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

Table of Contents (Click to select)

Alagille Syndrome	Transcript(s)
JAG1	NM_000214
NOTCH2	NM_024408

Chondrodysplasia punctata	Transcript(s)
AGPS	NM_003659
ARSE	NM_000047
EBP	NM_006579
GNPAT	NM_014236
PEX7	NM_000288

Combined Pituitary Hormone Deficiency	Transcript(s)
HESX1	NM_003865
POU1F1	NM_000306
PROP1	NM_006261
LHX3	NM_014564
LHX4	NM_033343

Congenital Generalised Lipodystrophy	Transcript(s)
AGPAT2	NM_006412
BSCL2	NM_032667
CAV1	NM_001753
PPARG	NM_015869
PTRF	NM_012232

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

Click here to return to page 1

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

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

Eye Movement disorders (Congenital fibrosis of the extraocular muscles, Duane radial ray syndrome, Duane retraction syndrome)	Transcript(s)
CHN1	NM_001822
HOXA1	NM_005522
HOXB1	NM_002144
KIF21A	NM_001173464
PHOX2A	NM_005169
ROBO3	NM_022370
SALL4	NM_020436
TUBB3	NM_006086

Familial Glucocorticoid Deficiency	Transcript(s)
MC2R	NM_000529
MCM4	NM_005914
MRAP	NM_178817
NNT	NM_012343
STAR	NM_000349

Click here to return to page 1

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

Familial Hypocalciuric Hypercalcaemia	Transcript(s)
AP2S1	NM_004069
GNA11	NM_002067
CASR	NM_000388

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

Familial Hypoparathyroidism	Transcript(s)
CASR	NM_000388
GCM2	NM_004752
GNA11	NM_002067
PTH	NM_000315

Familial Partial Lipodystrophy	Transcript(s)
LMNA	NM_170707
PLIN1	NM_002666
PPARG	NM_015869

Familial Porencephaly and HANAC syndrome	Transcript(s)
COL4A1	NM_001845
COL4A2	NM_001846
JAM3	NM_032801.4

Click here to return to page 1

Familial Tumoral Calcinosis	Transcript(s)
FGF23	NM_020638
GALNT3	NM_004482
KL	NM_004795
SAMD9	NM_017654

Feingold syndrome	Transcript(s)
MYCN	NM_005378
MIR17HG	NR_027350

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

Generalised Arterial Calcification in Infancy	Transcript(s)
ABCC6	NM_001171
ENPP1	NM_006208
NT5E	NM_002526.3

Holoprosencephaly	Transcript(s)
GLI2	NM_005270
PTCH1	NM_000264
SHH	NM_000193
SIX3	NM_005413
TGIF1	NM_173208
ZIC2	NM_007129

Click here to return to page 1

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

Hypophosphatemic Rickets	Transcript(s)
DMP1	NM_004407
ENPP1	NM_006208
FGF23	NM_020638
PHEX	NM_000444
SLC34A3	NM_080877

Isolated Growth Hormone Deficiency	Transcript(s)
GH1	NM_000515
GHRHR	NM_000823

Kabuki syndrome	Transcript(s)
KDM6A	NM_001291415
KMT2D	NM_003482

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

Mandibulofacial Dysostosis with Microcephaly	Transcript(s)
EFTUD2	NM_004247
SF3B4	NM_005850.4

Click here to return to page 1

Moebius syndrome	Transcript(s)
PLXND1	NM_015103.2
REV3L	NM_002912.4

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

Multiple Exostosis	Transcript(s)
EXT1	NM_000127.2
EXT2	NM_207122

Click here to return to page 1

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

Click here to return to page 1

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

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

Primary pigmented nodular adrenocortical disease	Transcript(s)
PDE11A	NM_016953
PDE8B	NM_003719
PRKAR1A	NM_002734

Pseudohypoaldosteronism	Transcript(s)
CUL3	NM_003590
KLHL3	NM_017415
WNK1	NM_018979
WNK4	NM_032387

Spondylocostal Dysostosis	Transcript(s)
DLL3	NM_016941
HES7	NM_032580
LFNG	NM_001040167
MESP2	NM_001039958
TBX6	NM_004608
RIPPLY2	NM_001009994

Click here to return to page 1

Visceral heterotaxy	Transcript(s)
CFC1	NM_032545
ZIC3	NM_003413

Click here to return to page 1