

# WES VISION DISORDERS DG 2.18

| <i>Gene</i> | <i>Median coverage</i> | <i>% covered &gt;10x</i> | <i>% covered &gt;20x</i> | <i>OMIM disease ID</i>      |
|-------------|------------------------|--------------------------|--------------------------|-----------------------------|
| ABCA4       | 123.6                  | 99.9                     | 99.3                     | 248200;601718;153800;604116 |
| ABCC6       | 117.1                  | 93.6                     | 92.4                     | 264800;177850               |
| ABHD12      | 102.3                  | 98.7                     | 92.3                     | 612674                      |
| ACBD5       | 181.1                  | 100.0                    | 99.2                     | -                           |
| ACO2        | 122.9                  | 96.3                     | 90.3                     | 616289                      |
| ADAM9       | 174.5                  | 99.8                     | 99.1                     | 612775                      |
| ADAMTS18    | 163.4                  | 100.0                    | 99.7                     | 615458                      |
| ADAMTSL4    | 134.5                  | 100.0                    | 99.2                     | 225200                      |
| ADGRV1      | 155.0                  | 99.6                     | 98.6                     | 605472                      |
| ADIPOR1     | 111.9                  | 99.9                     | 97.8                     | -                           |
| AGBL1       | 122.0                  | 98.5                     | 98.5                     | 615523                      |
| AGBL5       | 124.5                  | 99.9                     | 99.3                     | 617023                      |
| AGK         | 136.1                  | 99.9                     | 97.6                     | 614691                      |
| AHI1        | 154.3                  | 99.7                     | 97.9                     | 608629                      |
| AHR         | 205.4                  | 100.0                    | 99.6                     | 618345                      |
| AIPL1       | 107.9                  | 100.0                    | 99.8                     | 604393                      |
| ALDH1A3     | 112.3                  | 97.2                     | 94.5                     | 615113                      |
| ALMS1       | 203.4                  | 99.8                     | 99.5                     | 203800                      |
| AP3B1       | 140.4                  | 99.2                     | 95.8                     | 608233                      |
| AP3D1       | 125.3                  | 99.8                     | 98.6                     | 617050                      |
| APOPT1      | 92.5                   | 81.9                     | 80.7                     | 220110                      |
| ARHGEF18    | 151.9                  | 95.4                     | 92.3                     | 617433                      |
| ARL13B      | 108.1                  | 100.0                    | 99.2                     | 612291                      |
| ARL2BP      | 81.9                   | 95.9                     | 88.3                     | 615434                      |
| ARL3        | 89.4                   | 100.0                    | 98.4                     | 618173                      |

|          |       |       |       |                                    |
|----------|-------|-------|-------|------------------------------------|
| ARL6     | 127.8 | 99.9  | 98.6  | 613575;209900                      |
| ARR3     | 102.2 | 100.0 | 99.8  | 301010                             |
| ARSG     | 132.9 | 100.0 | 99.5  | 618144                             |
| ASB10    | 97.2  | 99.4  | 95.7  | 603383                             |
| ASPH     | 129.9 | 99.9  | 98.8  | 601552                             |
| ASRGL1   | 140.9 | 100.0 | 100.0 | -                                  |
| ATF6     | 158.7 | 100.0 | 99.9  | 616517                             |
| ATOH7    | 103.5 | 96.0  | 91.2  | 221900                             |
| B3GLCT   | 116.4 | 99.6  | 96.3  | 261540                             |
| BBIP1    | 152.5 | 98.6  | 92.4  | 615995                             |
| BBS1     | 172.8 | 100.0 | 100.0 | 209900                             |
| BBS10    | 168.2 | 100.0 | 99.8  | 615987                             |
| BBS12    | 212.9 | 100.0 | 100.0 | 615989                             |
| BBS2     | 180.0 | 100.0 | 99.5  | 616562;615981                      |
| BBS4     | 127.5 | 99.9  | 99.3  | 615982                             |
| BBS5     | 113.0 | 99.0  | 93.9  | 615983                             |
| BBS7     | 173.7 | 98.7  | 95.5  | 615984                             |
| BBS9     | 131.5 | 99.7  | 97.6  | 615986                             |
| BCOR     | 118.0 | 99.6  | 97.4  | 300166                             |
| BEST1    | 147.6 | 99.4  | 96.4  | 611809;613194;153700;608161;193220 |
| BFSP1    | 111.8 | 99.0  | 89.9  | 611391                             |
| BFSP2    | 94.5  | 99.8  | 97.6  | 611597                             |
| BLOC1S3  | 40.2  | 98.5  | 81.3  | 614077                             |
| BLOC1S6  | 117.5 | 99.9  | 97.1  | 614171                             |
| BMP4     | 178.6 | 100.0 | 100.0 | 607932                             |
| C12orf65 | 122.8 | 99.8  | 98.5  | 613559;615035                      |
| C19orf12 | 122.6 | 100.0 | 99.8  | 614298                             |
| C1QTNF5  | 118.0 | 90.9  | 78.5  | 605670                             |
| C21orf2  | 107.3 | 100.0 | 99.3  | 617547                             |
| C2orf71  | 124.3 | 99.6  | 98.5  | 613428                             |

