

Genes

ABCA4

ACAD9

ACADM

ACADS

ACADSB

ACADVL

ACAT1

ACOX1

ACTN4

ADA

ADAMTS2

AFG3L2

AGA

AGL

AGPS

AGRN

AGXT

AHI1

AIPL1

AKR1D1

ALAS2

ALDOA

ALDOB

ALPL

AMACR

AMT

ANK56

ANO5

APOB

APOC2

APOE

APTX

AR

ARG1

ARL13B

ARL6

ARSA

ARSB

ARX

ASL

ASPA
ASPM
ASS1
ATP2A1
ATP7A
ATP7B
ATXN1
ATXN2
ATXN3
ATXN7
B9D1
B9D2
BBS1
BBS10
BBS12
BBS2
BBS4
BBS5
BBS7
BBS9
BCKDHA
BCKDHB
BCS1L
BEST1
BICC1
BRCA1
BRCA2
BRIP1
BSND
BTD
BTK
C3
C5ORF42
CA2
CACNA1A
CAPN3
CAV3
CBS
CC2D2A
CD2AP
CD40LG

COL6A2
COL6A3
COL7A1
COL9A1
COL9A2
COLQ
COQ2
CPS1
CPT1A
CPT2
CRB1
CRTAP
CRX
CSTB
CTNS
CTSD
CTSK
CYP21A2
CYP27A1
CYP4V2
CYP7A1
CYP7B1
DBT
DECR1
DFNB31
DFNB59
DHCR24
DHCR7
DHDDS
DLD
DMD
DMP1
DMPK
DNAJB6
DNM2
DOK7
DPAGT1
DPM2
DPM3
DSP
DYSF

EBP
EDA
EDN3
EDNRB
EGR2
EMD
ENO3
ENPP1
ERCC4
ERCC6
ERCC8
ESCO2
ESPN
ESRRB
ETFA
ETFB
ETFDH
EYS
F11
F5
F8
F9
FAH
FAM126A
FAM20C
FANCA
FANCB
FANCC
FANCD2
FANCE
FANCG
FANCI
FANCL
FANCM
FBP1
FGD4
FHL1
FIG4
FKRP
FKTN
FMR1

FOXG1
FOXN1
FUCA1
FXN
G6PC
G6PC3
GAA
GALC
GALE
GALNS
GALT
GAMT
GATM
GBA
GBE1
GCDH
GCH1
GCSH
GDAP1
GFPT1
GJB2
GJB3
GJB6
GJC2
GLA
GLB1
GLDC
GLE1
GLIS2
GM2A
GNE
GNPAT
GNPTAB
GNPTG
GNS
GPR143
GPR98
GRHPR
GRXCR1
GTDC2
GUCY2D

GUSB
GYS2
HADHA
HADHB
HAL
HBA
HBB
HESX1
HEXA
HEXB
HFE
HGD
HGF
HGSNAT
HIBCH
HLCS
HMGCS2
HPD
HPRT1
HPS1
HSD17B4
HSD3B7
HSPG2
HYLS1
IDH3B
IDS
IDUA
IFT80
IGF1
IGHMBP2
IKBKAP
IL2RG
IMPDH1
IMPG2
INF2
INPP5E
INSR
INVS
IQCB1
ISCU
ISPD

ITGA3
ITGA7
IVD
JAK3
KCNC3
KCNJ13
KCNQ2
KCNQ3
KCNV2
KIF7
L1CAM
LAMA2
LAMB2
LARGE
LBR
LDHA
LDLR
LEPRE1
LHFPL5
LHX3
LIFR
LMNA
LMX1B
LOXHD1
LPL
LRAT
LRP5
LRPPRC
LRTOMT
MAK
MAN2B1
MANBA
MARVELD2
MAT1A
MATN3
MBTPS2
MCCC1
MCCC2
MCEE
MCOLN1
MCPH1

MECP2
MED25
MEFV
MERTK
MFRP
MFSD8
MGAT2
MKKS
MKS1
MMAA
MMAB
MMACHC
MMADHC
MOCS1
MOCS2
MPI
MPZ
MTHFR
MTM1
MTMR2
MUSK
MUT
MVK
MYO15A
MYO3A
MYO6
MYO7A
NAGLU
NAGS
NDRG1
NEB
NEFL
NEK8
NMNAT1
NOTCH2
NPC1
NPC2
NPHP1
NPHP3
NPHP4
NPHS1

NPHS2
NR0B1
NSDHL
NTRK1
NYX
OAT
OCA2
OCRL
OFD1
OPA3
OSTM1
OTC
OTOA
OTOF
OXCT1
PAH
PALB2
PAX3
PAX6
PC
PCCA
PCCB
PCDH15
PDE6A
PDE6B
PDE6C
PDE6G
PDSS1
PDSS2
PDYN
PDZD7
PEX1
PEX10
PEX12
PEX13
PEX14
PEX2
PEX26
PEX3
PEX5
PEX6

PEX7
PFKM
PGM1
PHKA2
PHKG2
PHYH
PKD1
PKD2
PKHD1
PKLR
PLCE1
PLEC
PLEKHG5
PLG
PLOD1
PLP1
PNPO
POLG
POMGNT1
POMT1
POMT2
POR
POU1F1
POU3F4
PPP2R2B
PPT1
PRCD
PRKCG
PRKRA
PROM1
PROP1
PRPS1
PRRT2
PRX
PSAP
PTPRO
PTRF
PTS
PXR1
PYGL
PYGM

RAD51
RAD51C
RAG1
RAG2
RAPSN
RAX
RDH12
RDX
RELN
RGR
RHO
RLBP1
RP2
RPE65
RPGR
RPGRIP1L
SACS
SBDS
SBF2
SC5D
SCN1A
SCN2A
SCN4A
SDCCAG8
SEC63
SEMA4A
SEPN1
SERPINA1
SETX
SGCA
SGCB
SGCD
SGCG
SGSH
SH3TC2
SIX6
SLC22A5
SLC25A13
SLC25A15
SLC25A20
SLC25A22

SLC26A2
SLC26A4
SLC26A5
SLC27A1
SLC2A1
SLC2A2
SLC37A4
SLC45A2
SLC4A11
SLC5A1
SLC6A8
SLC7A7
SLX4
SMN1
SMPD1
SNAI2
SPG11
SPG20
SPG7
STAR
STIL
STRA6
STRC
SUMF1
SUOX
SYNE1
SYNE2
TAF1
TAT
TBC1D24
TBP
TCAP
TCIRG1
TCTN1
TCTN3
TECTA
TFR2
TH
TMC1
TMEM138
TMEM216

TMEM231
TMEM237
TMEM43
TMEM67
TMIE
TMPRSS3
TNNT1
TPP1
TPRN
TRIM32
TRIOBP
TRPC6
TSHR
TTC21B
TTN
TULP1
TYR
TYRP1
UBA1
UBE2T
UBR1
UGT1A1
UMOD
UQCRB
UQCRQ
USH1C
USH1G
USH2A
VDR
WAS
WDR19
WDR62
WFS1
WNT10A
WT1
ZFYVE26
ZNF423