
ALDH3A2	52.72 +/- 7.49	100.0 +/- 0.02	99.99 +/- 0.03	98.94 +/- 1.83
ALOX12B	49.1 +/- 8.41	100.0 +/- 0.0	99.9 +/- 0.52	97.52 +/- 6.59
ALOXE3	49.59 +/- 8.27	100.0 +/- 0.0	99.84 +/- 0.82	97.14 +/- 7.31
AP1B1	51.79 +/- 7.78	100.0 +/- 0.0	99.9 +/- 0.47	98.44 +/- 4.23
AP1S1	48.22 +/- 8.25	100.0 +/- 0.0	99.81 +/- 0.9	95.16 +/- 8.63
ARSL	41.36 +/- 15.54	99.91 +/- 0.44	92.84 +/- 11.83	66.48 +/- 38.44
ASPRV1	52.31 +/- 8.98	100.0 +/- 0.0	100.0 +/- 0.0	98.46 +/- 5.39
CARD14	52.79 +/- 7.84	100.0 +/- 0.0	99.94 +/- 0.37	98.84 +/- 4.38
CASP14	48.33 +/- 7.91	100.0 +/- 0.0	99.96 +/- 0.3	97.88 +/- 5.64
CDSN	50.84 +/- 7.97	99.98 +/- 0.04	99.92 +/- 0.24	98.42 +/- 4.23
CERS3	53.58 +/- 7.73	100.0 +/- 0.0	99.97 +/- 0.14	99.16 +/- 3.3
CLDN1	55.35 +/- 8.11	100.0 +/- 0.0	99.98 +/- 0.16	99.41 +/- 1.76
CSTA	54.25 +/- 8.6	100.0 +/- 0.0	100.0 +/- 0.0	99.63 +/- 1.24
CYP4F22	51.3 +/- 8.04	100.0 +/- 0.0	99.8 +/- 0.69	97.11 +/- 6.24
DCLRE1C	54.63 +/- 7.65	100.0 +/- 0.0	99.99 +/- 0.05	99.54 +/- 0.94
DOLK	47.02 +/- 8.47	100.0 +/- 0.0	99.57 +/- 2.64	95.74 +/- 10.02
EBP	40.8 +/- 15.34	99.9 +/- 0.48	94.07 +/- 12.54	68.6 +/- 36.38
ELOVL1	49.99 +/- 8.87	100.0 +/- 0.0	99.97 +/- 0.18	98.24 +/- 4.37
ELOVL4	54.92 +/- 7.56	100.0 +/- 0.0	99.82 +/- 1.01	98.68 +/- 2.86
ERCC2	49.53 +/- 8.41	99.98 +/- 0.04	99.8 +/- 0.64	96.83 +/- 6.71
ERCC3	52.37 +/- 7.58	100.0 +/- 0.0	100.0 +/- 0.01	99.13 +/- 2.26
FDPS	50.61 +/- 7.98	100.0 +/- 0.0	99.95 +/- 0.34	98.48 +/- 4.28
FLG	48.17 +/- 8.91	99.97 +/- 0.15	99.18 +/- 3.0	94.33 +/- 9.84
FLG2	52.76 +/- 8.0	100.0 +/- 0.0	99.97 +/- 0.17	98.55 +/- 2.95
GBA1	52.32 +/- 8.71	100.0 +/- 0.0	99.98 +/- 0.11	98.82 +/- 3.79
GTF2E2	52.38 +/- 7.5	100.0 +/- 0.0	99.99 +/- 0.07	98.43 +/- 3.13
GTF2H5	54.75 +/- 7.22	100.0 +/- 0.0	99.99 +/- 0.03	99.45 +/- 1.01
KDSR	54.89 +/- 7.31	100.0 +/- 0.0	99.92 +/- 0.36	99.22 +/- 1.71
KRT1	50.85 +/- 8.31	100.0 +/- 0.01	99.65 +/- 0.65	97.55 +/- 4.53

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
KRT10	48.86 +/- 7.47	99.96 +/- 0.17	99.84 +/- 0.87	96.22 +/- 5.69
KRT2	49.65 +/- 7.88	100.0 +/- 0.0	99.99 +/- 0.07	97.9 +/- 6.31
KRT83	54.71 +/- 8.32	99.98 +/- 0.04	99.98 +/- 0.04	99.06 +/- 3.27
LIPN	53.85 +/- 7.65	100.0 +/- 0.0	100.0 +/- 0.0	99.45 +/- 1.29
LORICRIN	42.03 +/- 6.91	100.0 +/- 0.0	98.86 +/- 3.05	86.22 +/- 15.66
LRP1	51.6 +/- 8.31	100.0 +/- 0.0	99.94 +/- 0.22	97.86 +/- 5.47
MBTPS2	43.43 +/- 16.02	99.94 +/- 0.32	94.48 +/- 9.81	71.49 +/- 33.85