|          |       |       |       |                                    |
|----------|-------|-------|-------|------------------------------------|
| C5orf42  | 148.8 | 99.7  | 98.4  | 614615                             |
| C8orf37  | 152.0 | 100.0 | 99.4  | 614500;617406                      |
| CA4      | 170.8 | 100.0 | 100.0 | 600852                             |
| CABP4    | 165.3 | 100.0 | 99.9  | 610427                             |
| CACNA1F  | 98.4  | 99.7  | 97.5  | 300600;300476;300071               |
| CACNA2D4 | 107.2 | 98.9  | 97.7  | 610478                             |
| CAPN5    | 155.8 | 100.0 | 100.0 | 193235                             |
| CC2D2A   | 133.1 | 99.7  | 97.7  | 612285;216360;612284               |
| CCT2     | 172.5 | 100.0 | 100.0 | -                                  |
| CDH23    | 187.3 | 100.0 | 100.0 | 601067                             |
| CDH3     | 158.6 | 100.0 | 99.5  | 601553;225280                      |
| CDHR1    | 162.2 | 99.2  | 98.1  | 613660                             |
| CDK10    | 124.8 | 100.0 | 99.9  | -                                  |
| CEP164   | 102.8 | 99.9  | 98.3  | 614845                             |
| CEP250   | 102.9 | 100.0 | 99.2  | 618358                             |
| CEP290   | 97.0  | 96.1  | 90.0  | 610188;611134;610189;615991;611755 |
| CEP41    | 92.6  | 99.8  | 97.4  | 614464                             |
| CEP78    | 130.5 | 99.7  | 97.6  | 617236                             |
| CEP83    | 133.1 | 99.8  | 97.4  | 615862                             |
| CERKL    | 124.1 | 99.5  | 96.9  | 608380                             |
| CFH      | 194.1 | 99.9  | 99.0  | 126700                             |
| CHD7     | 158.7 | 100.0 | 99.5  | 214800                             |
| CHM      | 113.9 | 98.5  | 94.5  | 303100                             |
| CHMP4B   | 147.0 | 100.0 | 99.3  | 605387                             |
| CHRD1    | 111.5 | 100.0 | 99.8  | 309300                             |
| CHST6    | 197.7 | 100.0 | 100.0 | 217800                             |
| CIB2     | 203.3 | 99.7  | 97.0  | 614869                             |
| CISD2    | 135.4 | 83.4  | 83.4  | 604928                             |
| CLCC1    | 121.1 | 99.8  | 98.0  | -                                  |
| CLN3     | 133.0 | 92.5  | 91.8  | 204200                             |

|         |       |       |       |                      |
|---------|-------|-------|-------|----------------------|
| CLN5    | 164.8 | 99.4  | 95.5  | 256731               |
| CLN6    | 141.3 | 99.9  | 97.1  | 601780               |
| CLN8    | 173.9 | 83.5  | 83.5  | 600143               |
| CLRN1   | 168.2 | 100.0 | 99.8  | 614180;276902        |
| CLUAP1  | 155.6 | 100.0 | 99.8  | 204000               |
| CNGA1   | 129.3 | 91.7  | 86.3  | 613756               |
| CNGA3   | 173.8 | 100.0 | 99.7  | 216900               |
| CNGB1   | 121.0 | 99.4  | 97.5  | 613767               |
| CNGB3   | 133.7 | 99.4  | 95.9  | 248200;262300        |
| CNNM4   | 176.3 | 99.8  | 98.9  | 217080               |
| COL11A1 | 116.2 | 99.2  | 95.7  | 604841;154780        |
| COL18A1 | 123.2 | 98.1  | 95.6  | 267750               |
| COL25A1 | 155.4 | 99.5  | 99.1  | 616219               |
| COL2A1  | 125.9 | 100.0 | 99.7  | 108300               |
| COL4A1  | 115.6 | 98.7  | 97.4  | 175780               |
| COL8A2  | 89.1  | 99.9  | 97.9  | 136800;609140        |
| COL9A1  | 159.8 | 100.0 | 99.2  | 614134               |
| COL9A2  | 103.5 | 99.9  | 99.0  | 614284               |
| COL9A3  | 109.1 | 98.7  | 95.5  | -                    |
| CPAMD8  | 105.4 | 95.8  | 92.8  | 617319               |
| CPSF1   | 152.5 | 98.2  | 96.5  | 618827               |
| CRB1    | 194.4 | 100.0 | 99.9  | 600105;613835;172870 |
| CRX     | 215.1 | 100.0 | 100.0 | 120970;613829        |
| CRYAA   | 121.9 | 99.9  | 97.5  | 604219               |
| CRYAB   | 108.3 | 100.0 | 99.2  | 613763               |
| CRYBA1  | 134.4 | 100.0 | 99.4  | 600881               |
| CRYBA2  | 163.0 | 100.0 | 100.0 | 115900               |
| CRYBA4  | 126.8 | 100.0 | 100.0 | 610425               |
| CRYBB1  | 129.8 | 100.0 | 100.0 | 611544               |
| CRYBB2  | 149.9 | 100.0 | 100.0 | 601547               |

