CGAT – Neurodevelopmental Disorder Panel v.1.0 (11/2022)

Panel coverage statistics

This panel contains 2616 RefSeq genes. The names of the genes are in HGNC format that is current as of the versioning date of this panel. There is on average 97.1% coverage of all coding exons at 20x and 98.8% coverage at 10x in analyzable target regions from the panel-based exome backbone (v8 SureSelect). This also includes 10 base pairs of flanking intronic DNA in all available transcripts. Asterisk(*) represent genes in which <20x coverage spanning at least 50% of a given exon is calculated. Please note that sequence variants in certain exons of genes can occur in repetitive DNA or highly homologous genomic architecture inherent in the human genome, and therefore will not be analyzed.

Panel description

This neurodevelopmental disorder (NDD) panel covers a large set of curated genes implicated in autism spectrum disorders (ASD), intellectual disability/developmental delay (ID/DD), and epilepsy. The etiology of these conditions can be diverse with overlapping phenotypes in which Mendelian inheritance patterns have been implicated. Exome sequencing of proband + parent trios is a useful approach for the identification of genes, which when disrupted can lead to pediatric NDD. Monogenic causes of ASD are reported in 10-20% of individuals with no single gene accounting for more than 1% of all ASD cases (PubMed IDs: 20466091, 26503795, 26289574, 31984132, 32429945, 31481879). Further de novo missense and likely gene disrupting variants are overall more frequent in ASD cases vs. controls, (PubMed IDs: 25363768, 30559488). Intellectual disability (ID) leads to an impairment of cognitive development with notable abnormalities of brain structure and/or function. ID is a general symptom of neurologic dysfunction diagnosed in ~1-3% of the worldwide population (PubMed IDs: 26503795, 21124998, 3249945). ID and ASD demonstrate significant comorbidity with damaging de novo variants being enriched in DD (PubMed ID: 30559488). Of note, both genetic and non-genetic factors (e.g. congenital infections, prenatal exposures, trauma) may contribute to the etiology of ID (PubMed ID: 32767738). Epilepsy is heterogeneous and includes monogenic causes as well as complex forms involving multiple genes, modifier genes, and/or environmental factors (PubMed IDs: 22612257, 26122601). Many forms of epilepsy are sporadic, occurring in families with no prior history whereas some cases demonstrate mild forms of epilepsy that can be inherited (PubMed ID: 23934111). Sporadic epilepsy is more commonly caused by dominant, de novo variants in genes that are neuronally-expressed. Epilepsy can also be X-linked or autosomal recessive with de novo pathogenic variants being enriched among genetic forms of epilepsy. Of note, missense variants in ion channels (channelopathies) as well as copy number events have been shown to impart both gain or loss-of-function effects (PubMed ID: 12023309) involving epilepsy-related genes. This panel is designed to detect sequence-based variants in the form of single nucleotide, insertion/deletion, and copy number alterations. Besides the full NDD panel, a single gene or subset of genes can be selected from this list as part of a custom panel.

