CHOP Rapid Targeted Analysis of Genome – Infant (rTAG-I)

Background: The CHOP Rapid Targeted Analysis of Genome – Infant (rTAG-I) is a next generation sequencing test designed to identify clinically significant copy number and single nucleotide variants that may provide information about diagnosis, management, and/or prognosis for infants.

Targeted gene/cytogenetic syndrome list: The laboratory will update the targeted gene and cytogenetic syndrome list periodically based on review of the literature and review of requests from clinical partners. rTAG-I tests ordered on or after the implementation date of the most current version will include analysis/reporting of the related genes/phenotypes on the updated gene/cytogenetic syndrome list. Please see below for the current version of the rTAG-I targeted gene list.

Please note: During the period of transition to an updated rTAG-I targeted panel list, the version of the rTAG-I utilized for a specific patient's analysis depends on the date that all necessary materials are received. rTAG-I ordered prior to the launch date for a new version may undergo analysis using the newer list if all samples are not received by the transition cutoff date.

Effective version dates for rTAG-I gene and cytogenetic syndrome lists:

Version	Effective Date	Details
v1.1	July 10, 2023	Initial launch of rTAG-I
v1.2	September 3, 2024	Incorporates the following changes to the gene list: • Removal of 14 genes where analysis is not appropriate using short-read NGS technology • Noting 10 genes where analysis may not be complete using short-read NGS technology
v2.0	March 18, 2025	Incorporates the following changes to the gene list: • Addition of 448 genes associated with phenotypes related to infantile disease • Removal of 31 genes not appropriate for inclusion given phenotype is not associated with severe infantile disease • Removal of 26 genes where analysis not appropriate using short-read NGS technology • Noting an additional 34 genes where analysis may not be complete using short-read NGS technology See next page for details.