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|--------|-------|-------|-------|---------------|
| CRYBB3 | 168.8 | 100.0 | 100.0 | 609741        |
| CRYGB  | 111.7 | 100.0 | 99.6  | 615188        |
| CRYGC  | 137.6 | 99.8  | 96.9  | 604307        |
| CRYGD  | 112.0 | 100.0 | 98.9  | 115700        |
| CRYGS  | 109.8 | 94.1  | 86.6  | 116100        |
| CSPP1  | 132.4 | 99.8  | 98.7  | 615636        |
| CTDP1  | 129.4 | 88.4  | 84.3  | 604168        |
| CTNNA1 | 125.7 | 99.3  | 98.1  | 608970        |
| CTNNB1 | 148.9 | 100.0 | 100.0 | 617272        |
| CTSD   | 174.3 | 98.4  | 95.0  | 610127        |
| CWC27  | 105.7 | 99.3  | 96.5  | 250410        |
| CYP1B1 | 97.2  | 100.0 | 100.0 | 231300        |
| CYP4V2 | 163.7 | 99.9  | 98.4  | 210370        |
| DCN    | 162.4 | 95.7  | 95.6  | 610048        |
| DDHD1  | 164.0 | 97.9  | 95.8  | -             |
| DHDDS  | 93.1  | 99.0  | 95.0  | 613861        |
| DHX38  | 120.3 | 100.0 | 99.3  | 618220        |
| DKC1   | 108.6 | 99.8  | 98.7  | 305000        |
| DNM1L  | 139.3 | 99.9  | 98.5  | 610708        |
| DNMBP  | 163.7 | 100.0 | 99.7  | 618415        |
| DRAM2  | 158.7 | 100.0 | 99.9  | 616502        |
| DTNBP1 | 126.7 | 99.8  | 98.7  | 614076        |
| EFEMP1 | 163.7 | 100.0 | 99.9  | 126600        |
| ELOVL1 | 94.0  | 99.8  | 97.6  | 618527        |
| ELOVL4 | 122.7 | 100.0 | 99.5  | 600110        |
| EMC1   | 133.6 | 100.0 | 99.3  | 616875        |
| EPG5   | 137.8 | 99.5  | 98.5  | 242840        |
| EPHA2  | 158.5 | 100.0 | 99.5  | 116600        |
| EXOSC2 | 117.6 | 100.0 | 100.0 | 617763        |
| EYA1   | 145.8 | 99.9  | 99.7  | 113650;602588 |

|         |       |       |       |               |
|---------|-------|-------|-------|---------------|
| EYS     | 159.0 | 99.7  | 98.2  | 602772        |
| FA2H    | 96.4  | 92.0  | 83.1  | 612319        |
| FAM161A | 167.2 | 100.0 | 99.7  | 606068        |
| FLVCR1  | 153.2 | 100.0 | 98.9  | 609033        |
| FOXC1   | 55.4  | 98.0  | 89.6  | 602482;601631 |
| FOXE3   | 46.0  | 82.6  | 72.0  | 610256;612968 |
| FREM1   | 135.0 | 99.9  | 99.1  | 248450        |
| FRMD7   | 115.7 | 99.9  | 99.1  | 310700        |
| FTL     | 144.1 | 98.5  | 89.4  | 600886        |
| FYCO1   | 124.7 | 100.0 | 99.9  | 610019        |
| FZD4    | 188.5 | 100.0 | 100.0 | 133780        |
| GALK1   | 142.1 | 100.0 | 99.1  | 230200        |
| GALM    | 106.1 | 100.0 | 99.9  | -             |
| GALT    | 167.6 | 100.0 | 99.7  | 230400        |
| GCNT2   | 180.0 | 99.5  | 99.5  | 116700        |
| GDF3    | 138.9 | 100.0 | 100.0 | 613703;613704 |
| GDF6    | 116.8 | 100.0 | 99.9  | 615360;613094 |
| GFER    | 91.5  | 99.6  | 93.9  | 613076        |
| GJA1    | 187.7 | 100.0 | 100.0 | 257850;164200 |
| GJA3    | 147.7 | 100.0 | 99.7  | 601885        |
| GJA8    | 173.4 | 100.0 | 100.0 | 116200        |
| GNAT1   | 161.6 | 100.0 | 100.0 | 610444;616389 |
| GNAT2   | 119.6 | 99.9  | 99.0  | 613856        |
| GNB3    | 153.9 | 100.0 | 100.0 | 617024        |
| GNPTG   | 156.0 | 99.1  | 94.3  | 252605        |
| GPR143  | 65.2  | 85.8  | 76.4  | 300500;300814 |
| GPR179  | 158.0 | 100.0 | 100.0 | 614515        |
| GRHL2   | 139.4 | 100.0 | 100.0 | 618031        |
| GRK1    | 159.5 | 100.0 | 100.0 | 613411        |
| GRM6    | 137.8 | 90.2  | 80.6  | 257270        |