MPDU1	46.06 +/- 8.62	100.0 +/- 0.0	99.73 +/- 1.27	93.79 +/- 10.9
MPLKIP	53.26 +/- 7.06	100.0 +/- 0.0	99.95 +/- 0.17	98.86 +/- 2.54
MVD	56.85 +/- 8.84	100.0 +/- 0.0	99.91 +/- 0.58	99.09 +/- 2.73
MVK	51.87 +/- 7.94	100.0 +/- 0.0	99.94 +/- 0.39	98.25 +/- 5.52
NIPAL4	50.21 +/- 8.14	100.0 +/- 0.0	99.99 +/- 0.09	97.91 +/- 5.68
NLRP1	50.81 +/- 8.39	100.0 +/- 0.0	99.93 +/- 0.49	98.19 +/- 5.4
NSDHL	43.15 +/- 16.35	99.85 +/- 0.66	94.42 +/- 10.41	70.97 +/- 35.21
PERP	54.86 +/- 7.55	99.99 +/- 0.03	99.94 +/- 0.33	99.01 +/- 2.92
PEX7	56.2 +/- 7.37	100.0 +/- 0.0	100.0 +/- 0.0	99.46 +/- 1.84
PHGDH	52.25 +/- 8.35	100.0 +/- 0.0	100.0 +/- 0.0	98.5 +/- 6.02
PHYH	55.21 +/- 7.7	99.96 +/- 0.05	99.95 +/- 0.05	99.45 +/- 1.74
PIGA	43.55 +/- 15.82	99.98 +/- 0.1	95.37 +/- 9.91	71.86 +/- 34.18
PIGL	53.89 +/- 7.6	100.0 +/- 0.0	100.0 +/- 0.0	99.23 +/- 3.17
PMVK	48.29 +/- 8.82	100.0 +/- 0.0	99.98 +/- 0.13	96.48 +/- 8.9
PNPLA1	50.91 +/- 8.45	100.0 +/- 0.0	99.92 +/- 0.26	98.35 +/- 3.34
POMP	54.71 +/- 7.69	100.0 +/- 0.0	100.0 +/- 0.0	99.39 +/- 1.71
PSAT1	55.63 +/- 7.93	100.0 +/- 0.0	99.99 +/- 0.04	99.44 +/- 1.36
PSPH	54.35 +/- 7.66	100.0 +/- 0.01	99.97 +/- 0.18	99.16 +/- 3.5
SDR9C7	53.22 +/- 8.71	100.0 +/- 0.0	100.0 +/- 0.0	99.1 +/- 3.58
SERPINB8	53.46 +/- 7.79	100.0 +/- 0.0	100.0 +/- 0.0	99.39 +/- 1.49
SLC17A9	52.47 +/- 8.43	100.0 +/- 0.0	99.81 +/- 0.53	96.88 +/- 5.9
SLC27A4	49.45 +/- 8.13	99.99 +/- 0.07	99.56 +/- 2.2	96.06 +/- 8.88
SLURP1	50.25 +/- 9.36	100.0 +/- 0.0	99.91 +/- 0.6	97.04 +/- 12.09
SNAP29	53.74 +/- 7.86	99.97 +/- 0.07	99.88 +/- 0.35	98.56 +/- 3.76
SPINK5	54.97 +/- 7.53	100.0 +/- 0.0	99.98 +/- 0.14	99.55 +/- 0.91
SSH1	53.08 +/- 7.79	100.0 +/- 0.0	99.9 +/- 0.33	98.84 +/- 2.58
ST14	52.86 +/- 8.44	100.0 +/- 0.0	99.85 +/- 0.7	97.95 +/- 6.64
STIM1	49.63 +/- 7.38	99.99 +/- 0.03	99.91 +/- 0.3	97.61 +/- 5.11
STS	41.78 +/- 15.38	99.87 +/- 0.53	92.86 +/- 12.46	68.71 +/- 35.58
SULT2B1	46.98 +/- 8.73	100.0 +/- 0.0	99.87 +/- 0.78	95.49 +/- 12.87
SUMF1	55.1 +/- 7.9	99.96 +/- 0.08	99.83 +/- 0.86	99.09 +/- 3.01
TARS1	55.66 +/- 7.52	100.0 +/- 0.0	100.0 +/- 0.0	99.5 +/- 1.53
TGM1	50.14 +/- 8.3	99.88 +/- 0.19	99.29 +/- 0.36	96.65 +/- 6.65
TRPM4	50.11 +/- 8.21	100.0 +/- 0.0	99.84 +/- 0.65	97.12 +/- 5.92
TRPV3	52.92 +/- 8.11	100.0 +/- 0.0	99.94 +/- 0.28	98.57 +/- 4.27
VPS33B	52.71 +/- 8.11	100.0 +/- 0.0	99.94 +/- 0.35	99.06 +/- 2.64