

Table 1. Specified transcripts for 105 gene symbols analysed by targeted NGS diagnostic testing.

HGNC symbol	RefSeq Transcript ID	HGNC symbol	RefSeq Transcript ID	HGNC symbol	RefSeq Transcript ID
<i>ABCA4</i>	NM_000350.2	<i>PRPF31</i>	NM_015629.3	<i>IMPG2</i>	NM_016247.3
<i>ADAM9</i>	NM_003816.2	<i>PRPF6</i>	NM_012469.3	<i>KCNV2</i>	NM_133497.3
<i>AIP1</i>	NM_014336.3	<i>PRPF8</i>	NM_006445.3	<i>KLHL7</i>	NM_001031710.2
<i>ARL6</i>	NM_032146.3	<i>PRPH2</i>	NM_000322.4	<i>IMPG2</i>	NM_016247.3
<i>BBS1</i>	NM_024649.4	<i>RAX2</i>	NM_032753.3	<i>KCNV2</i>	NM_133497.3
<i>BBS10</i>	NM_024685.3	<i>RBP3</i>	NM_002900.2	<i>KLHL7</i>	NM_001031710.2
<i>BBS12</i>	NM_001178007.1	<i>RD3</i>	NM_183059.2	<i>LCA5</i>	NM_181714.3
<i>BBS2</i>	NM_031885.3	<i>RDH12</i>	NM_152443.2	<i>LRAT</i>	NM_004744.3
<i>BBS4</i>	NM_033028.3	<i>RDH5</i>	NM_001199771.1	<i>LRP5</i>	NM_002335.2
<i>BBS5</i>	NM_152384.2	<i>RGR</i>	NM_002921.3	<i>MERTK</i>	NM_006343.2
<i>BBS7</i>	NM_176824.2	<i>RGS9</i>	NM_001165933.1	<i>MKKS</i>	NM_018848.2
<i>BBS7</i>	NM_018190.3	<i>RGS9</i>	NM_003835.3	<i>MKS1</i>	NM_001165927.1
<i>BBS9</i>	NM_198428.2	<i>RHO</i>	NM_000539.3	<i>MKS1</i>	NM_017777.3
<i>BEST1</i>	NM_004183.3	<i>RIMS1</i>	NM_014989.4	<i>MYO7A</i>	NM_000260.3
<i>CIQTNF5</i>	NM_015645.3	<i>RIMS1</i>	NM_001168407.1	<i>NDP</i>	NM_000266.3
<i>C2orf71</i>	NM_001029883.1	<i>CRB1</i>	NM_201253.2	<i>RIMS1</i>	NM_001168410.1
<i>CA4</i>	NM_000717.3	<i>CRX</i>	NM_000554.4	<i>RLBP1</i>	NM_000326.4
<i>CACNA2D4</i>	NM_172364.4	<i>DFNB31</i>	NM_015404.3	<i>ROM1</i>	NM_000327.3
<i>CDH23</i>	NM_022124.5	<i>DHDDS</i>	NM_024887.2	<i>RP1</i>	NM_006269.1
<i>CDHR1</i>	NM_001171971.1	<i>EFEMP1</i>	NM_001039348.2	<i>RP1L1**</i>	NM_178857.5
<i>CDHR1</i>	NM_033100.2	<i>ELOVL4</i>	NM_022726.3	<i>RP2</i>	NM_006915.2
<i>CEP290*</i>	NM_025114.3	<i>EYS</i>	NM_001142800.1	<i>RP9</i>	NM_203288.1
<i>CERKL</i>	NM_001030311.2	<i>FAM161A</i>	NM_001201543.1	<i>RPE65</i>	NM_000329.2
<i>CHM</i>	NM_000390.2	<i>FSCN2</i>	NM_001077182.2	<i>RPGR***</i>	NM_001034853.1
<i>CLRN1</i>	NM_052995.2	<i>FZD4</i>	NM_012193.3	<i>RPGRIP1</i>	NM_020366.3
<i>CLRN1</i>	NM_001195794.1	<i>GNAT2</i>	NM_005272.3	<i>RS1</i>	NM_000330.3
<i>CNGA1</i>	NM_001142564.1	<i>GPR98</i>	NM_032119.3	<i>SAG</i>	NM_000541.4
<i>CNGA3</i>	NM_001298.2	<i>GUCA1A</i>	NM_000409.3	<i>SEMA4A</i>	NM_022367.3
<i>CNGB1</i>	NM_001297.4	<i>GUCA1B</i>	NM_002098.5	<i>SNRNP200</i>	NM_014014.4
<i>CNGB3</i>	NM_019098.4	<i>GUCY2D</i>	NM_000180.3	<i>SPATA7</i>	NM_018418.4
<i>NR2E3</i>	NM_014249.2	<i>IDH3B</i>	NM_006899.2	<i>TEAD1</i>	NM_021961.5
<i>NRL</i>	NM_006177.3	<i>IDH3B</i>	NM_174855.1	<i>TIMP3</i>	NM_000362.4
<i>OTX2</i>	NM_021728.2	<i>IMPDH1</i>	NM_000883.3	<i>TOPORS</i>	NM_005802.4
<i>PCDH15</i>	NM_001142763.1	<i>IMPG2</i>	NM_016247.3	<i>TRIM32</i>	NM_012210.3
<i>PCDH15</i>	NM_001142769.1	<i>KCNV2</i>	NM_133497.3	<i>TTC8</i>	NM_144596.2
<i>PCDH15</i>	NM_001142771.1	<i>KLHL7</i>	NM_001031710.2	<i>TULP1</i>	NM_003322.3
<i>PCDH15</i>	NM_001142770.1	<i>LCA5</i>	NM_181714.3	<i>UNC119</i>	NM_005148.3
<i>PDE6A</i>	NM_000440.2	<i>LRAT</i>	NM_004744.3	<i>UNC119</i>	NM_054035.2
<i>PDE6B</i>	NM_000283.3	<i>LRP5</i>	NM_002335.2	<i>USH1C</i>	NM_005709.3
<i>PDE6C</i>	NM_006204.3	<i>MERTK</i>	NM_006343.2	<i>USH1C</i>	NM_153676.3
<i>PDE6G</i>	NM_002602.3	<i>MKKS</i>	NM_018848.2	<i>USH1G</i>	NM_173477.2
<i>PITPNM3</i>	NM_031220.3	<i>MKS1</i>	NM_001165927.1	<i>USH2A</i>	NM_206933.2
<i>PRCD</i>	NM_001077620.2	<i>MKS1</i>	NM_017777.3	<i>ZNF513</i>	NM_144631.5
<i>PROM1</i>	NM_006017.2	<i>MYO7A</i>	NM_000260.3		
<i>PRPF3</i>	NM_004698.2	<i>NDP</i>	NM_000266.3		

* Testing of the common intron 26 mutation c.2991+1655A>G in CEP290 is included in this analysis

** Analysis of the coding region of exon 4 of the RP1L1 gene is not included

*** Analysis of the coding region of the final exon (*orf15*) of *RPGR* it is not included *HGNC*, gene symbols approved by the HUGO Gene Nomenclature Committee¹

1. Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Genenames.org: the HGNC resources in 2015. *Nucleic Acids Res* 2015;43:D1079-85.