

Supplementary information for article:

Optimised, Broad NGS Panel for Inherited Eye Diseases to Diagnose 1000 Patients in Poland

- 1) Gene lists for three subsequent NGS panel versions:
 - a) Gene list for the initial version of the panel:

HGNC ID	Gene Symbol	RefSeq ID
34	<i>ABCA4</i>	NM_000350.3
57	<i>ABCC6</i>	NM_001171.6
15868	<i>ABHD12</i>	NM_001042472.3
23338	<i>ACBD5</i>	NM_145698.5
118	<i>ACO2</i>	NM_001098.3
216	<i>ADAM9</i>	NM_003816.3
17110	<i>ADAMTS18</i>	NM_199355.4
13839	<i>ADGRA3</i>	NM_145290.4
17416	<i>ADGRV1</i>	NM_032119.4
24040	<i>ADIPOR1</i>	NM_015999.6
315	<i>AFG3L2</i>	NM_006796.3
26147	<i>AGBL5</i>	NM_021831.6
21575	<i>AHI1</i>	NM_001134831.2
348	<i>AHR</i>	NM_001621.5
359	<i>AIPL1</i>	NM_014336.5
428	<i>ALMS1</i>	NM_001378454.1
17090	<i>ARHGEF18</i>	NM_001367823.1
17146	<i>ARL2BP</i>	NM_012106.4
694	<i>ARL3</i>	NM_004311.4
13210	<i>ARL6</i>	NM_001278293.3
32685	<i>ARMS2</i>	NM_001099667.3
24102	<i>ARSG</i>	NM_001267727.2
16448	<i>ASRGL1</i>	NM_001083926.2
791	<i>ATF6</i>	NM_007348.4
10560	<i>ATXN7</i>	NM_001377405.1
28093	<i>BBIP1</i>	NM_001195305.3
966	<i>BBS1</i>	NM_024649.5
26291	<i>BBS10</i>	NM_024685.4
26648	<i>BBS12</i>	NM_152618.3
967	<i>BBS2</i>	NM_031885.5
969	<i>BBS4</i>	NM_033028.5
970	<i>BBS5</i>	NM_152384.3
18758	<i>BBS7</i>	NM_176824.3
30000	<i>BBS9</i>	NM_198428.3
12703	<i>BEST1</i>	NM_004183.4
14344	<i>C1QTNF5</i>	NM_001278431.2
1248	<i>C2</i>	NM_000063.6

1318	<i>C3</i>	NM_000064.4
1375	<i>CA4</i>	NM_000717.5
1386	<i>CABP4</i>	NM_145200.5
1393	<i>CACNA1F</i>	NM_001256789.3
20202	<i>CACNA2D4</i>	NM_172364.5
1482	<i>CAPN5</i>	NM_004055.5
29253	<i>CC2D2A</i>	NM_001378615.1
1615	<i>CCT2</i>	NM_006431.3
13733	<i>CDH23</i>	NM_022124.6
1762	<i>CDH3</i>	NM_001793.6
14550	<i>CDHR1</i>	NM_033100.4
29182	<i>CEP164</i>	NM_014956.5
28209	<i>CEP19</i>	NM_032898.5
1859	<i>CEP250</i>	NM_007186.6
29021	<i>CEP290</i>	NM_025114.4
25740	<i>CEP78</i>	NM_001330691.3
21699	<i>CERKL</i>	NM_201548.5
1260	<i>CFAP410</i>	NM_004928.3
27232	<i>CFAP418</i>	NM_177965.4
1037	<i>CFB</i>	NM_001710.6
4883	<i>CFH</i>	NM_000186.4
1940	<i>CHM</i>	NM_000390.4
24579	<i>CIB2</i>	NM_006383.4
29675	<i>CLCC1</i>	NM_001377458.1
2074	<i>CLN3</i>	NM_001042432.2
12605	<i>CLRN1</i>	NM_174878.3
19009	<i>CLUAP1</i>	NM_015041.3
2148	<i>CNGA1</i>	NM_001379270.1
2150	<i>CNGA3</i>	NM_001298.3
2151	<i>CNGB1</i>	NM_001297.5
2153	<i>CNGB3</i>	NM_019098.5
105	<i>CNNM4</i>	NM_020184.4
2186	<i>COL11A1</i>	NM_001854.4
2200	<i>COL2A1</i>	NM_001844.5
2217	<i>COL9A1</i>	NM_001851.6
2343	<i>CRB1</i>	NM_201253.3
2383	<i>CRX</i>	NM_000554.6
26193	<i>CSPP1</i>	NM_001382391.1
2509	<i>CTNNA1</i>	NM_001903.5
23198	<i>CYP4V2</i>	NM_207352.4
20603	<i>DHDDS</i>	NM_205861.3
17211	<i>DHX38</i>	NM_014003.4
2928	<i>DMD</i>	NM_004006.3
28769	<i>DRAM2</i>	NM_001349884.2
37261	<i>DTHD1</i>	NM_001170700.3
3218	<i>EFEMP1</i>	NM_001039348.3
14418	<i>ELOVL1</i>	NM_022821.4
14415	<i>ELOVL4</i>	NM_022726.4
28957	<i>EMC1</i>	NM_015047.3
3438	<i>ERCC6</i>	NM_000124.4

