

## Torakal aortasygdom udvidet panel

## Panelbeskrivelse

Navn: Torakal aortasygdom udvidet panel

version: 1.2

Ibrugtagningsdato: 22/05-2022

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

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>ABCC6</i>	51.66 +/- 7.95	100.0 +/- 0.0	99.94 +/- 0.42	98.58 +/- 5.09
<i>ABL1</i>	53.1 +/- 7.95	100.0 +/- 0.0	99.88 +/- 0.4	98.07 +/- 4.65
<i>ACTA2</i>	52.83 +/- 8.32	100.0 +/- 0.0	100.0 +/- 0.0	99.41 +/- 2.94
<i>ADAMTS2</i>	53.33 +/- 8.29	100.0 +/- 0.0	99.78 +/- 0.96	97.91 +/- 4.83
<i>ADAMTSL4</i>	47.77 +/- 8.51	100.0 +/- 0.0	99.82 +/- 0.5	95.92 +/- 7.96
<i>ALDH18A1</i>	54.06 +/- 7.76	100.0 +/- 0.0	99.96 +/- 0.28	99.09 +/- 2.87
<i>ARIH1</i>	53.63 +/- 7.31	100.0 +/- 0.0	99.99 +/- 0.04	99.28 +/- 1.48
<i>ATP6V0A2</i>	53.58 +/- 7.51	100.0 +/- 0.0	99.99 +/- 0.07	99.26 +/- 1.69
<i>ATP7A</i>	43.34 +/- 15.44	99.96 +/- 0.18	95.8 +/- 8.34	72.24 +/- 33.0
<i>B3GAT3</i>	50.47 +/- 8.04	100.0 +/- 0.0	99.96 +/- 0.24	98.5 +/- 3.88
<i>B4GALT7</i>	49.98 +/- 8.36	99.99 +/- 0.07	99.76 +/- 1.15	96.99 +/- 6.52
<i>BGN</i>	37.87 +/- 14.99	99.68 +/- 0.84	88.47 +/- 18.41	60.18 +/- 42.58
<i>C1R</i>	51.67 +/- 8.43	100.0 +/- 0.0	99.96 +/- 0.25	98.28 +/- 7.35
<i>C1S</i>	53.68 +/- 7.81	100.0 +/- 0.0	100.0 +/- 0.0	99.51 +/- 2.08
<i>CBS</i>	52.88 +/- 8.49	100.0 +/- 0.0	99.8 +/- 0.65	98.07 +/- 4.61
<i>CHST14</i>	46.39 +/- 8.35	99.99 +/- 0.04	98.75 +/- 3.5	91.69 +/- 12.61
<i>COL11A2</i>	47.46 +/- 8.55	100.0 +/- 0.0	99.72 +/- 1.12	95.79 +/- 8.64
<i>COL12A1</i>	55.55 +/- 7.52	100.0 +/- 0.0	99.99 +/- 0.04	99.48 +/- 1.53
<i>COL1A1</i>	49.25 +/- 8.17	100.0 +/- 0.0	99.92 +/- 0.45	97.55 +/- 7.02
<i>COL1A2</i>	53.65 +/- 7.18	100.0 +/- 0.0	100.0 +/- 0.0	99.26 +/- 2.37
<i>COL2A1</i>	50.25 +/- 8.17	100.0 +/- 0.0	99.9 +/- 0.48	97.93 +/- 6.53
<i>COL3A1</i>	53.68 +/- 7.12	100.0 +/- 0.0	100.0 +/- 0.01	99.46 +/- 1.5
<i>COL4A1</i>	53.48 +/- 7.53	100.0 +/- 0.0	99.99 +/- 0.04	98.83 +/- 3.9
<i>COL4A5</i>	41.76 +/- 15.48	99.93 +/- 0.33	93.31 +/- 10.33	67.99 +/- 36.58
<i>COL5A1</i>	53.23 +/- 8.19	100.0 +/- 0.0	99.9 +/- 0.38	98.25 +/- 4.49
<i>COL5A2</i>	55.38 +/- 7.43	100.0 +/- 0.0	99.99 +/- 0.07	99.5 +/- 1.49
<i>COL6A1</i>	55.94 +/- 9.84	99.97 +/- 0.11	99.21 +/- 0.98	96.67 +/- 3.93
<i>COL6A2</i>	54.75 +/- 9.39	100.0 +/- 0.0	99.86 +/- 0.58	98.08 +/- 5.66
<i>COL6A3</i>	53.29 +/- 7.65	100.0 +/- 0.0	99.97 +/- 0.14	99.23 +/- 2.92
<i>DSE</i>	54.29 +/- 7.4	100.0 +/- 0.0	99.99 +/- 0.05	99.31 +/- 1.92
<i>EFEMP2</i>	50.77 +/- 8.1	100.0 +/- 0.01	99.87 +/- 0.6	97.71 +/- 7.99

