

Sjældne calcium og knoglesygdomme

Panelbeskrivelse

Navn: Sjældne calcium og knoglesygdomme

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 foretages in silico filtrering af data til relevante genregioner baseret på nedenstående tabel. Middelsekventeringsdybden er minimum 30X, og minimumsandelens af de kodede regioner der dækket minimum 10X er 90 %.

Genliste

Gener hvor middelsekventeringsdybden er < 30X er markeret med¹. Gener, hvor andelen af de kodede regioner der dækket minimum 10X er < 90 % er markeret med². Værdier er angivet med +/- standardafvigelse.

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
ACAN	49.74 +/- 7.63	97.93 +/- 1.61	95.82 +/- 2.03	91.87 +/- 5.34
AIRE	50.75 +/- 8.1	100.0 +/- 0.0	99.96 +/- 0.3	97.68 +/- 6.97
ALPL	50.32 +/- 8.38	100.0 +/- 0.0	99.81 +/- 0.62	97.84 +/- 4.49
AMER1	40.47 +/- 15.34	99.7 +/- 1.01	91.15 +/- 15.34	66.2 +/- 37.75
ANKH	53.85 +/- 7.9	99.94 +/- 0.18	99.66 +/- 0.41	98.23 +/- 2.48
ANO5	54.7 +/- 7.3	99.97 +/- 0.05	99.96 +/- 0.05	99.22 +/- 1.81
AP2S1	46.74 +/- 8.74	100.0 +/- 0.0	99.72 +/- 0.72	94.85 +/- 9.38
ATP6V0A2	53.58 +/- 7.51	100.0 +/- 0.0	99.99 +/- 0.07	99.26 +/- 1.69
B3GALT6	54.1 +/- 9.34	100.0 +/- 0.0	99.68 +/- 1.64	97.2 +/- 6.81
B3GAT3	50.47 +/- 8.04	100.0 +/- 0.0	99.96 +/- 0.24	98.5 +/- 3.88
B4GALT7	49.98 +/- 8.36	99.99 +/- 0.07	99.76 +/- 1.15	96.99 +/- 6.52
BMP1	51.31 +/- 8.21	100.0 +/- 0.0	99.89 +/- 0.69	97.85 +/- 5.68
CA2	55.23 +/- 8.05	100.0 +/- 0.0	99.94 +/- 0.4	99.07 +/- 2.18
CASR	51.65 +/- 7.73	100.0 +/- 0.0	99.7 +/- 0.41	97.83 +/- 3.14
CDC73	54.46 +/- 7.48	100.0 +/- 0.0	99.99 +/- 0.04	99.13 +/- 2.0
CDKN1A	52.14 +/- 8.44	100.0 +/- 0.0	99.97 +/- 0.18	98.45 +/- 3.44
CDKN1B	52.14 +/- 7.61	100.0 +/- 0.0	99.95 +/- 0.25	98.02 +/- 4.01
CDKN2B	55.08 +/- 7.63	100.0 +/- 0.0	100.0 +/- 0.01	99.5 +/- 1.63
CDKN2C	51.0 +/- 8.23	100.0 +/- 0.0	99.98 +/- 0.15	98.35 +/- 6.09
CLCN5	43.02 +/- 16.21	99.98 +/- 0.09	94.67 +/- 8.02	69.5 +/- 35.7
CLCN7	54.27 +/- 9.34	100.0 +/- 0.0	99.94 +/- 0.29	97.91 +/- 5.82
COL10A1	53.62 +/- 7.37	100.0 +/- 0.0	100.0 +/- 0.01	98.31 +/- 5.33
COL11A1	53.87 +/- 7.14	100.0 +/- 0.0	99.95 +/- 0.22	99.2 +/- 1.94
COL11A2	47.46 +/- 8.55	100.0 +/- 0.0	99.72 +/- 1.12	95.79 +/- 8.64
COL12A1	55.55 +/- 7.52	100.0 +/- 0.0	99.99 +/- 0.04	99.48 +/- 1.53
COL1A1	49.25 +/- 8.17	100.0 +/- 0.0	99.92 +/- 0.45	97.55 +/- 7.02
COL1A2	53.65 +/- 7.18	100.0 +/- 0.0	100.0 +/- 0.0	99.26 +/- 2.37
COL2A1	50.25 +/- 8.17	100.0 +/- 0.0	99.9 +/- 0.48	97.93 +/- 6.53
COL5A1	53.23 +/- 8.19	100.0 +/- 0.0	99.9 +/- 0.38	98.25 +/- 4.49
COPB2	55.74 +/- 7.61	100.0 +/- 0.0	100.0 +/- 0.0	99.68 +/- 1.01
CREB3L1	47.15 +/- 7.86	100.0 +/- 0.0	99.86 +/- 0.35	95.6 +/- 9.63

