

# TARGETED GENE PANELS AND REFERENCE SEQUENCE (RefSeq) TRANSCRIPTS BY PHENOTYPE

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## Table of Contents (Click to select)

<b>Alagille Syndrome</b>	<b>2</b>
<b>Chondrodysplasia punctata</b>	<b>2</b>
<b>Combined Pituitary Hormone Deficiency</b>	<b>2</b>
<b>Congenital Generalised Lipodystrophy</b>	<b>2</b>
<b>Congenital Hypothyroidism</b>	<b>2</b>
<b>Early-onset Diabetes and Autoimmunity</b>	<b>3</b>
<b>Endocrine Neoplasia Syndromes</b>	<b>3</b>
<b>Familial Glucocorticoid Deficiency</b>	<b>3</b>
<b>Familial Hyperparathyroidism</b>	<b>3</b>
<b>Familial Hypocalciuric Hypercalcaemia</b>	<b>3</b>
<b>Familial Hyperparathyroidism/hypercalcaemia</b>	<b>4</b>
<b>Familial Hypoparathyroidism</b>	<b>4</b>
<b>Familial Partial Lipodystrophy</b>	<b>4</b>
<b>Familial Porencephaly and HANAC syndrome</b>	<b>4</b>
<b>Familial Tumoral Calcinosis</b>	<b>4</b>
<b>Feingold syndrome</b>	<b>4</b>
<b>Gastrointestinal atresia</b>	<b>5</b>
<b>Generalised Arterial Calcification in Infancy</b>	<b>5</b>
<b>Holoprosencephaly</b>	<b>5</b>
<b>Hyperinsulinism</b>	<b>5</b>
<b>Hypophosphatemic Rickets</b>	<b>6</b>
<b>Isolated Growth Hormone Deficiency</b>	<b>6</b>
<b>Kabuki syndrome</b>	<b>6</b>
<b>Kallmann syndrome</b>	<b>6</b>
<b>Mandibulofacial Dysostosis with Microcephaly</b>	<b>6</b>
<b>Moebius syndrome</b>	<b>6</b>
<b>Monogenic Diabetes of the Young (MODY)</b>	<b>7</b>
<b>Multiple Exostosis</b>	<b>7</b>
<b>Neonatal Diabetes</b>	<b>8</b>
<b>Phaeochromocytoma/Paraganglioma</b>	<b>9</b>
<b>Pontocerebellar Hypoplasia</b>	<b>9</b>
<b>Primary pigmented nodular adrenocortical disease</b>	<b>9</b>
<b>Pseudohypoaldosteronism</b>	<b>9</b>
<b>Spondylocostal Dysostosis</b>	<b>9</b>
<b>Visceral heterotaxy</b>	<b>10</b>

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<u>Alagille Syndrome</u>	Transcript(s)
<i>JAG1</i>	NM_000214
<i>NOTCH2</i>	NM_024408

<u>Chondrodysplasia punctata</u>	Transcript(s)
<i>AGPS</i>	NM_003659
<i>ARSE</i>	NM_000047
<i>EBP</i>	NM_006579
<i>GNPAT</i>	NM_014236
<i>PEX7</i>	NM_000288

<u>Combined Pituitary Hormone Deficiency</u>	Transcript(s)
<i>HESX1</i>	NM_003865
<i>POU1F1</i>	NM_000306
<i>PROP1</i>	NM_006261
<i>LHX3</i>	NM_014564
<i>LHX4</i>	NM_033343

<u>Congenital Generalised Lipodystrophy</u>	Transcript(s)
<i>AGPAT2</i>	NM_006412
<i>BSCL2</i>	NM_032667
<i>CAV1</i>	NM_001753
<i>PPARG</i>	NM_015869
<i>PTRF</i>	NM_012232

<u>Congenital Hypothyroidism</u>	Transcript(s)
<i>FOXE1</i>	NM_004473
<i>NKX2-1</i>	NM_001079668
<i>PAX8</i>	NM_003466
<i>TSHR</i>	NM_000369
<i>TPO</i>	NM_000547
<i>TG</i>	NM_003235
<i>DUOX2</i>	NM_014080
<i>THRA</i>	NM_199334

[Click here to return to page 1](#)