13281	<i>ESPN</i>	NM_031475.3
17097	<i>EXOSC2</i>	NM_014285.7
21555	<i>EYS</i>	NM_001142800.2
25808	<i>FAM161A</i>	NM_001201543.2
3602	<i>FBLN5</i>	NM_006329.4
24682	<i>FLVCR1</i>	NM_014053.4
3960	<i>FSCN2</i>	NM_012418.4
4042	<i>FZD4</i>	NM_012193.4
4221	<i>GDF6</i>	NM_001001557.4
4393	<i>GNAT1</i>	NM_144499.3
4394	<i>GNAT2</i>	NM_001377295.2
4400	<i>GNB3</i>	NM_002075.4
23026	<i>GNPTG</i>	NM_032520.5
31371	<i>GPR179</i>	NM_001004334.4
10013	<i>GRK1</i>	NM_002929.3
4598	<i>GRM6</i>	NM_000843.4
4678	<i>GUCA1A</i>	NM_001384910.1
4679	<i>GUCA1B</i>	NM_002098.6
4689	<i>GUCY2D</i>	NM_000180.4
4816	<i>HARS1</i>	NM_002109.6
26527	<i>HGSNAT</i>	NM_152419.3
4922	<i>HK1</i>	NM_000188.3
19194	<i>HMCN1</i>	NM_031935.3
5017	<i>HMX1</i>	NM_018942.3
9476	<i>HTRA1</i>	NM_002775.5
5385	<i>IDH3B</i>	NM_006899.5
29077	<i>IFT140</i>	NM_014714.4
30391	<i>IFT172</i>	NM_015662.3
18626	<i>IFT27</i>	NM_001177701.3
14313	<i>IFT81</i>	NM_014055.4
6052	<i>IMPDH1</i>	NM_000883.4
6055	<i>IMPG1</i>	NM_001563.4
18362	<i>IMPG2</i>	NM_016247.4
21474	<i>INPP5E</i>	NM_019892.6
17870	<i>INVS</i>	NM_014425.5
28949	<i>IQCB1</i>	NM_001023570.4
6174	<i>ITM2B</i>	NM_021999.5
6188	<i>JAG1</i>	NM_000214.3
6259	<i>KCNJ13</i>	NM_002242.4
19698	<i>KCNV2</i>	NM_133497.4
22219	<i>KIAA1549</i>	NM_001164665.2
6388	<i>KIF11</i>	NM_004523.4
15865	<i>KIZ</i>	NM_018474.6
15646	<i>KLHL7</i>	NM_001031710.3
6481	<i>LAMA1</i>	NM_005559.4
31923	<i>LCA5</i>	NM_001122769.3
6685	<i>LRAT</i>	NM_004744.5
24783	<i>LRIT3</i>	NM_198506.5
6697	<i>LRP5</i>	NM_002335.4
6741	<i>LZTFL1</i>	NM_020347.4