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>ELN</i>	49.45 +/- 8.61	100.0 +/- 0.0	99.96 +/- 0.19	96.48 +/- 7.88
<i>FBLN5</i>	50.92 +/- 8.2	100.0 +/- 0.0	99.99 +/- 0.09	97.89 +/- 6.0
<i>FBN1</i>	55.19 +/- 7.67	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.72
<i>FBN2</i>	55.2 +/- 7.45	100.0 +/- 0.0	100.0 +/- 0.0	99.26 +/- 1.87
<i>FKBP14</i>	55.69 +/- 7.48	100.0 +/- 0.01	99.99 +/- 0.05	99.64 +/- 1.0
<i>FLNA</i>	42.42 +/- 16.9	99.82 +/- 0.82	92.1 +/- 15.58	67.32 +/- 37.82
<i>FOXE3</i>	46.47 +/- 8.23	100.0 +/- 0.0	99.3 +/- 2.22	93.29 +/- 13.76
<i>GATA5</i>	52.14 +/- 8.67	100.0 +/- 0.0	99.83 +/- 0.73	97.21 +/- 6.41
<i>HCN4</i>	49.54 +/- 8.01	100.0 +/- 0.0	99.82 +/- 0.92	96.09 +/- 8.54
<i>IPO8</i>	54.84 +/- 7.42	100.0 +/- 0.0	99.97 +/- 0.18	99.33 +/- 1.49
<i>LOX</i>	54.96 +/- 7.59	100.0 +/- 0.0	99.84 +/- 1.01	98.89 +/- 2.86
<i>LTBP3</i>	53.39 +/- 8.11	100.0 +/- 0.0	100.0 +/- 0.02	98.78 +/- 4.42
<i>LTBP3</i>	50.54 +/- 8.79	100.0 +/- 0.0	99.32 +/- 1.89	93.6 +/- 9.28
<i>MAT2A</i>	54.96 +/- 7.75	100.0 +/- 0.0	100.0 +/- 0.0	99.28 +/- 1.9
<i>MED12</i>	38.78 +/- 15.09	99.63 +/- 2.0	88.98 +/- 17.17	63.13 +/- 40.43
<i>MFAP5</i>	54.06 +/- 8.0	100.0 +/- 0.0	100.0 +/- 0.0	99.37 +/- 1.97
<i>MYH11</i>	51.46 +/- 7.81	100.0 +/- 0.0	99.96 +/- 0.12	98.42 +/- 4.1
<i>MYLK</i>	52.05 +/- 7.77	100.0 +/- 0.0	99.94 +/- 0.15	98.25 +/- 4.44
<i>NOTCH1</i>	56.37 +/- 8.96	100.0 +/- 0.0	99.89 +/- 0.29	98.31 +/- 4.08
<i>PIEZO2</i>	53.53 +/- 7.37	100.0 +/- 0.0	99.94 +/- 0.29	98.81 +/- 2.76
<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>PLOD1</i>	46.65 +/- 7.88	100.0 +/- 0.0	99.95 +/- 0.13	95.59 +/- 8.98
<i>PLOD3</i>	47.86 +/- 8.16	100.0 +/- 0.0	99.92 +/- 0.37	96.05 +/- 9.0
<i>PRDM5</i>	55.39 +/- 7.34	100.0 +/- 0.01	99.93 +/- 0.27	99.3 +/- 1.91
<i>PRKG1</i>	54.6 +/- 7.21	100.0 +/- 0.0	100.0 +/- 0.0	99.62 +/- 0.76
<i>SKI</i>	47.35 +/- 7.68	98.77 +/- 1.52	94.23 +/- 3.27	86.36 +/- 7.49
<i>SLC2A10</i>	53.42 +/- 8.61	100.0 +/- 0.0	99.97 +/- 0.13	98.91 +/- 3.98
<i>SLC39A13</i>	51.89 +/- 8.65	100.0 +/- 0.0	99.94 +/- 0.24	98.07 +/- 4.65
<i>SMAD2</i>	54.49 +/- 7.16	100.0 +/- 0.02	99.91 +/- 0.21	99.0 +/- 1.55
<i>SMAD3</i>	51.58 +/- 8.01	100.0 +/- 0.0	99.62 +/- 0.93	96.5 +/- 4.34
<i>SMAD4</i>	54.51 +/- 7.48	99.99 +/- 0.03	99.8 +/- 0.66	98.74 +/- 2.31
<i>SMAD6</i>	51.73 +/- 8.53	100.0 +/- 0.0	99.79 +/- 0.88	96.29 +/- 8.82
<i>TGFB2</i>	52.63 +/- 7.04	100.0 +/- 0.0	99.96 +/- 0.17	98.75 +/- 2.65
<i>TGFB3</i>	51.25 +/- 8.18	100.0 +/- 0.0	99.99 +/- 0.05	98.79 +/- 4.24
<i>TGFBR1</i>	54.82 +/- 7.73	100.0 +/- 0.0	99.94 +/- 0.18	98.87 +/- 1.78
<i>TGFBR2</i>	53.75 +/- 7.48	100.0 +/- 0.0	100.0 +/- 0.03	99.28 +/- 2.33
<i>THSD4</i>	52.3 +/- 7.64	100.0 +/- 0.0	99.98 +/- 0.09	98.53 +/- 4.12
<i>TNXB</i>	52.24 +/- 8.62	100.0 +/- 0.0	99.91 +/- 0.46	97.93 +/- 5.47
<i>ZDHHC9</i>	39.93 +/- 15.04	99.67 +/- 1.37	90.45 +/- 15.61	65.82 +/- 38.13
<i>ZNF469</i>	52.25 +/- 8.99	100.0 +/- 0.0	99.86 +/- 0.54	97.38 +/- 7.44

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