|        |       |       |       |               |
|--------|-------|-------|-------|---------------|
| GSN    | 117.2 | 95.8  | 93.5  | 105120        |
| GUCA1A | 177.2 | 100.0 | 100.0 | 602093        |
| GUCA1B | 139.5 | 100.0 | 100.0 | 613827        |
| GUCY2D | 100.0 | 99.6  | 96.2  | 601777;204000 |
| HARS   | 152.0 | 100.0 | 100.0 | 614504        |
| HCCS   | 110.5 | 99.8  | 97.6  | 309801        |
| HGSNAT | 125.5 | 86.4  | 86.3  | 252930;616544 |
| HK1    | 140.2 | 100.0 | 100.0 | 617460        |
| HMX1   | 22.5  | 62.4  | 42.9  | 612109        |
| HPS1   | 123.1 | 100.0 | 100.0 | 203300        |
| HPS3   | 165.7 | 99.7  | 97.5  | 614072        |
| HPS4   | 142.1 | 100.0 | 100.0 | 614073        |
| HPS5   | 142.5 | 100.0 | 99.7  | 614074        |
| HPS6   | 172.2 | 97.1  | 89.0  | 614075        |
| HRAS   | 177.9 | 100.0 | 100.0 | 218040        |
| HSF4   | 131.0 | 99.6  | 97.2  | 116800        |
| HSPG2  | 122.2 | 99.2  | 97.7  | 255800        |
| IDH3B  | 150.2 | 95.4  | 95.4  | 612572        |
| IFT140 | 124.5 | 99.8  | 98.8  | 266920;617781 |
| IFT172 | 107.8 | 99.9  | 99.1  | 616394;615630 |
| IFT27  | 135.3 | 100.0 | 100.0 | 615996        |
| IFT43  | 134.3 | 100.0 | 100.0 | 617871        |
| IFT74  | 95.2  | 98.4  | 93.9  | 617119        |
| IFT81  | 108.2 | 93.5  | 90.1  | -             |
| IMPDH1 | 57.5  | 87.9  | 80.2  | 613837;180105 |
| IMPG1  | 110.6 | 99.7  | 98.5  | 616151        |
| IMPG2  | 150.2 | 99.8  | 98.4  | 616152;613581 |
| INPP5E | 107.5 | 97.1  | 92.7  | 213300;610156 |
| INVS   | 163.7 | 100.0 | 100.0 | -             |
| IQCB1  | 113.4 | 93.9  | 85.0  | 609254        |

|          |       |       |       |               |
|----------|-------|-------|-------|---------------|
| IRX1     | 135.1 | 87.4  | 81.3  | -             |
| ITPR1    | 152.5 | 100.0 | 99.9  | 206700        |
| JAG1     | 147.4 | 97.7  | 96.8  | 118450        |
| JAM3     | 149.8 | 100.0 | 99.9  | 613730        |
| KCNJ13   | 155.0 | 100.0 | 100.0 | 614186;193230 |
| KCNV2    | 111.4 | 100.0 | 99.9  | 610356        |
| KERA     | 204.3 | 100.0 | 100.0 | 217300        |
| KIAA1549 | 133.5 | 97.9  | 96.4  | 618613        |
| KIF11    | 105.2 | 97.6  | 94.8  | 152950        |
| KIF21A   | 147.7 | 99.9  | 99.3  | 135700        |
| KIF7     | 100.0 | 93.6  | 90.6  | 200990        |
| KIZ      | 170.2 | 100.0 | 99.2  | 615780        |
| KLHL7    | 137.3 | 99.9  | 99.8  | 612943        |
| KRT12    | 167.0 | 99.7  | 97.8  | 122100        |
| KRT3     | 129.0 | 100.0 | 100.0 | 122100        |
| LAMA1    | 142.0 | 100.0 | 99.7  | 615960        |
| LAMB2    | 171.7 | 100.0 | 99.7  | 609049        |
| LAMP2    | 108.9 | 99.2  | 95.6  | 300257        |
| LCA5     | 162.9 | 99.9  | 99.2  | 604537        |
| LEMD2    | 89.6  | 98.7  | 92.0  | 212500        |
| LIM2     | 120.6 | 100.0 | 100.0 | 615277        |
| LRAT     | 283.9 | 100.0 | 100.0 | 613341        |
| LRIT3    | 116.1 | 93.9  | 91.9  | 615058        |
| LRMDA    | 143.3 | 96.8  | 95.6  | 615179        |
| LRP2     | 171.0 | 100.0 | 99.9  | 222448        |
| LRP5     | 179.9 | 98.5  | 98.1  | 601813;259770 |
| LRPAP1   | 121.2 | 100.0 | 100.0 | 615431        |
| LSS      | 138.4 | 100.0 | 99.9  | 616509        |
| LTBP2    | 112.8 | 99.9  | 99.0  | 613086        |
| LYST     | 169.3 | 99.6  | 98.3  | 214500        |



