

## Øjenmisdannelse

## Panelbeskrivelse

Navn: Øjenmisdannelse

version: 1.0

Ibrugtagningsdato: 01/06-2023

**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 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>ABCB6</i>	52.51 +/- 8.49	100.0 +/- 0.0	99.96 +/- 0.24	98.57 +/- 7.1
<i>ADAMTS17</i>	52.75 +/- 8.09	99.94 +/- 0.09	99.8 +/- 0.58	98.35 +/- 3.37
<i>ADAMTS18</i>	53.29 +/- 7.54	100.0 +/- 0.0	99.95 +/- 0.33	98.82 +/- 3.85
<i>ADAMTSL4</i>	47.77 +/- 8.51	100.0 +/- 0.0	99.82 +/- 0.5	95.92 +/- 7.96
<i>ALDH1A3</i>	54.3 +/- 7.71	100.0 +/- 0.0	99.94 +/- 0.32	99.07 +/- 2.48
<i>ALX1</i>	52.17 +/- 7.78	100.0 +/- 0.0	100.0 +/- 0.0	98.89 +/- 2.81
<i>ATOH7</i>	56.29 +/- 8.97	100.0 +/- 0.0	99.95 +/- 0.33	98.88 +/- 4.06
<i>B3GLCT</i>	54.5 +/- 7.44	99.86 +/- 0.09	99.86 +/- 0.09	99.11 +/- 1.48
<i>BCOR</i>	41.47 +/- 15.57	99.57 +/- 1.64	91.31 +/- 13.69	67.34 +/- 35.48
<i>BMP4</i>	54.43 +/- 7.75	100.0 +/- 0.0	100.0 +/- 0.0	98.78 +/- 3.56
<i>C12orf57</i>	49.91 +/- 7.86	100.0 +/- 0.0	99.84 +/- 1.09	98.39 +/- 6.52
<i>CAPN15</i>	52.89 +/- 9.44	99.9 +/- 0.63	98.72 +/- 2.25	93.52 +/- 7.52
<i>CDH2</i>	55.68 +/- 7.35	99.99 +/- 0.06	99.92 +/- 0.31	99.21 +/- 1.5
<i>CHD7</i>	53.95 +/- 7.71	100.0 +/- 0.0	99.91 +/- 0.23	98.79 +/- 2.42
<i>CHRD1</i>	42.13 +/- 15.98	99.92 +/- 0.25	93.64 +/- 10.14	68.51 +/- 36.9
<i>CLDN19</i>	49.64 +/- 8.98	100.0 +/- 0.0	99.84 +/- 1.0	96.88 +/- 9.06
<i>COL4A1</i>	53.48 +/- 7.53	100.0 +/- 0.0	99.99 +/- 0.04	98.83 +/- 3.9
<i>CPAMD8</i>	50.01 +/- 7.88	100.0 +/- 0.0	99.76 +/- 0.73	96.91 +/- 6.71
<i>CRIM1</i>	53.07 +/- 7.32	99.98 +/- 0.13	99.64 +/- 1.16	97.16 +/- 4.13
<i>CYP1B1</i>	56.24 +/- 7.0	100.0 +/- 0.0	100.0 +/- 0.03	99.59 +/- 1.33
<i>DYRK1A</i>	53.44 +/- 7.54	100.0 +/- 0.0	99.82 +/- 0.49	98.08 +/- 2.69
<i>ERCC1</i>	51.05 +/- 8.18	100.0 +/- 0.01	99.84 +/- 0.72	97.82 +/- 7.33
<i>EYA1</i>	55.89 +/- 7.45	100.0 +/- 0.0	100.0 +/- 0.0	99.62 +/- 0.99
<i>FGFR1</i>	52.45 +/- 8.49	100.0 +/- 0.0	99.9 +/- 0.47	97.99 +/- 6.49
<i>FOXC1</i>	46.75 +/- 7.72	100.0 +/- 0.0	99.25 +/- 2.61	91.23 +/- 12.68
<i>FOXC2</i>	49.23 +/- 8.25	100.0 +/- 0.01	99.68 +/- 1.66	95.47 +/- 10.36
<i>FOXE3</i>	46.47 +/- 8.23	100.0 +/- 0.0	99.3 +/- 2.22	93.29 +/- 13.76
<i>FOXL2</i>	50.59 +/- 8.73	100.0 +/- 0.0	99.31 +/- 2.64	93.97 +/- 10.49
<i>FRAS1</i>	53.65 +/- 7.53	100.0 +/- 0.0	100.0 +/- 0.03	99.33 +/- 1.76
<i>FREM1</i>	54.46 +/- 7.51	100.0 +/- 0.0	100.0 +/- 0.0	99.35 +/- 2.7
<i>FREM2</i>	55.08 +/- 7.76	100.0 +/- 0.0	99.99 +/- 0.06	99.35 +/- 2.4