Table 1: Genes added to rTAG-I v2.0

ABCC2	*ABHD16A*	*ACBD6*	*ACOX1*	*ADAMTS15*	*ADAMTS19*	*ADAMTS3*	*ADAMTSL4*
ADGRE2	*ADGRL1*	*AFG2B*	*AGR2*	*AMFR*	*ANAPC1*	*AP1B1*	*AP1G1*
ARF1	*ARPC4*	*ARPC5*	*ASPH*	*ASPRV1*	*ATF6*	*ATG7*	*ATOH7*
ATP11A	*ATP13A3*	*ATP2B1*	*ATP9A*	*AVIL*	*AXIN1*	*BACH2*	*BCAS3*
BCAT2	*BCL10*	*BET1*	*BFSP1*	*BLOC1S3*	*BLOC1S5*	*BORCS8*	*C1GALT1C1*
C1QA	*C1QB*	*C1QC*	*C2*	*C2orf69*	*C3orf52*	*CAMSAP1*	*CAP2*
CAPN15	*CAPNS1*	*CAPRIN1*	*CARD14*	*CASP2*	*CASP8*	*CAST*	*CBLB*
CCDC32	*CCT3*	*CD36*	*CD8A*	*CDC42BPB*	*CDC45*	*CDCA7*	*CEP250*
CEP295	*CFAP52*	*CFHR5*	*CHASERR*	*CHKA*	*CHRM3*	*CHRNA3*	*CIROP*
CLDN10	*CLDN11*	*CLEC3B*	*CLXN*	*CNGA3*	*CNOT2*	*COL27A1*	*COLEC10*
COX11	*COX16*	*COX6A2*	*CPE*	*CREB3L1*	*CRLS1*	*CRYBB1*	*CRYGB*
CSGALNACT1	*CTNND1*	*CTSC*	*CYB5A*	*CYP19A1*	*DACT1*	*DAGLA*	*DAW1*
DCLRE1B	*DCN*	*DCT*	*DDB1*	*DEF6*	*DHRSX*	*DHX9*	*DMGDH*
DNAH7	*DNAJB4*	*DNMBP*	*DOCK2*	*DOHH*	*DPH2*	*DPH5*	*DPP9*
DPYSL5	*DRG1*	*DTNBP1*	*DTYMK*	*DUSP6*	*DUT*	*DVL3*	*EDEM3*
EDNRA	*EED*	*EIF5A*	*ELF4*	*EMC10*	*ERI1*	*EXOC2*	*EXOC3L2*
EXOSC2	*EXOSC5*	*FAM111B*	*FAM149B1*	*FBXL3*	*FGF16*	*FGF9*	*FIBP*
FILIP1	*FNIP1*	*FOCAD*	*FOSL2*	*FOXI3*	*FRA10AC1*	*FTH1*	*FYCO1*
FZD4	*FZD5*	*FZD6*	*GABBR1*	*GANAB*	*GCNT2*	*GDF2*	*GEMIN5*
GFRA1	*GGPS1*	*GHR*	*GHRHR*	*GHSR*	*GIMAP5*	*GLMN*	*GNAT1*
GNB2	*GPIHBP1*	*GPR156*	*GPRC5B*	*GREB1L*	*GRIA1*	*GSX2*	*GTPBP1*
GZF1	*H4C9*	*HCK*	*HELLS*	*HID1*	*HMOX1*	*HNRNPA2B1*	*HNRNPC*
HOGA1	*HOXB1*	*HOXD10*	*HRURF*	*HS2ST1*	*HSF4*	*IDH3A*	*IDH3B*
IFNAR2	*IFT56*	*IL36RN*	*IL6R*	*IL6ST*	*INTS1*	*INTS11*	*IPO8*
IRF1	*IRX5*	*JAG2*	*JAK1*	*JARID2*	*KAT5*	*KCNJ16*	*KCNN2*
KCNQ1OT1	*KDELR2*	*KDM4B*	*KERA*	*KIF26A*	*KLC2*	*KLK11*	*KPNA3*
KRT4	*KRT6B*	*LACC1*	*LCAT*	*LCP2*	*LEP*	*LGI3*	*LIG3*
LIM2	*LRIG2*	*LRMDA*	*LRRC32*	*LRRC8C*	*LTV1*	*LYN*	*MAB21L2*
MAD1L1	*MAFA*	*MAN2C1*	*MAP3K14*	*MAP3K20*	*MAPKAPK5*	*MBTPS1*	*MCM10*
MCTS1	*MESD*	*METTL5*	*MFRP*	*MINAR2*	*MINPP1*	*MIP*	*MIR140*
MIR184	*MNS1*	*MRPL39*	*MTHFD1*	*MTX2*	*MYL11*	*MYL9*	*MYO1E*
MYOD1	*NAA20*	*NAA60*	*NAPB*	*NCDN*	*NCKAP1L*	*NDUFC2*	*NECTIN4*
NEUROG1	*NLRP1*	*NPR3*	*NSMCE2*	*NSRP1*	*NSUN6*	*NUDT2*	*NUP188*
NUP54	*NXN*	*OTUD5*	*OTUD7A*	*OTULIN*	*PALB2*	*PAX7*	*PCDHGC4*
PDE3A	*PDE6B*	*PDZD8*	*PERCC1*	*PERP*	*PGM2L1*	*PHOX2A*	*PI4K2A*
PI4KB	*PIDD1*	*PIGF*	*PIK3C2A*	*PITX3*	*PKHD1L1*	*PLA2G4A*	*PLAAT3*
PLAG1	*PLAU*	*PLXNA1*	*POC1B*	*POLD3*	*POLR1B*	*POLR3GL*	*POLRMT*
POU4F1	*PPFIBP1*	*PPIL1*	*PPP2R3C*	*PRDX3*	*PRKACA*	*PRKACB*	*PRORP*
PSKH1	*PSMB10*	*PSMB8*	*PTCD3*	*PTPN14*	*PTPRJ*	*PTRHD1*	*PXDN*
RAB34	*RACGAP1*	*RALA*	*RAP1GDS1*	*RAX*	*RBL2*	*RBP4*	*RBSN*

