

# CONGENITAL HYPOTONIA XPANDED PANEL

## GENE LIST (~1424)

Note: Xpanded panel gene lists are regularly updated/improved. This list should be considered an approximation only as the gene list is subject to change at any time. The patient's test report includes a list of genes evaluated and includes a coverage parameter for each gene.

A2ML1	AAAS	AARS	AASS	ABAT	ABC7	ABCC8	ABCC9
ABCD4	ABHD11	ACACA	ACAD8	ACAD9	ACADL	ACADM	ACADS
ACADSB	ACADVL	ACAT2	AC02	ACOX1	ACP2	ACP6	ACSL4
ACTA1	ACTB	ACTG1	ACTG2	ACY1	ADAMTS2	ADAP2	ADAR
ADAT3	ADCK3	ADCY5	ADCY6	ADGRG1	ADK	ADNP	ADPGK
ADSL	AFF2	AFF4	AFG3L2	AGA	AGK	AGO1	AGRN
AGTPBP1	AHCY	AHDC1	AHI1	AHSG	AIFM1	AIFM3	AIMP1
AK9	AKT1	AKT3	ALDH18A1	ALDH3A2	ALDH5A1	ALDH6A1	ALDH7A1
ALDOA	ALG1	ALG11	ALG12	ALG13	ALG14	ALG2	ALG3
ALG6	ALG8	ALG9	ALMS1	ALPL	AMER1	AMMECR1	AMPD1
AMPD2	AMT	ANK3	ANKRD11	ANOS1	ANTXR2	AP1S1	AP1S2
AP3B2	AP3D1	AP4B1	AP4E1	AP4M1	AP4S1	APC2	APOA1BP
APOPT1	AR	ARFGF2	ARHGAP11B	ARHGAP31	ARID1A	ARID1B	ARID2
ARL13B	ARL3	ARMC9	ARSA	ARV1	ARVCF	ARX	ASAH1
ASCC1	ASCL1	ASH1L	ASNS	ASPA	ASPM	ASXL1	ASXL2
ASXL3	ATAD3A	ATCAY	ATIC	ATL1	ATM	ATP10A	ATP1A2
ATP1A3	ATP2B3	ATP5A1	ATP6V0A2	ATP6V1A	ATP6V1B2	ATP6V1E1	ATP7A
ATP7B	ATP8A2	ATPAF2	ATRX	AUTS2	B3GALNT2	B3GALT6	B3GAT3
B4GALT1	B4GALT7	B4GAT1	B9D1	BAZ1B	BBS1	BCAP31	BCKDHA
BCKDHB	BCL11A	BCL11B	BCOR	BCS1L	BDNF	BICD2	BIN1
BLK	BMP1	BMP2	BMP4	BMPER	BOLA3	BPTF	BRAF
BRAT1	BRPF1	BRWD3	BSCL2	BSND	BTD	BUB1B	BUB3
C10orf2	C11orf73	C12orf4	C12orf57	C12orf65	C21orf2	C5orf42	CA8
CACNA1A	CACNA1C	CACNA1D	CACNA1E	CACNA1G	CACNA1S	CAD	CAMK2B
CAMKMT	CAMTA1	CANT1	CAPN3	CARS2	CASK	CASR	CBL
CC2D2A	CCDC115	CCDC141	CCDC174	CCDC22	CCDC47	CCDC78	CCDC88A
CCNF	CD59	CD96	CDC42	CDH23	CDK13	CDKL5	CDKN1C
CDRT15	CEP104	CEP120	CEP290	CEP41	CEP57	CFL2	CFTR
CHAMP1	CHAT	CHD1	CHD2	CHD3	CHD4	CHD7	CHD8
CHKB	CHMP1A	CHRNA1	CHRNA7	CHRNA1	CHRNA1	CHRNA1	CHRNA1
CHST14	CHST3	CKAP2L	CLCN4	CLCN7	CLCNKA	CLCNKB	CLIP2
CLP1	CLPB	CLTC	CNKSR2	CNTN1	CNTNAP1	CNTNAP2	COA6
COA7	COG1	COG4	COG5	COG6	COG7	COG8	COL11A2
COL12A1	COL13A1	COL18A1	COL1A1	COL1A2	COL2A1	COL3A1	COL4A1