|          |       |       |       |                      |
|----------|-------|-------|-------|----------------------|
| LZTFL1   | 132.0 | 99.9  | 99.2  | 615994               |
| MAB21L2  | 181.5 | 100.0 | 100.0 | 615877               |
| MAF      | 79.2  | 83.5  | 78.0  | 610202               |
| MAK      | 174.4 | 98.7  | 96.8  | 614181               |
| MAPKAPK3 | 97.0  | 100.0 | 99.6  | 617111               |
| MERTK    | 159.6 | 99.5  | 98.8  | 613862               |
| MFN2     | 138.2 | 100.0 | 99.9  | 601152;609260        |
| MFRP     | 139.4 | 100.0 | 100.0 | 611040;609549        |
| MFSD8    | 143.3 | 100.0 | 99.7  | 610951;616170        |
| MIP      | 114.2 | 100.0 | 98.9  | 615274               |
| MIR184   |       |       |       | 614303               |
| MITF     | 160.6 | 100.0 | 99.9  | 103470               |
| MKKS     | 193.6 | 83.2  | 83.2  | 209900               |
| MKS1     | 104.7 | 99.8  | 97.9  | 249000;617121;615990 |
| MVK      | 120.8 | 90.9  | 90.5  | 610377;260920        |
| MYO7A    | 137.0 | 99.3  | 97.4  | 276900               |
| MYOC     | 161.9 | 100.0 | 98.6  | 137750               |
| NAA10    | 116.8 | 99.7  | 98.5  | 309800               |
| NBAS     | 169.2 | 100.0 | 99.6  | 614800               |
| NDP      | 89.0  | 100.0 | 99.7  | 310600;305390        |
| NDUFS2   | 118.5 | 100.0 | 100.0 | 252010               |
| NEK2     | 96.6  | 99.7  | 95.5  | 615565               |
| NEUROD1  | 152.8 | 100.0 | 99.1  | -                    |
| NHS      | 128.1 | 95.4  | 93.9  | 302350;302200        |
| NMNAT1   | 131.6 | 100.0 | 99.2  | 608553               |
| NPHP1    | 152.0 | 100.0 | 99.0  | 266900;609583        |
| NPHP3    | 143.4 | 99.7  | 98.4  | 267010               |
| NPHP4    | 137.8 | 100.0 | 99.8  | 606996               |
| NR2E3    | 103.9 | 100.0 | 99.6  | 611131;268100        |
| NR2F1    | 182.5 | 100.0 | 100.0 | 615722               |

|        |       |       |       |                                    |
|--------|-------|-------|-------|------------------------------------|
| NRL    | 93.0  | 99.5  | 94.8  | 613750                             |
| NYX    | 95.7  | 96.3  | 94.1  | 310500                             |
| OAT    | 87.0  | 85.2  | 76.3  | 258870                             |
| OCA2   | 128.6 | 99.9  | 98.7  | 203200                             |
| OCRL   | 127.1 | 99.9  | 98.6  | 309000                             |
| OFD1   | 61.2  | 88.0  | 73.7  | 300424;300804                      |
| OPA1   | 153.3 | 99.7  | 97.6  | 165500;125250                      |
| OPA3   | 124.4 | 100.0 | 99.0  | 258501;165300                      |
| OPN1LW | 77.1  | 67.2  | 60.6  | 303700                             |
| OPN1MW | 68.2  | 66.3  | 58.5  | 303700                             |
| OPTN   | 119.9 | 100.0 | 99.9  | 137760                             |
| OTX2   | 152.3 | 100.0 | 99.7  | 610125                             |
| OVOL2  | 111.0 | 95.7  | 89.5  | 122000                             |
| P3H2   | 110.8 | 99.8  | 98.0  | 614292                             |
| P4HA2  | 138.5 | 100.0 | 99.2  | 617238                             |
| PANK2  | 185.6 | 100.0 | 99.3  | 234200;607236                      |
| PAX2   | 201.8 | 100.0 | 99.9  | 120330                             |
| PAX6   | 141.4 | 100.0 | 100.0 | 106210;120430;120200;136520;604229 |
| PCDH15 | 169.5 | 98.6  | 97.5  | 602083;601067                      |
| PCYT1A | 113.2 | 98.9  | 95.5  | 608940                             |
| PDE6A  | 120.4 | 100.0 | 99.6  | 613810                             |
| PDE6B  | 155.9 | 100.0 | 99.9  | 613801;163500                      |
| PDE6C  | 136.5 | 99.9  | 97.8  | 613093                             |
| PDE6D  | 128.7 | 100.0 | 100.0 | 615665                             |
| PDE6G  | 140.0 | 100.0 | 99.7  | 613582                             |
| PDE6H  | 73.7  | 100.0 | 97.9  | 610024                             |
| PDZD7  | 90.4  | 98.8  | 96.3  | 605472                             |
| PET100 | 107.1 | 100.0 | 99.6  | 220110                             |
| PEX1   | 155.5 | 99.9  | 99.4  | 214100;601539                      |
| PEX2   | 158.8 | 100.0 | 100.0 | 614867;614866                      |