A2M
A2ML1
AAAS
AARS1
AARS2
AASS
ABAT
ABCA1
ABCA12
ABCA2
ABCB6
ABCB7
ABCC8
ABCC9
*ABCD1**
ABCD3
ABCD4
ABHD12
ABHD16A
ABHD5
ABI2
ABL1
ACAD8
ACAD9
ACADM
ACADS
ACADSB
ACADVL
ACAT1
ACBD6
ACD
ACHE
ACO2
ACOX1
ACP5
ACSF3
ACSL4
ACTA1
ACTB
ACTG1
ACTL6A
ACTL6B
ACTN1
COLEC11
COLGALT1
COMT
COQ2
COQ4
COQ5
COQ6
COQ8A
COQ9
COX10
COX14
COX15
COX20
COX4I2
COX6B1
COX7B
CP
CPA6
CPE
CPEB4
CPLANE1
CPLX1
CPS1
CPT1A
CPT1B
CPT2
CR2
CRADD
CRB2
CRBN
CREB3L1
CREBBP
CRIPT
CRLF1
CRPPA
CRTAP
CSDE1
CSF1R
CSMD1
CSNK2A1
CSNK2B
CSPP1
CSTB
GSS
GSX2
GTF2E2
GTF2H5
GTF2IRD1
GTF3C3
GTPBP2
GTPBP3
GUCY1A1
GUF1
GUSB
GYS1
H1-4
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H3-3A
H3-3B
HACE1
HACL1
HADH
HADHA
HADHB
HAL
HAP1
HARS1
HARS2
HAX1
HCCS
HCFC1
HCN1
HCN2
HCN4
HCRT
HDAC4
HDAC6
HDAC8
HDC
HDLBP
HECTD4
HECW2
HELLS
HEPACAM
HERC1
HESX1
NDUFV1
NDUFV2
*NEB**
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NEDD4L
NEFH
NEFL
NEMF
NEU1
NEUROD2
NEXMIF
NF1
NFASC
NFE2L3
NFIA
NFIB
NFIL3
NFIX
NFKB1
NFKB2
NFU1
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NHP2
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NINL
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NIPAL4
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NKX2-5
NKX6-2
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SCO2
SCP2
SCYL1
SCYL2
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SDHA
SDHAF1
SDHB
SDK2
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SEC24D
SEC31A
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SEMA3E
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SEMA6B
SEMA6D
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SETD1B
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SHANK2
SHANK3
UROS
USB1
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USP27X
USP45
USP7
USP8
USP9X
VAC14
VAMP2
VAPB
VARS1
VARS2
VAX1
VCP
VDR
VEZF1
VILL
VIPAS39
VLDLR
VPS11
VPS13A
VPS13B
VPS13C
VPS13D
VPS33B
VPS37A
VPS41
VPS45
VPS4A
VPS51
VPS53
VRK1
VSIG4
VWA3B
WAC
WARS2
WASF1
WASHC4
WASHC5
WDFY3
WDFY4
WDPCP

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ACY1
ADA
ADA2
ADAM17
ADAM22
ADAMTS10
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ADAT3
ADCY1
ADCY3
ADCY5
ADD3
ADGRG1
ADGRV1
ADK
ADNP
ADPRS
ADRA2B
ADSL
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AFF4
AFG3L2
AGA
AGAP2
AGK
AGL
AGMO
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AGO2
AGO4
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AGRN
AGTPBP1
AGTR2
AHCY
AHDC1
AHI1
AHNAK
AIFM1
AIMP1
AIMP2
AKAP9
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CTC1
CTCF
CTDP1
CTDSPL2
CTNNA2
CTNNB1
CTNND2
CTSA
CTSD
CTSF
CTTNBP2
CTU2
CUL3
CUL4B
CUL7
CUX1
CUX2
CWC27
CWF19L1
CYB5R3
CYC1
CYFIP1
CYFIP2
CYP24A1
CYP27A1
CYP27B1
CYP2U1
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DAG1
DAPP1
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DARS2
DBH
DBT
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DCDC2
DCHS1
DCPS
DCTN1
DCX
DDB1
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DDHD2
DDOST
DDR2
DDX10
DDX3X
HEXA
HEXB
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HHAT
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HIKESHI
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HK1
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HPDL
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HPS3
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HSPD1
HSPG2
HTRA1
HTRA2
HUWE1
HYLS1
IARS1
IARS2
IBA57
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NODAL
NOG
NONO
NOP10
NOTCH1
*NOTCH2**
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NPC1
NPC2
NPHP1
NPHP3
NPRL2
NPRL3
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NR0B2
NR2F1
NR3C2
NR4A2
NR5A1
NRAS
NRROS
NRXN1
NRXN2
NRXN3
NSD1
NSD2
NSDHL
NSMF
NSUN2
NSUN3
NT5C2
NTNG1
NTNG2
NTRK1
NTRK2
NUAK1
NUBPL
NUDCD2
NUP107
NUP155
NUP188
NUP62
*NUS1**
NYX
OBSL1
OCA2
*OCLN**
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SHMT2
SHOC2
SHOX
SHOX2
SHROOM4
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SIGMAR1
SIK1
SIL1
SIM1
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SIN3B
SIX1
SIX3
SIX6
SKI
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WNT7A
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XRCC4
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XYLT2
YAP1
YARS1
YARS2
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YWHAG
YY1
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ZEB2
ZFHX4
ZFP92