COL4A3BP	COL5A1	COL5A2	COL6A1	COL6A2	COL6A3	COLQ	COMT
COQ4	COQ7	COQ9	COX10	COX14	COX15	COX20	COX6B1
COX8A	CPLX1	CPS1	CPT1A	CPT2	CREBBP	CRYAB	CSNK2A1
CSNK2B	CSPP1	CSTB	CTBP1	CTCF	CTNNA1	CTNNA2	CTNS
CTU2	CUL4B	CWF19L1	CY5R3	CYFIP2	CYP24A1	CYP27B1	CYP2R1
D2HGDH	DAG1	DAO	DARS	DBT	DCC	DCHS1	DCPS
DCX	DDC	DDOST	DDR2	DDX11	DDX3X	DEAF1	DEPDC5

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DGAT1	DGCR14	DGCR2	DGCR6	DGCR8	DGUOK	DHCR7	DHDDS
DHFR	DHTKD1	DHX30	DIAPH1	DIS3L2	DKK1	DLAT	DLD
DLG3	DLG4	DMD	DNA2	DNAJB2	DNAJC12	DNAJC19	DNAJC21
DNM1	DNM1L	DNM2	DNMT3A	DOCK6	DOK7	DOLK	DPAGT1
DPF2	DPH1	DPM1	DPM2	DPM3	DPYD	DSE	DST
DUOX2	DUOXA2	DYNC1H1	DYRK1A	EARS2	EBF3	EBP	ECHS1
EDN1	EDN3	EDNRB	EEF1A2	EFEMP2	EFNB1	EFTUD1	EFTUD2
EGR2	EHMT1	EIF2AK3	EIF2B1	EIF2B2	EIF2B3	EIF2B4	EIF2B5
EIF2S3	ELAC2	ELN	ELP2	EMC1	EMD	EP300	EPCAM
EPG5	EPHA4	ERCC1	ERCC2	ERCC5	ERCC6	ERCC6L2	ERCC8
ERF	ERMARD	ETFA	ETFB	ETFDH	ETHE1	EXOSC3	EXOSC9
EXT2	EXTL3	EZH2	FAM126A	FAM134B	FANCG	FAR1	FARS2
FARSB	FASTKD2	FAT4	FBN1	FBN2	FBP1	FBXL4	FBXO11
FDX1L	FDXR	FEZF1	FGF12	FGF17	FGF8	FGFR1	FGFR2
FGFR3	FGFRL1	FH	FHL1	FIG4	FKBP14	FKRP	FKTN
FLAD1	FLG	FLII	FLNA	FLNB	FLRT3	FLVCR1	FMN2
FMR1	FOXC1	FOXE1	FOXG1	FOXP1	FOXP2	FOXRED1	FRG1
FRRS1L	FUCA1	G6PD	GAA	GABRA1	GABRB1	GABRB2	GABRB3
GABRD	GABRG2	GALC	GALE	GALNS	GAMT	GAN	GARS
GATA1	GATA4	GATA6	GATAD2B	GBA	GBE1	GCDH	GCH1
GCK	GCSH	GDI1	GDNF	GFAP	GFER	GFM1	GFPT1
GJA1	GJB2	GJC2	GK	GLB1	GLDC	GLE1	GLRA1
GLRA4	GLRX5	GLUL	GLYCTK	GM2A	GMPPA	GMPPB	GNA11
GNAI3	GNAO1	GNAS	GNB1	GNB5	GNE	GNPAT	GNPTAB
GORAB	GP1BB	GPAA1	GPC3	GPC4	GPHN	GPT2	GPX4
GRIA3	GRIA4	GRID2	GRIN1	GRIN2A	GRIN2B	GRIN2D	GRM1
GTF2I	GTF2IRD1	GTPBP2	GTPBP3	GUCY2D	GUSB	HACE1	HADH
HADHA	HADHB	HBA1	HBA2	HCFC1	HCN1	HDAC4	HDAC8
HECW2	HEPACAM	HERC1	HERC2	HESX1	HEXA	HFE	HIBCH
HINT1	HIRA	HIST1H1E	HIVEP2	HLCS	HMBS	HMGA2	HMGCS2
HNF1A	HNF4A	HNRNPH2	HNRNPK	HNRNPU	HPD	HPRT1	HRAS
HS6ST1	HSD17B10	HSD17B4	HSPB1	HSPB8	HSPD1	HTRA2	HUWE1
HYLS1	IARS	IBA57	IDH2	IDUA	IER3IP1	IFIH1	IFT140
IFT81	IGF2	IGHMBP2	IKBKAP	IL17RD	IL1RAPL1	INPP5E	INPP5K