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>CREBBP</i>	49.39 +/- 7.58	99.77 +/- 0.53	98.19 +/- 2.38	92.76 +/- 5.98
<i>CRTAP</i>	54.11 +/- 7.73	100.0 +/- 0.0	99.96 +/- 0.21	99.04 +/- 2.59
<i>CTSK</i>	53.75 +/- 8.09	100.0 +/- 0.0	100.0 +/- 0.0	99.09 +/- 2.56
<i>CTSK</i>	54.71 +/- 7.36	100.0 +/- 0.0	99.99 +/- 0.04	99.32 +/- 3.28
<i>CYP26B1</i>	51.88 +/- 8.53	99.99 +/- 0.06	99.8 +/- 0.84	97.5 +/- 6.38
<i>CYP27B1</i>	52.54 +/- 8.13	100.0 +/- 0.0	99.92 +/- 0.4	98.8 +/- 4.02
<i>CYP2R1</i>	54.25 +/- 7.37	100.0 +/- 0.0	100.0 +/- 0.03	99.37 +/- 1.94
<i>CYP3A4</i>	54.19 +/- 8.14	100.0 +/- 0.0	99.97 +/- 0.21	99.21 +/- 2.58
<i>DLX3</i>	52.03 +/- 8.13	100.0 +/- 0.0	100.0 +/- 0.0	98.65 +/- 4.99
<i>DMP1</i>	54.29 +/- 8.03	99.99 +/- 0.03	99.97 +/- 0.07	99.36 +/- 1.8
<i>DSPP</i>	51.23 +/- 6.79	98.8 +/- 0.73	97.3 +/- 1.15	95.35 +/- 2.67
<i>ENPP1</i>	54.22 +/- 7.72	100.0 +/- 0.0	99.96 +/- 0.13	98.8 +/- 2.67
<i>ESR1</i>	53.51 +/- 7.57	100.0 +/- 0.0	99.98 +/- 0.09	99.05 +/- 2.92
<i>FAH</i>	51.24 +/- 8.32	100.0 +/- 0.0	99.76 +/- 0.84	97.67 +/- 6.81
<i>FAM20C</i>	51.07 +/- 8.57	100.0 +/- 0.0	99.22 +/- 2.25	92.96 +/- 10.25
<i>FERMT3</i>	51.79 +/- 8.56	100.0 +/- 0.0	99.84 +/- 0.73	97.08 +/- 6.16
<i>FGF23</i>	51.71 +/- 7.67	100.0 +/- 0.0	99.99 +/- 0.04	98.61 +/- 3.89
<i>FGFR1</i>	52.45 +/- 8.49	100.0 +/- 0.0	99.9 +/- 0.47	97.99 +/- 6.49
<i>FKBP10</i>	47.97 +/- 8.73	99.95 +/- 0.05	99.8 +/- 0.33	95.91 +/- 7.73
<i>GATA3</i>	52.12 +/- 7.99	100.0 +/- 0.0	99.91 +/- 0.47	97.78 +/- 4.33
<i>GCM2</i>	53.6 +/- 7.85	100.0 +/- 0.0	99.99 +/- 0.09	99.23 +/- 2.9
<i>GHR</i>	55.0 +/- 7.54	99.79 +/- 0.43	99.68 +/- 0.45	98.59 +/- 2.88
<i>GJA1</i>	53.43 +/- 7.32	100.0 +/- 0.0	99.99 +/- 0.03	99.14 +/- 1.51
<i>GNA11</i>	51.71 +/- 8.68	99.97 +/- 0.16	99.48 +/- 1.48	96.04 +/- 7.4
<i>GNAS</i>	47.03 +/- 7.06	99.83 +/- 0.62	94.82 +/- 6.95	83.43 +/- 8.94
<i>GNPTAB</i>	55.28 +/- 7.66	99.99 +/- 0.03	99.71 +/- 0.63	98.17 +/- 2.37
<i>GORAB</i>	55.01 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.39
<i>GSC</i>	51.21 +/- 8.98	100.0 +/- 0.0	99.98 +/- 0.15	97.74 +/- 5.19
<i>HPGD</i>	55.41 +/- 7.62	100.0 +/- 0.0	100.0 +/- 0.0	99.64 +/- 0.99
<i>IFITM5</i>	52.0 +/- 9.46	100.0 +/- 0.0	99.91 +/- 0.61	97.54 +/- 7.85
<i>KDELR2</i>	54.84 +/- 7.83	99.92 +/- 0.04	99.86 +/- 0.38	98.48 +/- 3.48
<i>LEMD3</i>	54.66 +/- 7.66	100.0 +/- 0.0	99.99 +/- 0.05	98.85 +/- 2.97
<i>LMNA</i>	49.81 +/- 8.27	100.0 +/- 0.0	99.96 +/- 0.16	97.29 +/- 6.96
<i>LRP4</i>	51.36 +/- 8.09	100.0 +/- 0.0	99.95 +/- 0.26	98.26 +/- 4.32
<i>LRP5</i>	52.35 +/- 8.26	99.98 +/- 0.07	99.55 +/- 1.19	96.63 +/- 5.5
<i>LRP6</i>	55.06 +/- 7.31	100.0 +/- 0.0	99.96 +/- 0.18	99.26 +/- 1.76
<i>LRRK1</i>	51.53 +/- 7.79	99.24 +/- 0.69	98.63 +/- 0.78	96.86 +/- 3.34
<i>MAFB</i>	53.89 +/- 8.28	100.0 +/- 0.0	99.99 +/- 0.07	98.51 +/- 5.73
<i>MATN3</i>	53.8 +/- 7.29	100.0 +/- 0.0	99.86 +/- 0.58	98.8 +/- 2.74
<i>MBTPS2</i>	43.43 +/- 16.02	99.94 +/- 0.32	94.48 +/- 9.81	71.49 +/- 33.85
<i>MEN1</i>	49.7 +/- 8.05	100.0 +/- 0.0	99.86 +/- 0.57	96.63 +/- 7.2
<i>MESD</i>	52.48 +/- 7.71	100.0 +/- 0.0	99.92 +/- 0.36	99.1 +/- 2.11
<i>MMP2</i>	51.21 +/- 8.12	100.0 +/- 0.0	100.0 +/- 0.03	98.32 +/- 4.36
<i>NBAS</i>	55.47 +/- 7.24	100.0 +/- 0.0	100.0 +/- 0.0	99.52 +/- 1.16
<i>NOTCH2</i>	53.62 +/- 7.84	100.0 +/- 0.0	99.99 +/- 0.04	99.02 +/- 2.39
<i>NUDT6</i>	55.43 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.0	99.72 +/- 0.95