6816	<i>MAK</i>	NM_001242957.3
6888	<i>MAPKAPK3</i>	NM_001243925.2
7027	<i>MERTK</i>	NM_006343.3
16877	<i>MFN2</i>	NM_014874.4
18121	<i>MFRP</i>	NM_031433.4
28486	<i>MFSD8</i>	NM_001371596.2
31582	<i>MIR204</i>	NR_029621.1
7108	<i>MKKS</i>	NM_170784.3
7121	<i>MKS1</i>	NM_017777.4
26784	<i>MTRFR</i>	NM_152269.5
7467	<i>MTTP</i>	NM_001386140.1
7530	<i>MVK</i>	NM_000431.4
7606	<i>MYO7A</i>	NM_000260.4
15625	<i>NBAS</i>	NM_015909.4
7678	<i>NDP</i>	NM_000266.4
7745	<i>NEK2</i>	NM_002497.4
7762	<i>NEUROD1</i>	NM_002500.5
17877	<i>NMNAT1</i>	NM_022787.4
7905	<i>NPHP1</i>	NM_001128178.3
7907	<i>NPHP3</i>	NM_153240.5
19104	<i>NPHP4</i>	NM_015102.5
7974	<i>NR2E3</i>	NM_014249.4
7975	<i>NR2F1</i>	NM_005654.6
8002	<i>NRL</i>	NM_001354768.3
8082	<i>NYX</i>	NM_001378477.3
8091	<i>OAT</i>	NM_000274.4
2567	<i>OFD1</i>	NM_003611.3
8140	<i>OPA1</i>	NM_130837.3
8142	<i>OPA3</i>	NM_025136.4
1012	<i>OPN1SW</i>	NM_001385125.1
8522	<i>OTX2</i>	NM_021728.4
15894	<i>PANK2</i>	NM_001386393.1
8616	<i>PAX2</i>	NM_000278.5
34383	<i>PCARE</i>	NM_001029883.3
14674	<i>PCDH15</i>	NM_001384140.1
8754	<i>PCYT1A</i>	NM_001312673.2
8785	<i>PDE6A</i>	NM_000440.3
8786	<i>PDE6B</i>	NM_000283.4
8787	<i>PDE6C</i>	NM_006204.4
8789	<i>PDE6G</i>	NM_002602.4
8790	<i>PDE6H</i>	NM_006205.3
26257	<i>PDZD7</i>	NM_001195263.2
8850	<i>PEX1</i>	NM_000466.3
9717	<i>PEX2</i>	NM_000318.3
8860	<i>PEX7</i>	NM_000288.4
8896	<i>PGK1</i>	NM_000291.4
8940	<i>PHYH</i>	NM_006214.4
21043	<i>PITPNM3</i>	NM_031220.4
9038	<i>PLA2G5</i>	NM_000929.3
11397	<i>PLK4</i>	NM_014264.5