INPPL1	INS	INSR	IQCB1	IQSEC2	ISCA2	ISCU	ISPD
ITGA3	ITGA7	ITGB6	ITPA	ITPR1	IYD	JAG1	JMJD1C
KANK1	KANSL1	KAT6A	KAT6B	KBTBD13	KCNA2	KCNAB2	KCNB1
KCNC1	KCNC3	KCNE1L	KCNH1	KCNJ10	KCNJ11	KCNJ2	KCNK9
KCNMA1	KCNQ2	KCNQ3	KCNQ5	KCNT1	KCTD7	KDM1A	KDM5C
KDM6A	KIAA0196	KIAA0556	KIAA0586	KIAA0753	KIAA1109	KIAA1279	KIAA2022
KIDINS220	KIF11	KIF1A	KIF21A	KIF22	KIF2A	KIF5A	KIF5C

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KIF7	KISS1R	KLF13	KLHL40	KLHL41	KLHL7	KMT2A	KMT2B
KMT2C	KMT2D	KPTN	KRAS	L1CAM	L2HGDH	LAGE3	LAMA1
LAMA2	LAMB1	LAMB2	LAMP2	LARGE	LARP7	LARS	LAS1L
LEMD3	LETM1	LGI4	LHX3	LHX4	LIAS	LIFR	LIMK1
LIMS2	LINS	LIPT1	LMBR1	LMBRD1	LMNA	LMOD3	LMX1B
LONP1	LPL	LRP4	LRP5	LRPPRC	LTBP4	LTC4S	LYRM4
LYRM7	LZTR1	MAF	MAG	MAGEL2	MALL	MAN1B1	MAN2B1
MANBA	MAP2K1	MAP2K2	MAP3K7	MAPRE2	MARS2	MBD5	MBOAT7
MBTPS2	MCCC1	MCCC2	MCM3AP	MCOLN1	MECP2	MED12	MED13
MED13L	MED25	MEF2C	MEFV	MEGF10	MEIS2	METTL23	MFF
MFN2	MFSD2A	MFSD8	MGAT2	MICU1	MIPEP	MKS1	MLXIPL
MLYCD	MMAA	MMAB	MMACHC	MMADHC	MOGS	MORC2	MPC1
MPDU1	MPI	MPLKIP	MPV17	MPZ	MRE11A	MRPS16	MRPS22
MRPS34	MSTO1	MTFMT	MTM1	MTMR14	MTO1	MTOR	MTR
MTRR	MTTP	MUSK	MUT	MVK	MYBPC3	MYCN	MYF6
MYH3	MYH7	MYL2	MYO18B	MYO1H	MYO5A	MYO7A	MYO9A
MYOT	MYPN	MYT1L	NAA10	NAA15	NACC1	NADK2	NAGA
NALCN	NANS	NARS2	NAT8L	NBAS	NDN	NDNL2	NDP
NDST1	NDUFA10	NDUFA11	NDUFA12	NDUFA13	NDUFA2	NDUFA4	NDUFA9
NDUFAF2	NDUFAF4	NDUFAF5	NDUFAF6	NDUFB11	NDUFB3	NDUFS1	NDUFS2
NDUFS3	NDUFS4	NDUFS7	NDUFS8	NDUFV1	NDUFV2	NEB	NECAP1
NEDD4L	NEFL	NEK1	NELFA	NEU1	NF1	NFIA	NFIB
NFIX	NGLY1	NHLRC2	NHS	NIPBL	NKX2-1	NKX2-5	NKX6-2
NLRC4	NMNAT1	NONO	NOTCH3	NPAP1	NPC1	NPC2	NPHP1
NPHS1	NPRL3	NR2F1	NR2F2	NR3C2	NRAS	NRXN1	NSD1
NSDHL	NSMF	NSUN2	NTRK2	NUBPL	NUP107	NUP62	NUS1
OCLN	OCRL	OFD1	OGDH	OPA1	OPHN1	ORAI1	OSGEP
OSTM1	OTUD6B	OTX2	P4HB	PACS1	PACS2	PAFAH1B1	PAH
PAM16	PANK2	PAX6	PAX8	PC	PCBD1	PCCA	PCCB
PCDH12	PCDH19	PCL0	PCYT1A	PDE10A	PDE4D	PDE6D	PDHA1
PDHB	PDHX	PDP1	PDSS2	PDX1	PET100	PEX1	PEX10
PEX11B	PEX12	PEX13	PEX14	PEX16	PEX19	PEX2	PEX26
PEX3	PEX5	PEX6	PEX7	PGAP1	PGAP2	PGAP3	PGM3
PHF21A	PHF6	PHGDH	PHIP	PHKA2	PHKB	PHKG2	PHOX2B