|         |       |       |       |                                    |
|---------|-------|-------|-------|------------------------------------|
| PEX26   | 94.0  | 100.0 | 100.0 | 614872                             |
| PEX6    | 113.0 | 94.5  | 86.7  | 614862                             |
| PEX7    | 135.9 | 87.8  | 80.7  | 614879;215100                      |
| PGK1    | 50.7  | 92.8  | 79.3  | 300653                             |
| PHOX2A  | 36.9  | 91.6  | 72.7  | 602078                             |
| PHYH    | 89.1  | 100.0 | 99.6  | 266500                             |
| PIKFYVE | 164.2 | 99.9  | 99.4  | 121850                             |
| PITX2   | 158.5 | 99.9  | 97.7  | 137600                             |
| PITX3   | 59.0  | 100.0 | 98.4  | 610623;107250                      |
| PLA2G5  | 144.8 | 100.0 | 100.0 | 228980                             |
| PLK4    | 178.1 | 99.9  | 98.2  | 616171                             |
| PNPLA6  | 139.7 | 100.0 | 99.7  | 275400;215470;245800               |
| POC1B   | 96.6  | 99.8  | 98.8  | 615973                             |
| POC5    | 168.8 | 99.7  | 97.6  | -                                  |
| POMGNT1 | 131.5 | 100.0 | 99.9  | 617123                             |
| PPT1    | 164.4 | 90.3  | 90.3  | 256730                             |
| PRCD    | 125.0 | 100.0 | 100.0 | 610599                             |
| PRDM13  | 133.0 | 99.2  | 94.1  | 136550                             |
| PRDM5   | 162.7 | 99.9  | 99.2  | 614170                             |
| PRIMPOL | 138.1 | 97.5  | 94.6  | 615421                             |
| PROM1   | 133.3 | 97.2  | 96.1  | 608051;612095;612657;603786        |
| PRPF3   | 90.5  | 98.8  | 95.3  | 601414                             |
| PRPF31  | 133.4 | 100.0 | 98.7  | 600138                             |
| PRPF4   | 148.3 | 100.0 | 99.8  | 615922                             |
| PRPF6   | 123.4 | 100.0 | 99.8  | 613983                             |
| PRPF8   | 122.4 | 100.0 | 99.3  | 600059                             |
| PRPH2   | 242.9 | 100.0 | 100.0 | 608133;169150;136880;608161;613105 |
| PRSS56  | 76.9  | 99.9  | 96.4  | 613517                             |
| PXDN    | 150.6 | 100.0 | 99.6  | 269400                             |
| RAB28   | 80.2  | 99.7  | 96.0  | 615374                             |