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AKT2
AKT3
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ALDOB
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ALG12
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ALX3
ALX4
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INTS1
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INTS6
INTS7
IPO8
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IQSEC1
IQSEC2
IREB2
IRF2BPL
IRX5
ISCA2
ITCH
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ITGA9
ITGB3
ITK
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OFD1
OGDHL
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OPA1
OPA3
OPHN1
OPTN
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ORC1
ORC4
ORC6
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ANXA1
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AP2S1
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AP4B1
AP4E1
AP4M1
AP4S1
AP5Z1
APBB1
APC2
APH1A
APOC3
APP
APTX
AQP2
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ARG1
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ARHGAP44
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ARHGEF9
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ARX
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DRP2
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DSG4
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DUOX2
DUOXA2
DUSP6
DVL1
DYM
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DYNC1I2
DYNC2H1
DYNC2I1
DYNC2I2
DYRK1A
EARS2
EBAG9
EBF3
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ECHS1
ECM1
EDA
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ITPR1
ITSN1
IVD
IYD
JAG1
JAM3
JARID2
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JMJD7
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KAT6B
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KCNT2
KCTD13
PAX8
PBX1
PC
PCBD1
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PCCB
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PCK1
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PCNT
PCYT2
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PDHA1
PDHB
PDHX
PDP1
PDSS1
PDSS2
PDX1
PEPD
PER2
PET100
PEX1
PEX10
PEX11B
PEX12
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PEX7
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SNAP29
SNIP1
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SNRPB
SNRPN
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SNX5
SOBP
SON
SORCS3
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SOS2
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SPECC1L

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AUTS2
AVP
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EN2
ENC1
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KIAA1109
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KRAS
KRIT1
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KRT83
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PHB
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PHF8
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PHKA2
PHKG2
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PHRF1
PHYH
*PI4KA**
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PIDD1
*PIEZO2**
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PIGB
PIGC
PIGG
PIGH
PIGK
PIGL
PIGM
PIGN
PIGO
PIGP
PIGQ
PIGS
PIGT
PIGU
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PIK3C2A
PIK3CA
PIK3R1
PIK3R2
PIK3R5
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SPEN
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SPG21
SPG7
SPINK5
SPOP
SPOUT1
SPR
SPRED1
SPRY1
SPRY2
SPRY4
SPTAN1
SPTBN1
SPTBN2
SPTBN4
SQSTM1
SRCAP
SRD5A3
SRP54
SRPRA
SRPX2
SRSF11
SSR4
ST3GAL3
ST3GAL5
ST7
ST8SIA2
STAC3
STAG1
STAG2
STAMBP
STARD7
STARD9
STEEP1
STIL
STIM1
STRA6
STRADA
STS
STT3A
STT3B
STUB1
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STX1B
STX3
STXBP1

