

Arvelige kolestatiske og fibrotiske leversygdomme basis panel

Panelbeskrivelse

Navn: Arvelige kolestatiske og fibrotiske leversygdomme basis panel

version: 1.0

Ibrugtagningsdato: 24/05-2023

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 [%]
<i>ABCB11</i>	52.87 +/- 7.46	99.97 +/- 0.05	99.94 +/- 0.14	99.21 +/- 1.76
<i>ABCB4</i>	54.98 +/- 7.55	100.0 +/- 0.0	100.0 +/- 0.03	99.5 +/- 1.41
<i>ABCC2</i>	53.16 +/- 7.78	100.0 +/- 0.0	100.0 +/- 0.0	99.36 +/- 2.0
<i>ADK</i>	56.47 +/- 7.12	100.0 +/- 0.0	100.0 +/- 0.0	99.4 +/- 1.66
<i>AKR1D1</i>	56.01 +/- 7.73	100.0 +/- 0.0	99.98 +/- 0.12	99.79 +/- 0.88
<i>ALDOB</i>	54.31 +/- 7.26	100.0 +/- 0.0	100.0 +/- 0.0	99.57 +/- 0.98
<i>ALG8</i>	55.41 +/- 7.63	100.0 +/- 0.0	99.95 +/- 0.31	99.55 +/- 1.3
<i>AMACR</i>	54.69 +/- 7.36	100.0 +/- 0.0	99.91 +/- 0.38	99.09 +/- 1.99
<i>ATP7B</i>	53.2 +/- 7.43	100.0 +/- 0.0	100.0 +/- 0.03	99.16 +/- 2.06
<i>ATP8B1</i>	53.7 +/- 7.13	100.0 +/- 0.0	99.98 +/- 0.06	99.03 +/- 2.58
<i>BAAT</i>	54.47 +/- 7.74	100.0 +/- 0.01	100.0 +/- 0.01	99.32 +/- 2.4
<i>BCS1L</i>	52.11 +/- 8.27	100.0 +/- 0.0	100.0 +/- 0.0	98.85 +/- 5.02
<i>CC2D2A</i>	54.8 +/- 7.76	100.0 +/- 0.0	99.99 +/- 0.09	99.36 +/- 2.46
<i>CFTR</i>	54.95 +/- 7.28	100.0 +/- 0.0	100.0 +/- 0.0	99.56 +/- 1.12
<i>CLDN1</i>	55.35 +/- 8.11	100.0 +/- 0.0	99.98 +/- 0.16	99.41 +/- 1.76
<i>COG7</i>	53.44 +/- 7.49	100.0 +/- 0.0	100.0 +/- 0.0	99.33 +/- 1.98
<i>CYP27A1</i>	51.96 +/- 8.62	100.0 +/- 0.0	99.86 +/- 0.92	98.15 +/- 6.72
<i>CYP7A1</i>	55.8 +/- 7.54	100.0 +/- 0.0	100.0 +/- 0.03	99.53 +/- 1.11
<i>CYP7B1</i>	54.34 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.37 +/- 1.84
<i>DCDC2</i>	55.85 +/- 7.47	100.0 +/- 0.0	100.0 +/- 0.01	99.64 +/- 0.95
<i>DGUOK</i>	52.26 +/- 7.35	100.0 +/- 0.0	100.0 +/- 0.0	99.43 +/- 1.77
<i>DNAJB11</i>	53.15 +/- 7.3	100.0 +/- 0.0	99.98 +/- 0.1	98.11 +/- 3.26
<i>FAH</i>	51.24 +/- 8.32	100.0 +/- 0.0	99.76 +/- 0.84	97.67 +/- 6.81
<i>GALE</i>	50.08 +/- 7.85	100.0 +/- 0.0	99.85 +/- 1.03	97.84 +/- 5.9
<i>GALK1</i>	52.67 +/- 9.14	100.0 +/- 0.0	99.91 +/- 0.39	98.3 +/- 5.26
<i>GALM</i>	54.92 +/- 11.56	100.0 +/- 0.0	100.0 +/- 0.0	99.27 +/- 2.39
<i>GALT</i>	51.51 +/- 8.19	100.0 +/- 0.0	100.0 +/- 0.0	99.05 +/- 2.48
<i>GANAB</i>	50.54 +/- 7.79	100.0 +/- 0.0	100.0 +/- 0.0	98.47 +/- 3.74
<i>GBA1</i>	52.32 +/- 8.71	100.0 +/- 0.0	99.98 +/- 0.11	98.82 +/- 3.79
<i>GBE1</i>	55.38 +/- 7.26	100.0 +/- 0.0	100.0 +/- 0.0	99.5 +/- 1.34
<i>HADHA</i>	53.12 +/- 7.75	100.0 +/- 0.0	99.91 +/- 0.35	98.23 +/- 5.5