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>OCRL</i>	40.92 +/- 15.61	99.87 +/- 0.59	91.55 +/- 13.83	66.68 +/- 38.47
<i>OFD1</i>	42.17 +/- 15.78	99.93 +/- 0.36	93.98 +/- 9.99	68.61 +/- 36.55
<i>OSTM1</i>	55.32 +/- 7.28	100.0 +/- 0.0	99.97 +/- 0.13	99.21 +/- 2.03
<i>P3H1</i>	52.34 +/- 8.36	100.0 +/- 0.0	99.98 +/- 0.15	98.37 +/- 6.06
<i>P4HB</i>	52.79 +/- 8.45	100.0 +/- 0.0	100.0 +/- 0.0	98.38 +/- 6.59
<i>PAX3</i>	53.43 +/- 7.53	100.0 +/- 0.0	99.98 +/- 0.16	98.96 +/- 2.34
<i>PHEX</i>	42.24 +/- 15.5	99.9 +/- 0.34	94.19 +/- 10.41	68.9 +/- 35.67
<i>PLEKHM1</i>	53.82 +/- 8.17	100.0 +/- 0.0	99.95 +/- 0.15	98.73 +/- 3.73
<i>PLOD1</i>	46.65 +/- 7.88	100.0 +/- 0.0	99.95 +/- 0.13	95.59 +/- 8.98
<i>PLOD2</i>	53.87 +/- 7.13	100.0 +/- 0.0	99.96 +/- 0.18	98.68 +/- 2.42
<i>PLS3</i>	43.27 +/- 16.09	99.97 +/- 0.14	94.79 +/- 10.07	70.34 +/- 35.09
<i>POC1A</i>	53.75 +/- 8.69	100.0 +/- 0.0	100.0 +/- 0.01	98.95 +/- 4.26
<i>PPIB</i>	50.0 +/- 8.67	100.0 +/- 0.0	99.82 +/- 0.89	97.3 +/- 8.01
<i>PTDSS1</i>	53.32 +/- 8.13	100.0 +/- 0.0	100.0 +/- 0.02	98.93 +/- 3.06
<i>PTH</i>	55.4 +/- 8.22	100.0 +/- 0.0	100.0 +/- 0.0	99.78 +/- 0.85
<i>PTH1R</i>	50.27 +/- 8.68	100.0 +/- 0.0	99.9 +/- 0.58	97.38 +/- 9.54
<i>PTPRQ</i>	56.04 +/- 7.29	100.0 +/- 0.0	100.0 +/- 0.0	99.72 +/- 0.81
<i>PYCR1</i>	53.24 +/- 9.16	100.0 +/- 0.0	99.92 +/- 0.32	98.62 +/- 3.25
<i>RASGRP2</i>	48.74 +/- 7.78	100.0 +/- 0.0	99.82 +/- 0.81	96.15 +/- 9.08
<i>RET</i>	52.09 +/- 8.29	100.0 +/- 0.0	99.98 +/- 0.12	98.51 +/- 3.77
<i>SEC24D</i>	54.33 +/- 7.53	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.52
<i>SERPINF1</i>	50.56 +/- 7.69	100.0 +/- 0.0	99.77 +/- 1.43	98.16 +/- 6.61