|          |       |       |       |                             |
|----------|-------|-------|-------|-----------------------------|
| RAB3GAP2 | 108.0 | 99.5  | 97.0  | 614225                      |
| RARB     | 109.5 | 100.0 | 100.0 | 615524                      |
| RAX      | 96.0  | 96.0  | 87.0  | 611038                      |
| RAX2     | 76.5  | 100.0 | 92.3  | 610381                      |
| RBP3     | 150.0 | 100.0 | 100.0 | 615233                      |
| RBP4     | 140.5 | 99.9  | 97.7  | 615147;616428               |
| RCBTB1   | 119.9 | 99.9  | 99.5  | 617175                      |
| RD3      | 147.5 | 100.0 | 100.0 | 610612                      |
| RDH11    | 107.7 | 100.0 | 99.0  | 616108                      |
| RDH12    | 102.1 | 100.0 | 98.6  | 612712                      |
| RDH5     | 162.8 | 100.0 | 99.9  | 136880                      |
| REEP6    | 193.7 | 100.0 | 100.0 | 617304                      |
| RGS9     | 121.1 | 98.5  | 97.1  | 608415                      |
| RGS9BP   | 94.3  | 99.6  | 95.0  | 608415                      |
| RHO      | 172.6 | 100.0 | 100.0 | 613731;610445               |
| RIMS1    | 138.1 | 99.8  | 97.7  | 603649                      |
| RLBP1    | 133.4 | 100.0 | 99.9  | 607476;607475;136880        |
| ROM1     | 115.6 | 100.0 | 99.9  | 608133                      |
| RP1      | 124.0 | 91.5  | 90.6  | 180100                      |
| RP1L1    | 135.3 | 100.0 | 100.0 | 613587;618826               |
| RP2      | 177.8 | 100.0 | 99.8  | 312600                      |
| RP9      | 67.9  | 80.8  | 75.9  | 180104                      |
| RPE65    | 153.2 | 99.8  | 97.8  | 204100;618697;613794        |
| RPGR     | 76.0  | 77.3  | 72.7  | 300455;304020;300834;300029 |
| RPGRIP1  | 146.2 | 100.0 | 99.9  | 613826;608194               |
| RPGRIP1L | 155.5 | 96.7  | 95.7  | 611561;611560;216360        |
| RS1      | 60.6  | 99.8  | 93.9  | 312700                      |
| RTN4IP1  | 96.9  | 99.9  | 98.7  | 616732                      |
| SAG      | 150.1 | 100.0 | 100.0 | 613758;258100               |
| SAMD11   | 94.9  | 90.2  | 81.6  | -                           |

|          |       |       |       |                      |
|----------|-------|-------|-------|----------------------|
| SC5D     | 192.4 | 100.0 | 99.5  | 6607330              |
| SCAPER   | 177.8 | 99.7  | 98.2  | 618195               |
| SCO2     | 107.3 | 100.0 | 100.0 | 608908               |
| SDCCAG8  | 138.0 | 100.0 | 99.9  | 613615               |
| SEMA4A   | 134.8 | 100.0 | 99.8  | 610283;610282        |
| SGSH     | 149.1 | 94.4  | 94.1  | 252900               |
| SHH      | 128.7 | 100.0 | 99.5  | 611638               |
| SIPA1L3  | 171.8 | 99.9  | 98.9  | 616851               |
| SIX6     | 263.6 | 100.0 | 100.0 | 212550               |
| SLC16A12 | 152.1 | 100.0 | 99.9  | 612018               |
| SLC24A1  | 169.3 | 100.0 | 99.9  | 613830               |
| SLC24A5  | 113.8 | 99.9  | 99.1  | 113750               |
| SLC25A46 | 199.3 | 99.7  | 97.3  | 616505               |
| SLC33A1  | 156.2 | 99.9  | 98.9  | 614482               |
| SLC38A8  | 84.4  | 99.9  | 97.3  | 609218               |
| SLC39A5  | 145.3 | 99.9  | 99.0  | 615946               |
| SLC45A2  | 137.7 | 100.0 | 99.9  | 606574               |
| SLC4A11  | 174.2 | 100.0 | 99.9  | 217700;217400;613268 |
| SLC52A2  | 170.8 | 100.0 | 100.0 | 614707               |
| SLC7A14  | 157.5 | 100.0 | 100.0 | 615725               |
| SMOC1    | 132.3 | 99.9  | 98.4  | 206920               |
| SNRNP200 | 139.0 | 99.9  | 99.1  | 610359               |
| SOX2     | 207.1 | 100.0 | 100.0 | 206900               |
| SOX5     | 112.9 | 99.9  | 98.9  | 616803               |
| SPATA7   | 138.4 | 99.8  | 98.2  | 604232               |
| SPP2     | 123.7 | 100.0 | 99.9  | -                    |
| STRA6    | 130.5 | 100.0 | 99.8  | 601186               |
| TACSTD2  | 214.8 | 99.0  | 96.4  | 204870               |
| TCF4     | 132.4 | 100.0 | 99.8  | 613267               |
| TCTN1    | 114.9 | 96.7  | 93.0  | 614173               |