PIBF1	PIEZO2	PIGA	PIGG	PIGH	PIGL	PIGN	PIGO
PIGS	PIGT	PIGV	PIGY	PIK3CA	PIK3R1	PIK3R2	PKD1
PKHD1	PLA2G6	PLAA	PLAGL1	PLCB1	PLCB4	PLEKHG2	PLEKHG5
PLOD1	PLP1	PLXND1	PMM2	PMP22	PMPCA	PNKP	PNP
PNPLA2	PNPLA6	PNPLA8	PNPO	PNPT1	POC1A	POGZ	POLG
POLG2	POLR3A	POMGNT1	POMGNT2	POMK	POMT1	POMT2	PON1
PON2	PON3	POU1F1	PPA2	PPARGC1A	PPM1B	PPM1D	PPP1CB

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PPP2R1A	PPP2R5D	PPP3CA	PPT1	PQBP1	PRDM12	PRDM16	PRDM5
PRDX1	PREPL	PRF1	PRKAR1A	PRKCG	PRKD1	PRMT7	PRODH
PROK2	PROKR2	PROP1	PROSC	PRPH	PRPS1	PRRT2	PRUNE
PRX	PSAP	PSAT1	PSMD12	PTCH1	PTCHD1	PTDSS1	PTEN
PTPLA	PTPN11	PTRF	PTRH2	PTS	PUF60	PURA	PUS1
PUS3	PYCR1	PYCR2	PYGM	PYROXD1	QARS	QDPR	RAB11B
RAB18	RAB3GAP1	RAB3GAP2	RAC1	RAD21	RAF1	RAI1	RAP1A
RAP1B	RAPSN	RARB	RARS	RARS2	RASA2	RBM10	REEP1
RELN	RERE	RET	REV3L	RFC2	RFT1	RHOBTB2	RIN2
RIT1	RMND1	RNASEH2A	RNASEH2B	RNASEH2C	RNF125	RORA	RPGRIP1L
RPL10	RPS23	RPS6KA3	RRAS	RREB1	RRM2B	RSPRY1	RUSC2
RYR1	RYR2	SACS	SALL1	SAMD9	SAMHD1	SARS2	SATB2
SBDS	SC5D	SCN1A	SCN1B	SCN2A	SCN3A	SCN4A	SCN8A
SCN9A	SCO1	SCO2	SDHA	SDHAF1	SDHD	SEC24C	SEC24D
SEMA3A	SEMA3E	SEMA5A	SEPN1	SEPSECS	SERAC1	SERPINA1	SERPINH1
SET	SETBP1	SETD1B	SETD2	SETD5	SETX	SFXN4	SGCA
SGCB	SGCD	SGCE	SGCG	SGPL1	SGSH	SH2B1	SH3TC2
SHANK2	SHANK3	SHOG2	SIGMAR1	SIK1	SIL1	SIM1	SIN3A
SIX3	SKI	SLC12A6	SLC13A5	SLC16A2	SLC17A5	SLC18A2	SLC18A3
SLC19A3	SLC1A2	SLC1A3	SLC1A4	SLC22A5	SLC25A1	SLC25A12	SLC25A15
SLC25A19	SLC25A20	SLC25A22	SLC25A24	SLC25A26	SLC25A3	SLC25A4	SLC25A46
SLC26A2	SLC26A4	SLC29A3	SLC2A1	SLC2A10	SLC30A9	SLC33A1	SLC34A1
SLC34A3	SLC35A2	SLC35A3	SLC35C1	SLC39A14	SLC39A8	SLC3A1	SLC45A1
SLC46A1	SLC52A2	SLC52A3	SLC5A5	SLC5A7	SLC6A1	SLC6A19	SLC6A3
SLC6A5	SLC6A8	SLC6A9	SLC7A7	SLC7A9	SLC9A6	SMAD2	SMAD4
SMARCA2	SMARCA4	SMARCB1	SMARCE1	SMC1A	SMC3	SMG9	SMPD1
SMS	SNAP25	SNAP29	SNIP1	SNRPN	SNX14	SON	SOS1
SOS2	SOX10	SOX11	SOX2	SOX5	SOX9	SPARC	SPAST
SPATA5	SPECC1L	SPEG	SPG20	SPR	SPRED1	SPRY4	SPTAN1
SPTBN2	SPTBN4	SRCAP	SRD5A3	SRP54	SSR4	ST3GAL3	ST3GAL5
STAMPB	STAT1	STAT3	STIM1	STRA6	STRADA	STT3A	STT3B