<i>SERPINH1</i>	54.12 +/- 8.51	100.0 +/- 0.0	99.89 +/- 0.66	98.21 +/- 6.19
<i>SFRP4</i>	55.24 +/- 8.01	100.0 +/- 0.0	99.98 +/- 0.15	99.42 +/- 1.37
<i>SGK3</i>	55.88 +/- 7.71	100.0 +/- 0.0	99.99 +/- 0.04	99.6 +/- 0.82
<i>SGMS2</i>	55.01 +/- 7.33	100.0 +/- 0.0	99.96 +/- 0.19	99.1 +/- 1.73
<i>SHOX</i>	51.15 +/- 7.42	99.72 +/- 0.46	98.11 +/- 1.86	93.63 +/- 6.83
<i>SLC34A1</i>	54.33 +/- 7.81	100.0 +/- 0.0	100.0 +/- 0.0	99.54 +/- 1.23
<i>SLC34A1</i>	50.01 +/- 8.87	100.0 +/- 0.0	99.92 +/- 0.29	97.59 +/- 6.36
<i>SLC34A3</i>	48.22 +/- 8.4	100.0 +/- 0.0	99.7 +/- 0.85	95.31 +/- 8.2
<i>SLCO2A1</i>	50.42 +/- 8.09	100.0 +/- 0.0	99.96 +/- 0.21	98.1 +/- 5.5
<i>SNX10</i>	55.5 +/- 7.68	100.0 +/- 0.0	99.92 +/- 0.3	98.88 +/- 2.33
<i>SOST</i>	52.76 +/- 7.84	100.0 +/- 0.0	99.94 +/- 0.25	98.49 +/- 4.74
<i>SOX3</i>	37.81 +/- 14.98	98.86 +/- 5.25	85.73 +/- 21.08	60.87 +/- 40.35
<i>SP7</i>	49.96 +/- 8.44	99.99 +/- 0.07	99.74 +/- 0.82	96.38 +/- 7.11
<i>SPARC</i>	54.03 +/- 7.77	100.0 +/- 0.0	99.99 +/- 0.07	99.3 +/- 1.78
<i>STX16</i>	54.08 +/- 7.8	100.0 +/- 0.0	99.95 +/- 0.31	99.02 +/- 2.85
<i>SUCO</i>	54.85 +/- 7.3	100.0 +/- 0.0	100.0 +/- 0.0	99.46 +/- 1.14
<i>TAPT1</i>	54.49 +/- 7.22	100.0 +/- 0.0	99.88 +/- 0.44	98.78 +/- 2.08
<i>TBCE</i>	55.0 +/- 8.14	100.0 +/- 0.0	100.0 +/- 0.0	99.46 +/- 1.54
<i>TBX1</i>	44.43 +/- 8.42	100.0 +/- 0.0	97.39 +/- 7.75	87.47 +/- 18.59
<i>TBXAS1</i>	52.3 +/- 8.21	100.0 +/- 0.0	99.98 +/- 0.12	98.59 +/- 5.7
<i>TCIRG1</i>	54.47 +/- 9.52	100.0 +/- 0.0	100.0 +/- 0.03	98.06 +/- 6.1
<i>TDP2</i>	54.25 +/- 7.56	100.0 +/- 0.0	99.98 +/- 0.12	99.44 +/- 1.86
<i>TENT5A</i>	54.14 +/- 7.42	100.0 +/- 0.0	99.98 +/- 0.08	99.33 +/- 1.49
<i>TGFB1</i>	45.88 +/- 7.75	99.99 +/- 0.05	98.76 +/- 3.85	92.3 +/- 11.8