|          |       |       |       |  |
|----------|-------|-------|-------|--|
| TCTN3    | 136.4 | 100.0 | 100.0 | 614815   |
| TDRD7    | 162.3 | 99.9  | 99.1  | 613887   |
| TEAD1    | 157.5 | 100.0 | 99.9  | 108985   |
| TEK      | 191.8 | 100.0 | 100.0 | 617272   |
| TENM3    | 175.3 | 100.0 | 99.7  | 615145   |
| TGFBI    | 134.7 | 99.5  | 94.6  | 121820;602082;122200;608471;607541;608470;121900 |
| TIMM8A   | 58.8  | 98.0  | 90.1  | 304700;311150                                    |
| TIMP3    | 145.1 | 100.0 | 100.0 | 136900   |
| TMCO3    | 151.3 | 100.0 | 99.4  | -  |
| TMEM126A | 125.2 | 96.3  | 84.4  | 612989   |
| TMEM138  | 95.1  | 100.0 | 99.1  | 614465   |
| TMEM216  | 114.4 | 99.9  | 98.1  | 608091   |
| TMEM231  | 106.4 | 100.0 | 99.6  | 615397;614970                                    |
| TMEM237  | 141.7 | 100.0 | 99.9  | 614424   |
| TMEM67   | 96.9  | 99.5  | 95.0  | 607361;216360;610688                             |
| TMEM98   | 140.7 | 99.3  | 97.8  | 615972   |
| TOPORS   | 202.2 | 100.0 | 100.0 | 609923   |
| TPP1     | 146.7 | 100.0 | 100.0 | 204500   |
| TRAF3IP1 | 91.1  | 99.6  | 97.6  | 616629   |
| TREX1    | 244.5 | 100.0 | 100.0 | 192315   |
| TRIM32   | 131.1 | 100.0 | 100.0 | 615988   |
| TRNT1    | 108.4 | 99.5  | 96.5  | 616959   |
| TRPM1    | 151.0 | 100.0 | 99.8  | 613216   |
| TSPAN12  | 164.0 | 100.0 | 99.8  | 613310   |
| TTC8     | 135.1 | 99.6  | 98.1  | 613464;615985                                    |
| TTLL5    | 167.0 | 100.0 | 99.7  | 615860   |
| TUB      | 111.7 | 99.4  | 97.1  | 616188   |
| TUBA3D   | 114.9 | 100.0 | 99.2  | 617928   |
| TUBB3    | 113.8 | 98.3  | 96.9  | 600638   |
| TUBB4B   | 81.1  | 99.9  | 96.9  | 617879   |

|         |       |       |       |                      |
|---------|-------|-------|-------|----------------------|
| TUBGCP4 | 123.4 | 99.2  | 96.4  | 616335               |
| TUBGCP6 | 157.8 | 100.0 | 99.3  | 251270               |
| TULP1   | 134.3 | 100.0 | 99.5  | 600132;613843        |
| TYR     | 164.5 | 100.0 | 100.0 | 103470;203100;606952 |
| TYRP1   | 183.7 | 100.0 | 99.8  | 203290               |
| UBIAD1  | 250.0 | 99.5  | 96.0  | 121800               |
| UNC45B  | 126.0 | 99.3  | 98.0  | 616279               |
| USH1C   | 109.5 | 100.0 | 99.8  | 276904               |
| USH1G   | 161.0 | 99.6  | 97.9  | 606943               |
| USH2A   | 156.7 | 100.0 | 99.8  | 613809;276901        |
| USP45   | 120.3 | 99.6  | 98.1  | 618513               |
| VAX1    | 97.2  | 97.5  | 91.5  | 614402               |
| VCAN    | 180.4 | 100.0 | 100.0 | 143200               |
| VIM     | 121.9 | 99.3  | 97.0  | 116300               |
| VPS13B  | 163.9 | 99.5  | 98.2  | 216550               |
| VSX1    | 69.0  | 84.7  | 80.5  | 148300               |
| VSX2    | 100.3 | 100.0 | 99.3  | 610092;610093        |
| WDPCP   | 122.2 | 98.2  | 94.4  | 615992               |
| WDR19   | 153.1 | 100.0 | 99.4  | 614376;614378;616307 |
| WDR36   | 151.1 | 99.7  | 97.6  | 609887               |
| WFS1    | 194.8 | 100.0 | 99.9  | 614296;116400;222300 |
| WHRN    | 127.3 | 99.8  | 98.1  | 611383               |
| WRN     | 147.8 | 99.9  | 98.8  | 277700               |
| YAP1    | 106.6 | 96.4  | 89.4  | 120433               |
| YME1L1  | 133.4 | 99.0  | 95.2  | 617302               |
| ZEB1    | 170.5 | 100.0 | 99.4  | 613270;609141        |
| ZNF408  | 147.0 | 100.0 | 100.0 | 616468;616469        |
| ZNF423  | 195.9 | 100.0 | 100.0 | 614844               |
| ZNF469  | 154.6 | 100.0 | 100.0 | 229200               |
| ZNF513  | 142.5 | 100.0 | 100.0 | 613617               |

|        |       |       |       |        |
|--------|-------|-------|-------|--------|
| ZNF644 | 178.6 | 100.0 | 100.0 | 614167 |
|--------|-------|-------|-------|--------|

*Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.*

*Median Coverage describes the average number of reads seen across 50 exomes.*

*% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.*

*% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.*

*Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.*

*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*