

## Arvelige trombocytdefekter

## Panelbeskrivelse

Navn: Arvelige trombocytdefekter

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 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>ABCC4</i>	54.72 +/- 7.84	100.0 +/- 0.0	100.0 +/- 0.02	99.42 +/- 1.78
<i>ABCG5</i>	52.38 +/- 7.41	100.0 +/- 0.0	99.96 +/- 0.21	98.52 +/- 4.73
<i>ABCG8</i>	53.42 +/- 8.21	99.99 +/- 0.03	99.94 +/- 0.33	98.68 +/- 5.02
<i>ACTB</i>	52.7 +/- 9.38	100.0 +/- 0.0	99.97 +/- 0.13	98.35 +/- 4.52
<i>ACTN1</i>	51.27 +/- 8.06	100.0 +/- 0.0	99.74 +/- 1.39	97.31 +/- 6.88
<i>ADAMTS13</i>	50.65 +/- 8.67	100.0 +/- 0.0	99.78 +/- 0.98	96.81 +/- 6.59
<i>ANKRD26</i>	54.36 +/- 7.35	100.0 +/- 0.0	99.99 +/- 0.09	99.36 +/- 1.42
<i>ANO6</i>	54.35 +/- 7.16	100.0 +/- 0.0	99.98 +/- 0.11	99.37 +/- 1.33
<i>AP3B1</i>	55.4 +/- 7.36	100.0 +/- 0.0	100.0 +/- 0.03	99.47 +/- 1.56
<i>AP3D1</i>	54.8 +/- 8.84	100.0 +/- 0.0	99.96 +/- 0.17	98.2 +/- 6.19
<i>ARPC1B</i>	52.45 +/- 8.75	100.0 +/- 0.0	99.86 +/- 0.58	97.79 +/- 8.11
<i>BLOC1S3</i>	48.86 +/- 8.08	100.0 +/- 0.0	99.93 +/- 0.4	97.44 +/- 6.09
<i>BLOC1S6</i>	56.96 +/- 7.12	100.0 +/- 0.0	100.0 +/- 0.0	99.83 +/- 0.43
<i>CDC42</i>	54.71 +/- 7.9	99.89 +/- 0.09	99.77 +/- 0.22	98.97 +/- 2.19
<i>CYCS</i>	56.18 +/- 7.18	100.0 +/- 0.0	99.99 +/- 0.04	99.81 +/- 0.53
<i>DIAPH1</i>	51.35 +/- 8.06	99.96 +/- 0.19	99.39 +/- 1.31	96.37 +/- 4.26
<i>DTNBP1</i>	53.31 +/- 8.19	100.0 +/- 0.0	100.0 +/- 0.0	97.88 +/- 5.83
<i>ETV6</i>	49.91 +/- 7.75	100.0 +/- 0.0	99.85 +/- 0.66	96.75 +/- 5.0
<i>F10</i>	52.31 +/- 8.31	100.0 +/- 0.0	99.82 +/- 1.22	98.51 +/- 5.12
<i>F11</i>	54.29 +/- 7.59	100.0 +/- 0.0	100.0 +/- 0.0	99.37 +/- 1.46
<i>F12</i>	53.05 +/- 8.93	100.0 +/- 0.0	99.92 +/- 0.35	97.86 +/- 8.03
<i>F13A1</i>	54.58 +/- 7.77	100.0 +/- 0.0	100.0 +/- 0.01	99.46 +/- 2.44
<i>F13B</i>	56.32 +/- 7.13	100.0 +/- 0.0	100.0 +/- 0.0	99.88 +/- 0.44
<i>F2</i>	51.5 +/- 8.06	100.0 +/- 0.0	99.99 +/- 0.06	98.42 +/- 6.71
<i>F5</i>	53.31 +/- 7.6	100.0 +/- 0.0	99.97 +/- 0.22	98.82 +/- 2.72
<i>F7</i>	52.39 +/- 8.42	100.0 +/- 0.0	99.97 +/- 0.15	98.71 +/- 3.41
<i>F8</i>	41.99 +/- 15.81	99.94 +/- 0.37	94.09 +/- 10.17	68.25 +/- 36.91
<i>F9</i>	41.38 +/- 15.1	99.96 +/- 0.17	93.5 +/- 12.07	68.78 +/- 35.37
<i>FERMT3</i>	51.79 +/- 8.56	100.0 +/- 0.0	99.84 +/- 0.73	97.08 +/- 6.16
<i>FGA</i>	52.05 +/- 8.18	100.0 +/- 0.0	99.99 +/- 0.06	98.86 +/- 3.04