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<b>Early-onset Diabetes and Autoimmunity</b>	<b>Transcript(s)</b>
<i>AIRE</i>	NM_000383
<i>CD274</i>	NM_014143
<i>CTLA4</i>	NM_005214
<i>FOXP3</i>	NM_014009
<i>IL2RA</i>	NM_000417
<i>ITCH</i>	NM_001257138
<i>JAK1</i>	NM_002227
<i>LRBA</i>	NM_001199282
<i>SIRT1</i>	NM_012238
<i>STAT1</i>	NM_007315
<i>STAT3</i>	NM_139276
<i>STAT5B</i>	NM_012448
<i>TNFAIP3</i>	NM_001270508

<b>Endocrine Neoplasia Syndromes</b>	<b>Transcript(s)</b>
<i>AIP</i>	NM_003977
<i>CDKN1B</i>	NM_004064
<i>GPR101</i>	NM_054021.1
<i>MEN1</i>	NM_130799
<i>RET</i>	NM_020975
<i>SOS1</i>	NM_005633

<b>Familial Glucocorticoid Deficiency</b>	<b>Transcript(s)</b>
<i>MC2R</i>	NM_000529
<i>MCM4</i>	NM_005914
<i>MRAP</i>	NM_178817
<i>NNT</i>	NM_012343
<i>STAR</i>	NM_000349

<b>Familial Hyperparathyroidism</b>	<b>Transcript(s)</b>
<i>CASR</i>	NM_000388
<i>CDC73</i>	NM_024529
<i>CDKN1A</i>	NM_078467
<i>CDKN1B</i>	NM_004064
<i>CDKN2B</i>	NM_004936
<i>CDKN2C</i>	NM_001262
<i>MEN1</i>	NM_130799
<i>RET</i>	NM_020975

<b>Familial Hypocalciuric Hypercalcaemia</b>	<b>Transcript(s)</b>
<i>AP2S1</i>	NM_004069
<i>GNA11</i>	NM_002067
<i>CASR</i>	NM_000388

[Click here to return to page 1](#)

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<u>Familial Hyperparathyroidism/hypercalcaemia</u>	Transcript(s)
<i>AP2S1</i>	NM_004069
<i>CASR</i>	NM_000388
<i>CDC73</i>	NM_024529
<i>CDKN1A</i>	NM_078467
<i>CDKN1B</i>	NM_004064
<i>CDKN2B</i>	NM_004936
<i>CDKN2C</i>	NM_001262
<i>GNA11</i>	NM_002067
<i>MEN1</i>	NM_130799
<i>RET</i>	NM_020975

<u>Familial Hypoparathyroidism</u>	Transcript(s)
<i>CASR</i>	NM_000388
<i>GCM2</i>	NM_004752
<i>GNA11</i>	NM_002067
<i>PTH</i>	NM_000315

<u>Familial Partial Lipodystrophy</u>	Transcript(s)
<i>LMNA</i>	NM_170707
<i>PLIN1</i>	NM_002666
<i>PPARG</i>	NM_015869

<u>Familial Porencephaly and HANAC syndrome</u>	Transcript(s)
<i>COL4A1</i>	NM_001845
<i>COL4A2</i>	NM_001846
<i>JAM3</i>	NM_032801.4

<u>Familial Tumoral Calcinosis</u>	Transcript(s)
<i>FGF23</i>	NM_020638
<i>GALNT3</i>	NM_004482
<i>KL</i>	NM_004795
<i>SAMD9</i>	NM_017654

<u>Feingold syndrome</u>	Transcript(s)
<i>MYCN</i>	NM_005378
<i>MIR17HG</i>	NR_027350

[Click here to return to page 1](#)

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<u>Gastrointestinal atresia</u>	Transcript(s)
<i>CCDC11 – HGNC approved name CFAP53</i>	NM_145020
<i>CHD7</i>	NM_017780
<i>FANCB</i>	NM_001018113
<i>FANCC</i>	NM_000136
<i>GLI3</i>	NM_000168
<i>MID1</i>	NM_000381
<i>MYCN</i>	NM_005378
<i>RFX6</i>	NM_173560
<i>SOX2</i>	NM_003106
<i>TTC7A</i>	NM_020458
<i>EFTUD2</i>	NM_004247
<i>FOXF1</i>	NM_001451