16268	<i>PNPLA6</i>	NM_001166114.2
30836	<i>POC1B</i>	NM_172240.3
26658	<i>POC5</i>	NM_001099271.2
19139	<i>POMGNT1</i>	NM_017739.4
32528	<i>PRCD</i>	NM_001077620.3
13998	<i>PRDM13</i>	NM_021620.4
9402	<i>PRKCG</i>	NM_002739.5
9454	<i>PROM1</i>	NM_006017.3
17348	<i>PRPF3</i>	NM_004698.4
15446	<i>PRPF31</i>	NM_015629.4
17349	<i>PRPF4</i>	NM_001244926.2
15860	<i>PRPF6</i>	NM_012469.4
17340	<i>PRPF8</i>	NM_006445.4
9942	<i>PRPH2</i>	NM_000322.5
9462	<i>PRPS1</i>	NM_002764.4
9768	<i>RAB28</i>	NM_001017979.3
18286	<i>RAX2</i>	NM_001319074.4
9884	<i>RB1</i>	NM_000321.3
9921	<i>RBP3</i>	NM_002900.3
9922	<i>RBP4</i>	NM_006744.4
18243	<i>RCBTB1</i>	NM_018191.4
19689	<i>RD3</i>	NM_001164688.2
17964	<i>RDH11</i>	NM_016026.4
19977	<i>RDH12</i>	NM_152443.3
9940	<i>RDH5</i>	NM_002905.5
30078	<i>REEP6</i>	NM_138393.4
9990	<i>RGR</i>	NM_001012720.2
10004	<i>RGS9</i>	NM_003835.4
30304	<i>RGS9BP</i>	NM_207391.3
10012	<i>RHO</i>	NM_000539.3
17282	<i>RIMS1</i>	NM_014989.7
10024	<i>RLBP1</i>	NM_000326.5
10254	<i>ROM1</i>	NM_000327.4
10263	<i>RP1</i>	NM_006269.2
15946	<i>RP1L1</i>	NM_178857.6
10274	<i>RP2</i>	NM_006915.3
10288	<i>RP9</i>	NM_203288.2
10294	<i>RPE65</i>	NM_000329.3
10295	<i>RPGR</i>	NM_001034853.2
13436	<i>RPGRIP1</i>	NM_020366.4
29168	<i>RPGRIP1L</i>	NM_015272.5
10457	<i>RS1</i>	NM_000330.4
18647	<i>RTN4IP1</i>	NM_032730.5
10521	<i>SAG</i>	NM_000541.5
28706	<i>SAMD11</i>	NM_001385641.1
10671	<i>SDCCAG8</i>	NM_006642.5
10729	<i>SEMA4A</i>	NM_022367.4
10975	<i>SLC24A1</i>	NM_004727.3
25198	<i>SLC25A46</i>	NM_138773.4
29326	<i>SLC7A14</i>	NM_020949.3

30859	<i>SNRNP200</i>	NM_014014.5
20423	<i>SPATA7</i>	NM_018418.5
11256	<i>SPP2</i>	NM_006944.3
11714	<i>TEAD1</i>	NM_021961.6
11817	<i>TIMM8A</i>	NM_004085.4
11822	<i>TIMP3</i>	NM_000362.5
11849	<i>TLR3</i>	NM_003265.3
11850	<i>TLR4</i>	NM_138554.5
25382	<i>TMEM126A</i>	NM_032273.4
25018	<i>TMEM216</i>	NM_001173990.3
14432	<i>TMEM237</i>	NM_001044385.3
21653	<i>TOPORS</i>	NM_005802.5
12269	<i>TREX1</i>	NM_033629.6
16380	<i>TRIM32</i>	NM_012210.4
17341	<i>TRNT1</i>	NM_182916.3
7146	<i>TRPM1</i>	NM_001252024.2
21641	<i>TSPAN12</i>	NM_012338.4
20087	<i>TTC8</i>	NM_144596.4
19963	<i>TTL5</i>	NM_015072.5
12404	<i>TPA</i>	NM_000370.3
12406	<i>TUB</i>	NM_177972.3
16691	<i>TUBGCP4</i>	NM_014444.5
18127	<i>TUBGCP6</i>	NM_020461.4
12423	<i>TULP1</i>	NM_003322.6
12565	<i>UNC119</i>	NM_005148.4
12597	<i>USH1C</i>	NM_153676.4
16356	<i>USH1G</i>	NM_173477.5
12601	<i>USH2A</i>	NM_206933.4
2464	<i>VCAN</i>	NM_004385.5
2183	<i>VPS13B</i>	NM_152564.5
28027	<i>WDPCP</i>	NM_015910.7
18340	<i>WDR19</i>	NM_025132.4
12762	<i>WFS1</i>	NM_006005.3
16361	<i>WHRN</i>	NM_015404.4
20041	<i>ZNF408</i>	NM_024741.3
16762	<i>ZNF423</i>	NM_001379286.1
26498	<i>ZNF513</i>	NM_144631.6

b) Genes added incrementally to the second panel version:

HGNC ID	Gene symbol	RefSeq ID
21014	<i>ANTXR1</i>	NM_032208.3
25419	<i>ARL13B</i>	NM_001174150.2
20730	<i>ARMC9</i>	NM_001352754.2
801	<i>ATP1A3</i>	NM_152296.5
24123	<i>B9D1</i>	NM_015681.6
28636	<i>B9D2</i>	NM_030578.4
24564	<i>C2CD3</i>	NM_001286577.2

28163	<i>CCDC28B</i>	NM_024296.5
3231	<i>CELSR2</i>	NM_001408.3
24866	<i>CEP104</i>	NM_014704.4
26690	<i>CEP120</i>	NM_001375405.1
12370	<i>CEP41</i>	NM_018718.3
24212	<i>CISD2</i>	NM_001008388.5
25801	<i>CPLANE1</i>	NM_001384732.1
10664	<i>CWC27</i>	NM_005869.4
16410	<i>DNAJC30</i>	NM_032317.3
2973	<i>DNM1L</i>	NM_012062.5
2962	<i>DYNC2H1</i>	NM_001377.3
28296	<i>DYNC2I2</i>	NM_052844.4
3360	<i>ENSA</i>	NM_004436.4
24659	<i>EXOC8</i>	NM_175876.5
29162	<i>FAM149B1</i>	NM_173348.2
3642	<i>FDXR</i>	NM_024417.5
8079	<i>FRMD7</i>	NM_194277.3
4392	<i>GNAS</i>	NM_000516.7
20145	<i>GPR143</i>	NM_000273.3
26558	<i>HYLS1</i>	NM_001134793.2
21424	<i>IFT74</i>	NM_025103.4
29068	<i>KATNIP</i>	NM_015202.5
19960	<i>KIAA0586</i>	NM_001329943.3
29110	<i>KIAA0753</i>	NM_014804.3
6320	<i>KIF3B</i>	NM_004798.4
30497	<i>KIF7</i>	NM_198525.3
29622	<i>MCAT</i>	NM_173467.5
25979	<i>MIEF1</i>	NM_019008.6
9936	<i>OPN1LW</i>	NM_020061.6
4206	<i>OPN1MW</i>	NM_000513.2
8788	<i>PDE6D</i>	NM_002601.4
8819	<i>PDXK</i>	NM_003681.5
18145	<i>PHF6</i>	NM_001015877.2
23352	<i>PIBF1</i>	NM_006346.4
9325	<i>PPT1</i>	NM_000310.4
9456	<i>PROS1</i>	NM_000313.4
10680	<i>SDHA</i>	NM_004168.4
11033	<i>SLC4A7</i>	NM_001321103.2
11237	<i>SPG7</i>	NM_003119.4
11317	<i>SSBP1</i>	NM_003143.3
16466	<i>SUFU</i>	NM_016169.4
26113	<i>TCTN1</i>	NM_001082538.3
25774	<i>TCTN2</i>	NM_024809.5
24519	<i>TCTN3</i>	NM_015631.6
28128	<i>TMEM107</i>	NM_183065.4
26944	<i>TMEM138</i>	NM_016464.5
37234	<i>TMEM231</i>	NM_001077418.3
28396	<i>TMEM67</i>	NM_153704.6
17861	<i>TRAF3IP1</i>	NM_015650.4
25660	<i>TTC21B</i>	NM_024753.5

12843	<i>YME1L1</i>	NM_014263.4
12309	<i>ZNHIT3</i>	NM_004773.4

c) Genes added incrementally to the third panel version:

HGNC ID	Gene symbol	RefSeq ID
360	<i>AIRE</i>	NM_000383.4
403	<i>ALDH3A2</i>	NM_000382.3
20917	<i>ALPK1</i>	NM_025144.4
451	<i>AMACR</i>	NM_014324.6
11891	<i>CLEC3B</i>	NM_003278.3
2076	<i>CLN5</i>	NM_006493.4
2077	<i>CLN6</i>	NM_017882.3
2079	<i>CLN8</i>	NM_018941.4
26169	<i>CTC1</i>	NM_025099.6
2514	<i>CTNNB1</i>	NM_001904.4
2529	<i>CTSD</i>	NM_001909.5
3439	<i>ERCC8</i>	NM_000082.4
3604	<i>FBN2</i>	NM_001999.4
20883	<i>GDPD1</i>	NM_182569.4
4601	<i>GRN</i>	NM_002087.4
23302	<i>HKDC1</i>	NM_025130.4
5384	<i>IDH3A</i>	NM_005530.3
29669	<i>IFT43</i>	NM_001102564.3
5961	<i>IKBKG</i>	NM_001099857.5
6694	<i>LRP2</i>	NM_004525.3
11957	<i>MED12</i>	NM_005120.3
24858	<i>MFF</i>	NM_001277062.2
24525	<i>MMACHC</i>	NM_015506.3
29678	<i>MSTO1</i>	NM_018116.4
7711	<i>NDUFS4</i>	NM_002495.4
8859	<i>PEX6</i>	NM_000287.4
8999	<i>PISD</i>	NM_001326411.2
9399	<i>PRKCD</i>	NM_006254.4
17283	<i>RIMS2</i>	NM_001348484.3
17296	<i>RRM2B</i>	NM_015713.5
13081	<i>SCAPER</i>	NM_020843.4
10892	<i>SIX6</i>	NM_007374.3
11052	<i>SLC6A6</i>	NM_003043.6
17884	<i>SPATA3</i>	NM_139073.5
25812	<i>SRD5A3</i>	NM_024592.5
11824	<i>TINF2</i>	NM_001099274.3
25295	<i>TLCD3B</i>	NM_031478.6
27344	<i>TMEM218</i>	NM_001258244.2
2073	<i>TPP1</i>	NM_000391.4
20771	<i>TUBB4B</i>	NM_006088.6
20080	<i>USP45</i>	NM_001346022.3
2183	<i>VPS13B</i>	NM_152564.5

18326	YPEL2	NM_001005404.4
20761	ZFYVE26	NM_015346.4

- 2) Calculation of the *ABCA4* haplotype frequency in causal variants with a comparison to previous data for the Polish population Ścieżyńska et al. 2016 [53]:

To compare the *ABCA4* complex haplotype p.(Leu541Pro;Ala1038Val) with the previous work [53], we restricted our set of positive samples to those with confirmed variants identified in the *ABCA4* gene, as the previous work only considered molecular findings in the *ABCA4* gene.

We calculated the frequency of the haplotype p.(Leu541Pro;Ala1038Val) in all positive patients from Supplementary table 2 of the Ścieżyńska et al. 2016 [53]. In 65 positive samples the haplotype p.(Leu541Pro;Ala1038Val) was identified 6 times as homozygote and 24 times as heterozygote. This gives a total of 36 alleles of the haplotype p.(Leu541Pro;Ala1038Val) for 130 alleles, which estimates the frequency of the haplotype p.(Leu541Pro;Ala1038Val) in positive samples to be 27.7%.

Subsequently, among 135 positive samples in our cohort with confirmatory variants in *ABCA4* gene, we found 12 cases with the haplotype p.(Leu541Pro;Ala1038Val) as homozygote and 59 cases with heterozygous haplotype p.(Leu541Pro;Ala1038Val) in compound heterozygote with other causal *ABCA4* variant. These findings led to haplotype p.(Leu541Pro;Ala1038Val) frequency estimation of about 30.7% (83 alleles with haplotype divided by 270 alleles in total). The difference in frequency between present study and the previous work of Ścieżyńska is not significant (two-tailed Fisher's exact probability test, $p > 0.05$).