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<i>FZD5</i>	52.29 +/- 7.59	100.0 +/- 0.0	99.9 +/- 0.57	98.3 +/- 5.13
<i>GDF3</i>	51.57 +/- 9.16	100.0 +/- 0.0	99.93 +/- 0.4	99.02 +/- 4.27
<i>GDF6</i>	51.24 +/- 7.45	100.0 +/- 0.0	99.91 +/- 0.29	97.76 +/- 5.85
<i>GJA1</i>	53.43 +/- 7.32	100.0 +/- 0.0	99.99 +/- 0.03	99.14 +/- 1.51
<i>GLI3</i>	54.8 +/- 7.68	100.0 +/- 0.0	99.99 +/- 0.06	99.24 +/- 2.65
<i>GRIP1</i>	55.15 +/- 7.55	100.0 +/- 0.0	100.0 +/- 0.0	99.68 +/- 1.07
<i>HCCS</i>	42.82 +/- 16.21	99.83 +/- 1.1	93.94 +/- 11.67	70.0 +/- 35.47
<i>HESX1</i>	55.26 +/- 7.73	100.0 +/- 0.0	99.92 +/- 0.54	99.44 +/- 2.26
<i>HMGGB3</i>	41.83 +/- 15.06	99.94 +/- 0.2	94.9 +/- 8.89	69.18 +/- 35.19
<i>HMX1</i>	48.36 +/- 9.52	100.0 +/- 0.0	99.38 +/- 2.74	93.22 +/- 11.77
<i>KDM6A</i>	41.97 +/- 15.7	99.88 +/- 0.7	93.59 +/- 9.67	68.65 +/- 35.83
<i>KERA</i>	54.77 +/- 7.11	100.0 +/- 0.0	100.0 +/- 0.0	99.66 +/- 1.18
<i>LAMB2</i>	53.73 +/- 8.75	100.0 +/- 0.0	99.98 +/- 0.16	98.94 +/- 4.73
<i>LRP2</i>	54.29 +/- 7.49	100.0 +/- 0.0	100.0 +/- 0.01	99.42 +/- 1.35
<i>MAB21L2</i>	55.24 +/- 7.95	99.98 +/- 0.12	98.89 +/- 0.99	95.95 +/- 4.03
<i>MFRP</i>	48.77 +/- 8.21	100.0 +/- 0.0	99.75 +/- 1.58	96.88 +/- 8.76
<i>MITF</i>	55.39 +/- 7.95	100.0 +/- 0.0	100.0 +/- 0.01	99.3 +/- 1.73
<i>MYRF</i>	49.48 +/- 8.09	100.0 +/- 0.03	99.77 +/- 1.06	96.8 +/- 7.49
<i>NAA10</i>	39.31 +/- 15.48	99.74 +/- 0.91	89.47 +/- 17.91	64.0 +/- 40.61
<i>NDP</i>	42.78 +/- 16.21	99.96 +/- 0.25	93.63 +/- 13.19	70.24 +/- 35.63
<i>NHS</i>	42.18 +/- 15.83	99.7 +/- 1.66	93.66 +/- 10.27	68.66 +/- 35.99
<i>OCRL</i>	40.92 +/- 15.61	99.87 +/- 0.59	91.55 +/- 13.83	66.68 +/- 38.47
<i>OLFM2</i>	47.81 +/- 6.84	100.0 +/- 0.0	99.44 +/- 1.21	93.31 +/- 8.17
<i>OTX2</i>	52.09 +/- 8.29	100.0 +/- 0.0	100.0 +/- 0.0	98.91 +/- 2.84
<i>PAX2</i>	45.55 +/- 7.78	100.0 +/- 0.01	99.64 +/- 1.67	93.15 +/- 12.12
<i>PAX6</i>	52.35 +/- 7.1	100.0 +/- 0.0	99.97 +/- 0.13	98.35 +/- 3.15
<i>PIGL</i>	53.89 +/- 7.6	100.0 +/- 0.0	100.0 +/- 0.0	99.23 +/- 3.17
<i>PITX2</i>	52.06 +/- 7.04	100.0 +/- 0.0	99.92 +/- 0.52	98.8 +/- 4.55
<i>PITX3</i>	48.0 +/- 9.2	100.0 +/- 0.0	99.59 +/- 2.39	95.06 +/- 12.63
<i>PORCN</i>	38.1 +/- 15.1	99.86 +/- 0.53	87.69 +/- 18.16	61.92 +/- 41.01
<i>PRSS56</i>	50.07 +/- 9.18	100.0 +/- 0.0	99.81 +/- 0.76	96.34 +/- 9.76
<i>PTCH1</i>	52.9 +/- 7.4	100.0 +/- 0.0	99.8 +/- 0.72	97.29 +/- 5.08
<i>PUF60</i>	48.42 +/- 8.66	100.0 +/- 0.0	99.9 +/- 0.43	96.01 +/- 6.9
<i>PXDN</i>	54.31 +/- 8.0	100.0 +/- 0.0	99.96 +/- 0.2	98.9 +/- 3.69
<i>RAB18</i>	55.27 +/- 7.64	100.0 +/- 0.0	100.0 +/- 0.0	99.65 +/- 1.29
<i>RAB3GAP1</i>	52.78 +/- 7.46	100.0 +/- 0.0	99.69 +/- 1.41	97.72 +/- 4.82
<i>RAB3GAP2</i>	55.11 +/- 6.94	100.0 +/- 0.0	99.99 +/- 0.03	99.4 +/- 1.17
<i>RARB</i>	55.3 +/- 7.74	99.98 +/- 0.04	99.98 +/- 0.04	99.43 +/- 1.9
<i>RAX</i>	50.56 +/- 8.58	100.0 +/- 0.02	99.94 +/- 0.21	97.45 +/- 7.23
<i>RBP4</i>	51.69 +/- 8.13	100.0 +/- 0.0	99.86 +/- 0.91	97.65 +/- 4.38
<i>RPGRIP1L</i>	54.61 +/- 7.24	100.0 +/- 0.0	100.0 +/- 0.0	99.66 +/- 0.88
<i>SALL2</i>	50.76 +/- 7.89	99.99 +/- 0.06	99.85 +/- 0.37	97.49 +/- 4.41
<i>SALL4</i>	53.12 +/- 7.99	100.0 +/- 0.0	99.93 +/- 0.33	98.85 +/- 3.94
<i>SHH</i>	52.29 +/- 7.5	100.0 +/- 0.0	99.88 +/- 0.67	97.86 +/- 4.67
<i>SIX3</i>	48.41 +/- 7.11	100.0 +/- 0.02	99.59 +/- 1.34	93.9 +/- 10.24
<i>SIX6</i>	52.61 +/- 7.95	99.95 +/- 0.11	99.51 +/- 0.41	97.98 +/- 5.83