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>HNF1B</i>	49.89 +/- 7.94	100.0 +/- 0.0	99.66 +/- 1.6	96.98 +/- 6.57
<i>HSD3B7</i>	53.17 +/- 8.61	100.0 +/- 0.0	100.0 +/- 0.0	98.55 +/- 4.68
<i>INVS</i>	54.25 +/- 7.9	100.0 +/- 0.0	100.0 +/- 0.01	99.41 +/- 1.58
<i>JAG1</i>	54.22 +/- 7.67	100.0 +/- 0.0	99.94 +/- 0.37	98.84 +/- 2.65
<i>KIF12</i>	49.0 +/- 8.09	100.0 +/- 0.0	99.87 +/- 0.57	97.52 +/- 7.48
<i>LIPA</i>	55.33 +/- 7.73	100.0 +/- 0.0	99.89 +/- 0.75	98.98 +/- 5.99
<i>LRP5</i>	52.35 +/- 8.26	99.98 +/- 0.07	99.55 +/- 1.19	96.63 +/- 5.5
<i>MPI</i>	54.13 +/- 8.33	100.0 +/- 0.0	99.97 +/- 0.19	98.98 +/- 4.16
<i>MPV17</i>	53.45 +/- 8.53	100.0 +/- 0.0	99.98 +/- 0.12	98.83 +/- 4.25
<i>MVK</i>	51.87 +/- 7.94	100.0 +/- 0.0	99.94 +/- 0.39	98.25 +/- 5.52
<i>MYO5B</i>	55.97 +/- 7.41	100.0 +/- 0.0	99.93 +/- 0.22	98.6 +/- 2.82
<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>NPC1</i>	53.64 +/- 7.56	100.0 +/- 0.0	99.95 +/- 0.27	98.96 +/- 3.09
<i>NPC2</i>	51.2 +/- 8.72	100.0 +/- 0.0	99.95 +/- 0.36	98.58 +/- 4.1
<i>NR1H4</i>	54.18 +/- 7.39	100.0 +/- 0.0	100.0 +/- 0.01	99.39 +/- 1.58
<i>PEX1</i>	55.37 +/- 6.89	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.1
<i>PEX10</i>	56.38 +/- 8.62	100.0 +/- 0.0	99.93 +/- 0.45	99.24 +/- 2.69
<i>PEX12</i>	52.35 +/- 7.99	99.94 +/- 0.09	99.91 +/- 0.19	99.26 +/- 1.91
<i>PEX13</i>	54.34 +/- 7.3	100.0 +/- 0.0	99.96 +/- 0.28	99.26 +/- 2.16
<i>PEX14</i>	51.87 +/- 8.66	100.0 +/- 0.0	99.92 +/- 0.55	98.01 +/- 6.89
<i>PEX16</i>	51.67 +/- 9.09	100.0 +/- 0.0	99.96 +/- 0.3	98.06 +/- 4.43
<i>PEX19</i>	53.44 +/- 8.25	100.0 +/- 0.0	100.0 +/- 0.0	99.58 +/- 1.02
<i>PEX2</i>	54.84 +/- 7.73	100.0 +/- 0.0	100.0 +/- 0.0	99.74 +/- 0.88
<i>PEX26</i>	53.39 +/- 7.8	100.0 +/- 0.0	99.92 +/- 0.18	98.78 +/- 3.09
<i>PEX3</i>	55.63 +/- 7.13	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 1.19
<i>PEX6</i>	49.64 +/- 7.84	99.96 +/- 0.05	99.82 +/- 0.41	96.66 +/- 7.29
<i>PKD1</i>	56.64 +/- 9.29	100.0 +/- 0.0	99.66 +/- 0.8	97.51 +/- 4.02
<i>PKD2</i>	53.54 +/- 7.27	100.0 +/- 0.0	99.72 +/- 0.98	97.86 +/- 4.21
<i>PKHD1</i>	54.54 +/- 7.54	100.0 +/- 0.0	99.99 +/- 0.06	99.52 +/- 1.3
<i>POLG</i>	52.85 +/- 8.09	100.0 +/- 0.0	99.94 +/- 0.37	98.88 +/- 3.87
<i>PRKCSH</i>	49.54 +/- 8.71	99.99 +/- 0.03	99.8 +/- 1.09	97.39 +/- 7.65
<i>RINT1</i>	55.34 +/- 7.25	100.0 +/- 0.0	100.0 +/- 0.01	99.5 +/- 1.54
<i>SEC63</i>	53.98 +/- 7.17	99.88 +/- 0.08	99.84 +/- 0.08	98.78 +/- 1.99
<i>SERPINA1</i>	50.83 +/- 8.36	100.0 +/- 0.0	99.89 +/- 0.33	96.92 +/- 6.26
<i>SLC25A13</i>	55.41 +/- 7.71	100.0 +/- 0.0	100.0 +/- 0.0	99.31 +/- 2.0
<i>SMPD1</i>	51.77 +/- 8.44	99.99 +/- 0.05	99.6 +/- 2.02	97.14 +/- 8.99
<i>TALDO1</i>	51.29 +/- 9.24	100.0 +/- 0.0	99.73 +/- 0.81	95.51 +/- 7.1
<i>TJP2</i>	54.5 +/- 7.92	100.0 +/- 0.0	99.94 +/- 0.2	98.84 +/- 1.72
<i>TRMU</i>	53.87 +/- 8.83	100.0 +/- 0.0	99.98 +/- 0.1	98.75 +/- 3.81
<i>UGT1A1</i>	55.94 +/- 7.57	100.0 +/- 0.0	100.0 +/- 0.0	99.71 +/- 0.89
<i>UNC45A</i>	51.0 +/- 8.44	100.0 +/- 0.0	99.94 +/- 0.39	97.96 +/- 6.43
<i>USP53</i>	54.64 +/- 7.32	100.0 +/- 0.0	99.99 +/- 0.04	99.41 +/- 1.08
<i>VIPAS39</i>	53.27 +/- 8.33	100.0 +/- 0.0	99.95 +/- 0.31	98.67 +/- 3.87
<i>VPS33B</i>	52.71 +/- 8.11	100.0 +/- 0.0	99.94 +/- 0.35	99.06 +/- 2.64
<i>YARS1</i>	50.56 +/- 8.06	100.0 +/- 0.0	99.99 +/- 0.05	98.46 +/- 5.15

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
ZFYVE19	52.46 +/- 8.77	100.0 +/- 0.0	100.0 +/- 0.0	98.81 +/- 5.0

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