<i>FGB</i>	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 [%]
<i>FGG</i>	55.31 +/- 8.18	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 1.09
<i>FLI1</i>	52.82 +/- 7.92	98.94 +/- 3.36	98.47 +/- 3.5	96.09 +/- 5.32
<i>FLNA</i>	42.42 +/- 16.9	99.82 +/- 0.82	92.1 +/- 15.58	67.32 +/- 37.82
<i>FYB1</i>	53.76 +/- 7.27	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 1.28
<i>GATA1</i>	37.58 +/- 14.64	99.85 +/- 0.91	88.03 +/- 18.64	61.8 +/- 42.41
<i>GFI1B</i>	52.72 +/- 8.69	100.0 +/- 0.0	99.96 +/- 0.24	98.47 +/- 4.34
<i>GGCX</i>	53.45 +/- 7.74	100.0 +/- 0.0	100.0 +/- 0.03	99.36 +/- 1.89
<i>GNE</i>	54.84 +/- 7.57	99.98 +/- 0.04	99.98 +/- 0.04	99.41 +/- 1.23
<i>GP1BA</i>	48.42 +/- 8.44	99.87 +/- 0.27	98.18 +/- 1.5	93.59 +/- 6.77
<i>GP1BB</i>	54.66 +/- 10.37	100.0 +/- 0.0	99.86 +/- 0.94	97.17 +/- 11.47
<i>GP6</i>	52.78 +/- 8.88	100.0 +/- 0.0	100.0 +/- 0.0	98.71 +/- 4.14
<i>GP9</i>	52.57 +/- 9.02	100.0 +/- 0.0	100.0 +/- 0.0	98.71 +/- 4.22
<i>HOXA11</i>	52.28 +/- 8.64	100.0 +/- 0.0	99.98 +/- 0.15	98.37 +/- 5.12
<i>HPS1</i>	52.48 +/- 8.51	100.0 +/- 0.0	99.95 +/- 0.34	98.62 +/- 4.65
<i>HPS3</i>	54.51 +/- 7.71	100.0 +/- 0.0	99.95 +/- 0.2	98.99 +/- 2.03
<i>HPS4</i>	53.55 +/- 7.82	99.96 +/- 0.06	99.87 +/- 0.12	98.79 +/- 3.14
<i>HPS5</i>	55.41 +/- 7.19	100.0 +/- 0.0	100.0 +/- 0.0	99.69 +/- 0.82
<i>HPS6</i>	50.72 +/- 8.59	100.0 +/- 0.0	99.94 +/- 0.4	97.58 +/- 7.48
<i>HRG</i>	52.52 +/- 8.63	100.0 +/- 0.0	99.88 +/- 0.64	98.1 +/- 5.7
<i>IKZF5</i>	54.11 +/- 7.15	100.0 +/- 0.0	99.99 +/- 0.07	98.99 +/- 2.82
<i>ITGA2B</i>	49.48 +/- 7.95	100.0 +/- 0.0	99.95 +/- 0.36	97.84 +/- 6.39
<i>ITGB3</i>	52.94 +/- 7.61	100.0 +/- 0.0	99.92 +/- 0.4	98.76 +/- 4.03
<i>KDSR</i>	54.89 +/- 7.31	100.0 +/- 0.0	99.92 +/- 0.36	99.22 +/- 1.71
<i>KNG1</i>	54.58 +/- 7.48	100.0 +/- 0.0	99.98 +/- 0.1	99.47 +/- 1.48
<i>LMAN1</i>	55.98 +/- 7.58	100.0 +/- 0.0	99.99 +/- 0.09	99.16 +/- 1.86
<i>LYST</i>	55.53 +/- 7.3	100.0 +/- 0.0	100.0 +/- 0.0	99.59 +/- 1.01
<i>MCFD2</i>	53.56 +/- 7.43	99.99 +/- 0.06	99.89 +/- 0.35	99.15 +/- 2.69
<i>MECOM</i>	54.63 +/- 7.54	100.0 +/- 0.0	99.94 +/- 0.26	99.11 +/- 1.61
<i>MPIG6B</i>	51.3 +/- 8.07	100.0 +/- 0.0	99.98 +/- 0.16	98.54 +/- 3.84
<i>MPL</i>	52.02 +/- 8.45	100.0 +/- 0.0	99.99 +/- 0.04	98.5 +/- 3.75
<i>MYH9</i>	52.51 +/- 7.98	100.0 +/- 0.0	99.98 +/- 0.14	98.64 +/- 4.25