Gen	Middelsekventeringsdybde [x]	Andel >= 10x [%]	Andel >= 20x [%]	Andel >= 30x [%]
<b>SLC38A8</b>	51.31 +/- 8.37	100.0 +/- 0.0	99.95 +/- 0.3	97.96 +/- 6.97
<b>SMCHD1</b>	55.35 +/- 7.34	99.92 +/- 0.04	99.87 +/- 0.21	99.16 +/- 1.74
<b>SMO</b>	49.62 +/- 7.83	100.0 +/- 0.0	99.66 +/- 1.61	97.11 +/- 7.12
<b>SMOC1</b>	50.95 +/- 8.35	100.0 +/- 0.0	99.83 +/- 0.78	97.5 +/- 6.52
<b>SOX2</b>	48.55 +/- 6.93	100.0 +/- 0.0	99.81 +/- 0.54	95.32 +/- 7.31
<b>STRA6</b>	49.83 +/- 8.42	100.0 +/- 0.0	99.91 +/- 0.57	97.75 +/- 8.12
<b>TBC1D20</b>	51.14 +/- 8.26	100.0 +/- 0.0	99.86 +/- 0.53	97.98 +/- 3.49
<b>TENM3</b>	56.25 +/- 7.38	100.0 +/- 0.0	100.0 +/- 0.0	99.55 +/- 1.51
<b>TFAP2A</b>	49.36 +/- 7.02	100.0 +/- 0.0	99.05 +/- 1.38	93.93 +/- 4.72
<b>TMEM67</b>	54.92 +/- 7.82	100.0 +/- 0.0	100.0 +/- 0.03	99.09 +/- 1.67
<b>TMEM98</b>	51.69 +/- 8.15	99.99 +/- 0.07	99.74 +/- 0.58	98.1 +/- 4.26
<b>TMX3</b>	54.87 +/- 7.69	100.0 +/- 0.0	99.99 +/- 0.07	99.31 +/- 1.69
<b>TUBGCP4</b>	52.97 +/- 7.93	100.0 +/- 0.0	99.93 +/- 0.46	98.83 +/- 3.39
<b>VAX1</b>	45.85 +/- 7.37	99.81 +/- 0.73	98.31 +/- 3.93	90.83 +/- 10.5
<b>VCAN</b>	54.22 +/- 7.4	99.99 +/- 0.03	99.95 +/- 0.05	99.47 +/- 1.4
<b>VPS35L</b>	53.24 +/- 7.82	100.0 +/- 0.0	99.97 +/- 0.21	99.1 +/- 2.58
<b>VSX1</b>	54.81 +/- 7.26	100.0 +/- 0.0	100.0 +/- 0.0	99.42 +/- 1.6
<b>VSX2</b>	51.4 +/- 8.44	100.0 +/- 0.01	99.98 +/- 0.13	98.03 +/- 6.43
<b>WDR37</b>	54.11 +/- 7.84	100.0 +/- 0.0	99.92 +/- 0.38	98.82 +/- 2.34
<b>YAP1</b>	52.66 +/- 7.14	100.0 +/- 0.0	99.74 +/- 0.43	97.38 +/- 3.28

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