<u>Generalised Arterial Calcification in Infancy</u>	Transcript(s)
<i>ABCC6</i>	NM_001171
<i>ENPP1</i>	NM_006208
<i>NT5E</i>	NM_002526.3

<u>Holoprosencephaly</u>	Transcript(s)
<i>GLI2</i>	NM_005270
<i>PTCH1</i>	NM_000264
<i>SHH</i>	NM_000193
<i>SIX3</i>	NM_005413
<i>TGIF1</i>	NM_173208
<i>ZIC2</i>	NM_007129

<u>Hyperinsulinism</u>	Transcript(s)
<i>ABCC8</i>	NM_001287174
<i>AKT2</i>	NM_001626
<i>CACNA1D</i>	NM_000720
<i>GCK</i>	NM_000162
<i>GLUD1</i>	NM_005271
<i>GPC3</i>	NM_001164617
<i>HADH</i>	NM_005327
<i>HNF1A</i>	NM_000545
<i>HNF4A</i>	NM_175914
<i>INSR</i>	NM_000208
<i>KCNJ11</i>	NM_000525
<i>KDM6A</i>	NM_001291415
<i>KMT2D</i>	NM_003482
<i>PMM2</i>	NM_00303
<i>SLC16A1</i>	NM_003051
<i>TRMT10A</i>	NM_001134665

[Click here to return to page 1](#)

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<u>Hypophosphatemic Rickets</u>	Transcript(s)
<i>DMP1</i>	NM_004407
<i>ENPP1</i>	NM_006208
<i>FGF23</i>	NM_020638
<i>PHEX</i>	NM_000444
<i>SLC34A3</i>	NM_080877

<u>Isolated Growth Hormone Deficiency</u>	Transcript(s)
<i>GH1</i>	NM_000515
<i>GHRHR</i>	NM_000823

<u>Kabuki syndrome</u>	Transcript(s)
<i>KDM6A</i>	NM_001291415
<i>KMT2D</i>	NM_003482

<u>Kallmann syndrome</u>	Transcript(s)
<i>KAL1</i>	NM_000216.2
<i>FGFR1</i>	NM_023110.2
<i>FGF8</i>	NM_033163.3
<i>PROKR2</i>	NM_144773.2
<i>PROK2</i>	NM_001126128.1

<u>Mandibulofacial Dysostosis with Microcephaly</u>	Transcript(s)
<i>EFTUD2</i>	NM_004247
<i>SF3B4</i>	NM_005850.4

<u>Moebius syndrome</u>	Transcript(s)
<i>PLXND1</i>	NM_015103.2
<i>REV3L</i>	NM_002912.4

[Click here to return to page 1](#)

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<b>Monogenic Diabetes of the Young (MODY)</b>	<b>Transcript(s)</b>
<b>ABCC8</b>	NM_001287174
<b>APPL1</b>	NM_012096
<b>CEL</b>	NM_001807
<b>CISD2</b>	NM_001008388
<b>DCAF17</b>	NM_025000
<b>DNAJC3</b>	NM_006260
<b>DYRK1B</b>	NM_004714
<b>GATA4</b>	NM_002052
<b>GATA6</b>	NM_005257
<b>GCK</b>	NM_000162
<b>HNF1A</b>	NM_000545
<b>HNF1B</b>	NM_000458
<b>HNF4A</b>	NM_175914
<b>INS</b>	NM_001185098
<b>INSR</b>	NM_000208
<b>KCNJ11</b>	NM_000525
<b>LMNA</b>	NM_170707
<b>mtDNA_3243</b>	NC_012920
<b>NEUROD1</b>	NM_002500
<b>PAX6</b>	NM_001604
<b>PCBD1</b>	NM_000281
<b>PDX1</b>	NM_000209
<b>PIK3R1</b>	NM_181523
<b>PLIN1</b>	NM_002666
<b>POLD1</b>	NM_002691
<b>PPARG</b>	NM_015869
<b>PPP1R15B</b>	NM_032833
<b>RFX6</b>	NM_173560
<b>SLC29A3</b>	NM_018344
<b>TRMT10A</b>	NM_001134665
<b>WFS1</b>	NM_006005
<b>ZBTB20</b>	NM_001164342
<b>ZFP57</b>	NM_001109809