STX11	STX1B	STXBP1	SUCLA2	SUCLG1	SUMF1	SUOX	SURF1
SUV420H1	SYNE1	SYNE2	SYNGAP1	SYNJ1	SYT1	SYT2	SZT2
TAB2	TAC01	TACR3	TAF1	TAF6	TANGO2	TARS2	TAZ
TBC1D20	TBC1D23	TBC1D24	TBCD	TBCE	TBCK	TBL1XR1	TBL2
TBR1	TBX1	TBX4	TCAP	TCF20	TCF4	TCTN1	TCTN2
TCTN3	TDP2	TECPR2	TELO2	TFAP2B	TG	TGFB3	TGFBR1
TGFBR2	TH	THOC2	THRA	THRB	TIMM50	TK2	TKT
TLK2	TMC01	TMEM106B	TMEM138	TMEM165	TMEM199	TMEM216	TMEM231
TMEM237	TMEM5	TMEM67	TMEM70	TMEM8C	TMTC3	TNFRSF11A	TNNT1
TNPO3	TNXB	TOE1	TOR1A	TOR1AIP1	TP53RK	TP63	TP1

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<i>TPK1</i>	<i>TPM2</i>	<i>TPM3</i>	<i>TPO</i>	<i>TPRKB</i>	<i>TRAF7</i>	<i>TRAK1</i>	<i>TRAPPC11</i>
<i>TRAPPC9</i>	<i>TREX1</i>	<i>TRHR</i>	<i>TRIM2</i>	<i>TRIM32</i>	<i>TRIM37</i>	<i>TRIO</i>	<i>TRIP12</i>
<i>TRIP13</i>	<i>TRIP4</i>	<i>TRMT1</i>	<i>TRMT10C</i>	<i>TRMT5</i>	<i>TRMU</i>	<i>TRNT1</i>	<i>TRPS1</i>
<i>TRPV4</i>	<i>TSC1</i>	<i>TSEN15</i>	<i>TSEN2</i>	<i>TSEN54</i>	<i>TSFM</i>	<i>TSHB</i>	<i>TSHR</i>
<i>TTN</i>	<i>TUBA1A</i>	<i>TUBA8</i>	<i>TUBB</i>	<i>TUBB2A</i>	<i>TUBB2B</i>	<i>TUBB3</i>	<i>TUBB4A</i>
<i>TUFM</i>	<i>TXN2</i>	<i>TYMP</i>	<i>UBA1</i>	<i>UBA2</i>	<i>UBA5</i>	<i>UBE2A</i>	<i>UBE3A</i>
<i>UBE3B</i>	<i>UBR1</i>	<i>UBTF</i>	<i>UFD1L</i>	<i>UNC13A</i>	<i>UNC80</i>	<i>UPB1</i>	<i>UPF3B</i>
<i>UQCC2</i>	<i>UQCC3</i>	<i>UQCRQ</i>	<i>USP18</i>	<i>USP7</i>	<i>USP9X</i>	<i>VAC14</i>	<i>VAMP1</i>
<i>VAPB</i>	<i>VARS2</i>	<i>VDR</i>	<i>VIPAS39</i>	<i>VLDLR</i>	<i>VMA21</i>	<i>VPS11</i>	<i>VPS13B</i>
<i>VPS13D</i>	<i>VPS33B</i>	<i>VPS53</i>	<i>VRK1</i>	<i>WAC</i>	<i>WARS2</i>	<i>WDR11</i>	<i>WDR26</i>
<i>WDR4</i>	<i>WDR45</i>	<i>WDR62</i>	<i>WDR73</i>	<i>WDR81</i>	<i>WFS1</i>	<i>WHSC1</i>	<i>WNK1</i>
<i>WNT10A</i>	<i>WWOX</i>	<i>XYLT1</i>	<i>XYLT2</i>	<i>YARS2</i>	<i>YME1L1</i>	<i>YWHAE</i>	<i>YWHAG</i>
<i>YY1</i>	<i>ZBTB18</i>	<i>ZBTB20</i>	<i>ZC4H2</i>	<i>ZDHHC15</i>	<i>ZDHHC9</i>	<i>ZEB2</i>	<i>ZFP57</i>
<i>ZIC1</i>	<i>ZMYND11</i>	<i>ZNF148</i>	<i>ZNF423</i>	<i>ZNF469</i>	<i>ZNF592</i>	<i>ZNHIT3</i>	<i>ZSWIM6</i>