AVPR2
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B3GLCT
B4GALNT1
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BAG3
BAZ2B
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BBS2
BBS4
BBS5
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BBS9
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BCAS3
BCKDHA
BCKDHB
BCKDK
BCL11A
BCL11B
BCOR
BCORL1
BCS1L
BDH1
BDNF
BICD2
BICRA
BIN1
BLM
BLNK
BMP1
BMP4
BMPER
BOD1
BOLA3
BPNT2
BPTF
BRAF
BRAT1
BRD4
BRF1
ENO2
ENPP1
ENTPD1
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ERCC2
ERCC3
ERCC4
ERCC5
ERCC6
ERCC8
ERF
ERI1
ERLIN2
ERMARD
ESCO2
ETFA
ETFB
ETFDH
ETHE1
EVC
EVC2
EXOC2
EXOC6B
EXOC7
EXOSC2
EXOSC3
EXOSC8
EXOSC9
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EXTL3
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EZR
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FAM111B
FAM126A
FAM20C
FAM234B
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LENG8
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LGI3
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LHX3
LHX4
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LIG3
LIG4
LIN7B
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LIPT1
LIPT2
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LMNB2
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LRP1
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LRP4
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PLEC
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PLXNB1
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POLG
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POLR3B
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STXBP5L
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SUMF1
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SYN1
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SYP
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TARS2
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TBC1D2B
TBC1D31
TBC1D7
TBCD
TBCE

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BRIP1	*FANCA*	*LRPPRC*	*PPA2*	*TBCK*
BRPF1	*FANCB*	*LRRC4C*	*PPARG*	*TBK1*
BRSK2	*FANCC*	*LRRIQ3*	*PPFIA1*	*TBL1XR1*
BRWD3	*FANCD2*	*LRRTM4*	*PPIB*	*TBR1*
BSCL2	*FANCE*	*LSS*	*PPIL1*	*TBX1*
BSN	*FANCF*	*LTBP2*	*PPM1D*	*TBX18*
BSND	*FANCG*	*LTBP4*	*PPM1K*	*TBX19*
BTAF1	*FANCI*	*LYRM4*	*PPP1CB*	*TBX4*
BTBD2	*FANCL*	*LYRM7*	*PPP1R12A*	*TBX6*
BTD	*FAR1*	*LYST*	*PPP1R15B*	*TBXA2R*
BTK	*FARS2*	*LZTFL1*	*PPP1R21*	*TBXAS1*
BTN2A2	*FARSB*	*LZTR1*	*PPP1R9B*	*TCF12*
BTRC	*FASN*	*MAB21L1*	*PPP2CA*	*TCF20*
BUB1B	*FASTKD2*	*MAB21L2*	*PPP2R1A*	*TCF3*
C12orf4	*FAT1*	*MACF1*	*PPP2R5D*	*TCF4*
C12orf57	*FAT4*	*MACROD2*	*PPP3CA*	*TCF7L2*
C19orf12	*FBLN1*	*MADD*	*PPRC1*	*TCN2*
C2CD3	*FBLN5*	*MAF*	*PPT1*	*TCOF1*
C2orf69	*FBN1*	*MAG*	*PQBP1*	*TCTN1*
C3	*FBP1*	*MAGED2*	*PREPL*	*TCTN2*
	FBXL3	*MAGEL2*	*PREX1*	*TCTN3*
CA2	*FBXL4*	*MAGI2*	*PRF1*	*TDGF1*
CA5A	*FBXO11*	*MAGT1*	*PRICKLE1*	*TDP2*
CA8	*FBXO28*	*MAN1A1*	*PRICKLE2*	*TECPR2*
CABP2	*FBXO31*	*MAN1B1*	*PRIMA1*	*TECR*
CACNA1A	*FBXW11*	*MAN2B1*	*PRKACA*	*TEK*
CACNA1B	*FBXW4*	*MANBA*	*PRKACB*	*TELO2*
CACNA1C	*FCSK*	*MAOA*	*PRKAR1A*	*TENM3*
CACNA1D	*FDFT1*	*MAP1A*	*PRKCB*	*TERF2*
CACNA1E	*FDXR*	*MAP1B*	*PRKCD*	*TERT*
CACNA1G	*FEZF1*	*MAP2K1*	*PRKD1*	*TET1*
CACNA1H	*FEZF2*	*MAP2K2*	*PRKD2*	*TET2*
CACNA1I	*FGD1*	*MAP3K1*	*PRKG1*	*TET3*
CACNA2D1	*FGF12*	*MAP3K15*	*PRKN*	*TFAP2A*
CACNA2D2	*FGF13*	*MAP3K7*	*PRKRA*	*TFAP2B*
CACNA2D3	*FGF14*	*MAPK1*	*PRMT7*	*TFE3*
CACNB2	*FGF17*	*MAPK10*	*PROC*	*TFG*
CACNB4	*FGF23*	*MAPK8*	*PRODH2*	*TG*
CACNG2	*FGF3*	*MAPK8IP3*	*PROK2*	*TGDS*
CAD	*FGF8*	*MAPRE2*	*PROKR2*	*TGFB1*
CAMK2A	*FGFR1*	*MAPT*	*PROP1*	*TGFB2*
CAMK2B	*FGFR2*	*MARS2*	*PRPS1*	*TGFBR1*
CAMK2G	*FGFR3*	*MASP1*	*PRR12*	*TGFBR2*
CAMTA1	*FH*	*MAST1*	*PRRG4*	*TGIF1*
CANT1	*FIBP*	*MAST3*	*PRRT2*	*TGM1*
CAPN10	*FIG4*	*MAT1A*	*PRSS12*	*TGM5*
CAPRIN1	*FKBP10*	*MATR3*	*PRUNE1*	*TH*
CAPS	*FKRP*	*MBD5*	*PSAP*	*THG1L*