<b>Multiple Exostosis</b>	<b>Transcript(s)</b>
<b>EXT1</b>	NM_000127.2
<b>EXT2</b>	NM_207122

[Click here to return to page 1](#)

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<b>Neonatal Diabetes</b>	<b>Transcript(s)</b>
<b>ABCC8</b>	NM_001287174
<b>AGPAT2</b>	NM_006412
<b>BSCL2</b>	NM_032667
<b>CISD2</b>	NM_001008388
<b>COQ2</b>	NM_015697
<b>COQ9</b>	NM_020312
<b>EIF2S3</b>	NM_001415
<b>EIF2AK3</b>	NM_004836
<b>FOXP3</b>	NM_014009
<b>GATA4</b>	NM_002052
<b>GATA6</b>	NM_005257
<b>GCK</b>	NM_000162
<b>GLIS3</b>	NM_001042413
<b>HNF1B</b>	NM_000458
<b>IER3IP1</b>	NM_016097
<b>IL2RA</b>	NM_000417
<b>INS</b>	NM_001185098
<b>INSR</b>	NM_000208
<b>KCNJ11</b>	NM_000525
<b>LPL</b>	NM_00237
<b>LRBA</b>	NM_006726
<b>MNX1</b>	NM_005515
<b>NEUROD1</b>	NM_002500
<b>NEUROG3</b>	NM_020999
<b>NKX2-2</b>	NM_002509
<b>PDX1</b>	NM_000209
<b>PTF1A</b>	NM_178161
<b>C10orf115</b>	NR_103721
<b>RFX6</b>	NM_173560
<b>SLC19A2</b>	NM_006996
<b>SLC2A2</b>	NM_000340
<b>STAT3</b>	NM_139276
<b>WFS1</b>	NM_006005
<b>ZFP57</b>	NM_001109809

[Click here to return to page 1](#)



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<u>Phaeochromocytoma/Paraganglioma</u>	Transcript(s)
<i>FH</i>	NM_000143
<i>MAX</i>	NM_002382
<i>RET</i>	NM_020975
<i>SDHA</i>	NM_004168
<i>SDHAF2</i>	NM_017841
<i>SDHB</i>	NM_003000
<i>SDHC</i>	NM_003001
<i>SDHD</i>	NM_003002
<i>TMEM127</i>	NM_017849
<i>VHL</i>	NM_000551

<u>Pontocerebellar Hypoplasia</u>	Transcript(s)
<i>AMPD2</i>	NM_004037
<i>CASK</i>	NM_003688
<i>CHMP1A</i>	NM_002768
<i>CLP1</i>	NM_006831
<i>EXOSC3</i>	NM_016042
<i>PCLO</i>	NM_033026
<i>RARS2</i>	NM_020320
<i>SEPSECS</i>	NM_016955
<i>TSEN2</i>	NM_025265
<i>TSEN34</i>	NM_024075
<i>TSEN54</i>	NM_207346
<i>VPS53</i>	NM_001128159
<i>VRK1</i>	NM_003384

<u>Primary pigmented nodular adrenocortical disease</u>	Transcript(s)
<i>PDE11A</i>	NM_016953
<i>PDE8B</i>	NM_003719
<i>PRKAR1A</i>	NM_002734

<u>Pseudohypoaldosteronism</u>	Transcript(s)
<i>CUL3</i>	NM_003590
<i>KLHL3</i>	NM_017415
<i>WNK1</i>	NM_018979
<i>WNK4</i>	NM_032387

<u>Spondylocostal Dysostosis</u>	Transcript(s)
<i>DLL3</i>	NM_016941
<i>HES7</i>	NM_032580
<i>LFNG</i>	NM_001040167
<i>MESP2</i>	NM_001039958
<i>TBX6</i>	NM_004608
<i>RIPPLY2</i>	NM_001009994

[Click here to return to page 1](#)

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<u>Visceral heterotaxy</u>	Transcript(s)
<i>CFC1</i>	NM_032545
<i>ZIC3</i>	NM_003413

[Click here to return to page 1](#)