<i>NBEA</i>	55.43 +/- 7.51	100.0 +/- 0.03	99.83 +/- 0.56	98.67 +/- 2.14
<i>NBEAL2</i>	51.81 +/- 8.79	100.0 +/- 0.0	99.94 +/- 0.36	97.88 +/- 6.27
<i>P2RY12</i>	55.28 +/- 7.88	100.0 +/- 0.0	99.98 +/- 0.07	99.16 +/- 2.4
<i>PIGA</i>	43.55 +/- 15.82	99.98 +/- 0.1	95.37 +/- 9.91	71.86 +/- 34.18
<i>PLA2G4A</i>	54.87 +/- 7.62	100.0 +/- 0.0	100.0 +/- 0.0	99.48 +/- 1.38
<i>PLAU</i>	52.12 +/- 8.57	100.0 +/- 0.0	99.9 +/- 0.67	97.94 +/- 4.83
<i>PLG</i>	54.24 +/- 7.4	100.0 +/- 0.0	100.0 +/- 0.01	99.48 +/- 1.66
<i>PROC</i>	53.03 +/- 8.11	100.0 +/- 0.0	100.0 +/- 0.0	98.65 +/- 5.25
<i>PROS1</i>	55.51 +/- 7.49	100.0 +/- 0.0	100.0 +/- 0.01	99.55 +/- 1.17
<i>PTGS1</i>	51.36 +/- 8.16	100.0 +/- 0.0	99.95 +/- 0.24	98.69 +/- 4.0
<i>RASGRP2</i>	48.74 +/- 7.78	100.0 +/- 0.0	99.82 +/- 0.81	96.15 +/- 9.08
<i>RBM8A</i>	53.81 +/- 7.96	100.0 +/- 0.0	100.0 +/- 0.0	99.12 +/- 2.21
<i>RUNX1</i>	52.94 +/- 7.33	99.99 +/- 0.05	99.8 +/- 0.75	98.13 +/- 3.64
<i>SERPINC1</i>	54.25 +/- 7.49	100.0 +/- 0.0	100.0 +/- 0.0	99.8 +/- 0.71
<i>SERPIND1</i>	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 [%]
<i>SERPINE1</i>	50.74 +/- 7.92	100.0 +/- 0.0	99.9 +/- 0.67	98.1 +/- 6.63
<i>SERPINF2</i>	50.2 +/- 9.3	100.0 +/- 0.0	99.89 +/- 0.36	96.02 +/- 7.39
<i>SLFN14</i>	54.62 +/- 7.94	100.0 +/- 0.0	100.0 +/- 0.01	99.25 +/- 2.18
<i>SRC</i>	50.77 +/- 8.08	99.99 +/- 0.04	99.7 +/- 1.12	97.34 +/- 6.53
<i>STIM1</i>	49.63 +/- 7.38	99.99 +/- 0.03	99.91 +/- 0.3	97.61 +/- 5.11
<i>STXBP2</i>	51.74 +/- 9.08	100.0 +/- 0.0	99.83 +/- 0.82	97.49 +/- 5.75
<i>TBXA2R</i>	53.93 +/- 9.17	100.0 +/- 0.0	99.98 +/- 0.11	97.94 +/- 6.72
<i>TBXAS1</i>	52.3 +/- 8.21	100.0 +/- 0.0	99.98 +/- 0.12	98.59 +/- 5.7
<i>THBD</i>	54.08 +/- 7.98	100.0 +/- 0.0	99.9 +/- 0.37	98.18 +/- 5.04
<i>THPO</i>	48.8 +/- 8.23	100.0 +/- 0.0	99.98 +/- 0.07	97.43 +/- 5.75
<i>TPM4</i>	52.79 +/- 7.43	100.0 +/- 0.0	99.99 +/- 0.05	98.61 +/- 4.53
<i>TUBB1</i>	55.0 +/- 7.3	100.0 +/- 0.0	100.0 +/- 0.0	99.66 +/- 0.95
<i>VIPAS39</i>	53.27 +/- 8.33	100.0 +/- 0.0	99.95 +/- 0.31	98.67 +/- 3.87
<i>VKORC1</i>	50.72 +/- 8.8	100.0 +/- 0.0	99.97 +/- 0.21	97.48 +/- 9.27
<i>VPS33B</i>	52.71 +/- 8.11	100.0 +/- 0.0	99.94 +/- 0.35	99.06 +/- 2.64
<i>VWF</i>	52.06 +/- 7.8	100.0 +/- 0.0	99.99 +/- 0.04	98.7 +/- 3.84
<i>WAS</i>	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](mailto:mol-dia@rn.dk). Rapport genereret: 09/09-2024