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
TMCO1	53.44 +/- 7.62	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.16
TMEM38B	54.98 +/- 7.62	100.0 +/- 0.0	99.97 +/- 0.12	99.24 +/- 1.67
TNFRSF11A	52.91 +/- 7.65	100.0 +/- 0.0	99.96 +/- 0.2	98.72 +/- 2.89
TNFRSF11B	55.96 +/- 8.22	100.0 +/- 0.0	100.0 +/- 0.0	99.74 +/- 0.92
TNFSF11	54.5 +/- 7.34	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 0.97
TNXB	52.24 +/- 8.62	100.0 +/- 0.0	99.91 +/- 0.46	97.93 +/- 5.47
TREM2	50.36 +/- 9.01	100.0 +/- 0.0	100.0 +/- 0.0	98.36 +/- 8.32
TRMT10A	55.86 +/- 7.36	100.0 +/- 0.0	99.98 +/- 0.15	99.52 +/- 1.36
TRPV6	50.62 +/- 8.72	100.0 +/- 0.0	99.98 +/- 0.15	97.71 +/- 7.6
TYROBP	44.69 +/- 8.3	100.0 +/- 0.0	99.28 +/- 3.59	95.75 +/- 7.62
VDR	49.67 +/- 8.16	99.96 +/- 0.08	99.85 +/- 0.49	97.64 +/- 6.4
WNT1	51.93 +/- 9.06	100.0 +/- 0.0	99.83 +/- 1.09	97.89 +/- 5.84
ZMPSTE24	55.06 +/- 6.95	100.0 +/- 0.0	100.0 +/- 0.0	99.62 +/- 1.17

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