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CASZ1
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CCT5
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PTRHD1
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PUS7
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PYCR2
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MYO7A
MYO9B
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RUNX1T1
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RXYLT1
RYR2
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SATB2
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TTC5
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TTPA
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*TUBB2B**
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TUBGCP6
TUFM
TUSC3
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TWIST2
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TYK2
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UBE2H
UBE2T
UBE3A
UBE3B
UBE3C
UBE4A
UBN2
UBQLN2
UBR1
UBR3
UBR5
UBR7
UBTF
UCHL1
UFC1
UFM1
UGDH
UGP2

COG4	*GRID1*	*NDUFAF3*	*SBDS*	*UGT1A1*
COG5	*GRID2*	*NDUFAF4*	*SBF1*	*ULK1*
COG6	*GRIK2*	*NDUFAF5*	*SC5D*	*ULK4*
COG7	*GRIK5*	*NDUFAF6*	*SCAF4*	*UMPS*
COG8	*GRIN1*	*NDUFAF8*	*SCAMP5*	*UNC13A*
COL11A1	*GRIN2A*	*NDUFB11*	*SCAPER*	*UNC13D*
COL11A2	*GRIN2B*	*NDUFB3*	*SCARB2*	*UNC79*
COL18A1	*GRIN2D*	*NDUFB8*	*SCN10A*	*UNC80*
COL1A1	*GRIP1*	*NDUFB9*	*SCN11A*	*UPB1*
COL1A2	*GRIPAP1*	*NDUFS1*	*SCN1A*	*UPF3B*
COL25A1	*GRM1*	*NDUFS2*	*SCN1B*	*UQCC2*
COL27A1	*GRM7*	*NDUFS3*	*SCN2A*	*UQCC3*
COL3A1	*GRN*	*NDUFS4*	*SCN3A*	*UQCRB*
COL4A1	*GRXCR2*	*NDUFS6*	*SCN5A*	*UQCRC2*
COL4A2	*GSDME*	*NDUFS7*	*SCN8A*	*UQCRQ*
COL7A1	*GSPT2*	*NDUFS8*	*SCN9A*	*UROC1*

The following exons are not analyzed due to multiple homologous sequences in the genome (exon numbering based on RefSeq Select transcript):
NEB exons 82-105, *OCLN* exons 5-9, *TUBB2B* exon 4, *PHC1* exons 8 & 15, *KRT86* exons 3-9, *PIEZO2* exon 4, *IKBKG* exons 3-10, *TMLHE* exons 7-8, *SUZ12* exons 1-9, *KMT2C* exons 7-25, *PI4KA* exons 44-55, *TLK2* exons 1-10, *NUS1* exon 5, *ABCD1* exons 7-10, and *NOTCH2* exons 1-4