REL	*RFX7*	*RIMS2*	*RNF170*	*RNF220*	*RNF31*	*RNU12*	*RNU4-2*
RNU4ATAC	*ROBO2*	*RORC*	*RPA1*	*RPL13*	*RPL3L*	*RRAGC*	*RRAGD*
RSPO1	*RSPO2*	*SAG*	*SCAF4*	*SCUBE3*	*SEC61A1*	*SEMA4A*	*SERPINB7*
SERPINE1	*SERPINF2*	*SF3B2*	*SFRP4*	*SGMS2*	*SHARPIN*	*SHQ1*	*SIAH1*
SLC11A2	*SLC24A5*	*SLC25A36*	*SLC30A7*	*SLC31A1*	*SLC32A1*	*SLC35B2*	*SLC38A3*
SLC38A8	*SLC4A10*	*SMAD4*	*SMARCAD1*	*SMC5*	*SMG8*	*SNAPC4*	*SNORD118*
SNX10	*SOST*	*SPPL2A*	*SPTBN1*	*SPTSSA*	*SQOR*	*SREBF1*	*SRSF1*
SULT2B1	*SUPT16H*	*TAF4*	*TAF8*	*TAMM41*	*TASP1*	*TBC1D2B*	*TBC1D7*
TBK1	*TBXT*	*TCEAL1*	*TCP1*	*TEFM*	*TENM3*	*TET3*	*TF*
THUMPD1	*TLR8*	*TMEM163*	*TMEM222*	*TMEM63A*	*TMPRSS6*	*TNNC2*	*TNPO2*
TOMM7	*TPP2*	*TRAPPC10*	*TSHZ1*	*TTC12*	*TTC5*	*TUFT1*	*TULP3*
TWIST2	*TXNL4A*	*TYRP1*	*U2AF2*	*UBAP2L*	*UBE3C*	*UBE4A*	*UBIAD1*
UBR7	*UFSP2*	*UNC45B*	*USP27X*	*USP53*	*VIM*	*VKORC1*	*VPS33A*
VPS35L	*VPS41*	*VPS4A*	*VSX2*	*WBP4*	*WDR83OS*	*WLS*	*WNK3*
YARS1	*YIF1B*	*YIPF5*	*YRDC*	*YY1AP1*	*ZBTB11*	*ZBTB7A*	*ZMYM2*
ZMYM3	*ZNF526*	*ZNF668*	*ZNF699*	*ZNF808*	*ZNFX1*	*ZRSR2*	*ZSCAN10*

Table 2: Genes removed from the rTAG-I v2.0 gene list as they are not appropriate for inclusion given phenotype is not associated with severe infantile disease

ACTN1	*ACTN4*	*ANO3*	*ARHGEF10*	*CELSR1*	*CLIC2*	*CYP2C9*	*DNAH8*
DOCK11	*FLG*	*GPX1*	*KLKB1*	*KRT83*	*LEMD3*	*MASP2*	*MC4R*
MEF2A	*OPLAH*	*PRKCG*	*SETX*	*SLC39A5*	*SLC3A1*	*SLC5A2*	*SLCO2A1*
SPG7	*SQSTM1*	*STARD9*	*SUGCT*	*XK*	*XPR1*	*YWHAZ*	

Table 3: Genes removed from rTAG-I v2.0 gene list given gene is not appropriate for analysis utilizing short-read NGS technology

ATXN10	*CBS*	*CEL*	*CFC1*	*CSTB*	*CYP21A2*	*DIP2B*	*DMPK*
FXN	*H3F3A*	*HBA1*	*HBA2*	*HYDIN*	*IKBKG*	*KCNE1*	*NCF1*
NUS1	*SFTPA2*	*SIK1*	*SMN1*	*SMN2*	*STRC*	*TUBB2A*	*TUBB2B*
UQCRFS1	*USP18*						

Table 4: Genes on rTAG-I v2.0 that are not appropriate for complete analysis utilizing short-read NGS technology, will be noted on the gene list with a †

ABCC6	*AFF2*	*ARX*	*ATN1*	*CLCNKA*	*COX10*	*CYP11B1*	*CYP2R1*
DDX11	*DHFR*	*FMR1*	*GBA1*	*GCSH*	*HERC2*	*HOXA13*	*INTU*
ISCA1	*KRT14*	*KRT6A*	*MSTO1*	*NEB*	*NOTCH2*	*OCLN*	*ORC6*
PHC1	*PHOX2B*	*PKD1*	*RANBP2*	*RBM8A*	*SBDS*	*SDHA*	*SLC7A9*
SMPD4	*SORD*	*STAT5B*	*SUZ12*	*TBX20*	*TCF4*	*TLK2*	*TMEM231*
TUBA1A	*TUBG1*	*XYLT1*	*ZIC2*				

rTAG-I v2.0 Targeted Gene List, HGNC-approved gene symbol:

A2ML1, AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA12, ABCA2, ABCA3, ABCB11, ABCB4*, ABCB6, ABCB7, ABCC2, ABCC6*, ABCC8, ABCC9, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD16A, ABHD5, ABL1, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAN, ACAT1, ACBD5, ACBD6, ACD, ACE, ACER3, ACMSD, ACO2, ACOX1, ACP2, ACP5, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTG2, ACTL6B, ACTN2, ACVR1, ACVR2B, ACVRL1, ACY1, ADA, ADA2, ADAM17, ADAM22, ADAM28, ADAMTS10, ADAMTS13, ADAMTS15, ADAMTS17, ADAMTS19, ADAMTS2, ADAMTS3, ADAMTS9, ADAMTSL2, ADAMTSL4, ADAR*, ADARB1, ADAT3, ADCY1, ADCY5, ADCY6, ADD3, ADGRE2, ADGRG1, ADGRG6, ADGRL1, ADGRV1, ADK, ADNP, ADPRS, ADSL, AEBP1, AFF2†, AFF3, AFF4, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGL, AGMO, AGO1, AGPAT2, AGPS, AGR2, AGRN, AGT, AGTPBP1, AGTR1, AGXT, AHCY, AHDC1, AHI1, AHSG, AICDA, AIFM1, AIMP1, AIMP2, AIPL1, AIRE, AK1, AK2, AKR1D1, AKT2, AKT3, ALAS2, ALB*, ALDH18A1, ALDH1A2, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8*, ALG9, ALKBH8, ALMS1, ALOX12B, ALOXE3, ALPK1, ALPK3, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMELX, AMER1, AMFR, AMH, AMHR2, AMMECR1, AMN, AMPD1, AMPD2, AMT, ANAPC1, ANGPT2, ANGPTL6, ANK1, ANK2, ANK3, ANKH, ANKLE2, ANKRD1, ANKRD11, ANKRD17, ANKRD26, ANKS6, ANO10, ANO5, ANO6, ANOS1, ANTXR1*, ANTXR2, AP1B1, AP1G1, AP1S1, AP1S2, AP2M1, AP2S1, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOA2, APOB, APOC2, APPL1, APRT, APTX, AQP2, AR, ARCN1, ARF1, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGDIA, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3*, ARL6, ARL6IP1, ARMC9, ARNT2, ARPC1B, ARPC4, ARPC5, ARSA, ARSB, ARSL, ARV1, ARX†, ASAH1, ASCC1 ASH1L, ASL, ASNS, ASPA, ASPH, ASPM, ASPRV1, ASS1, ASXL1, ASXL2, ASXL3, ATAD1, ATCAY, ATF6, ATG7, ATIC, ATL1, ATM*, ATN1†, ATOH7, ATP11A, ATP13A2, ATP13A3, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B1, ATP2B2, ATP2B3, ATP2C1, ATP5F1A, ATP5F1B, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0A4, ATP6V0C, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP6V1E1, ATP7A, ATP7B, ATP8A2, ATP8B1, ATP9A, ATPAF2, ATR, ATRX, AUH, AUTS2, AVIL, AVPR2, AXIN1, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAAT, BACH2, BAG3, BAP1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCAT2, BCHE, BCKDHA, BCKDHB, BCKDK, BCL10, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BDP1, BET1, BFSP1, BGN, BHLHA9, BICD2, BICRA, BIN1, BLM, BLNK, BLOC1S3, BLOC1S5, BLOC1S6, BLTP1, BLVRA, BMP1, BMP2, BMP4, BMPER, BMPR1B, BMPR2, BOLA3, BORCS8, BPNT2, BPTF, BRAF, BRAT1, BRCA1*, BRCA2*, BRD4, BRF1, BRIP1*, BRPF1, BRWD3, BSCL2, BSND, BTD, BTK, BUB1, BUB1B*, C12orf57, C19orf12, C1GALT1C1, C1QA, C1QB, C1QBP, C1QC, C1R, C1S, C2, C2CD3, C2orf69, C3orf52, C5*, C6, C7, C8A, C8B, CA12, CA2, CA5A, CA8, CABP2, CABP4, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1F, CACNA1G, CACNA1I, CACNA1S, CACNA2D1, CACNA2D2, CACNG2, CAD, CALM1, CALM2, CALM3, CAMK2A, CAMK2B, CAMSAP1, CAMTA1, CANT1, CAP2, CAPN1, CAPN15, CAPN3, CAPNS1, CAPRIN1, CARD11, CARD14, CARMIL2, CARS1, CARS2, CASK, CASP10, CASP14, CASP2, CASP8, CASQ1, CASQ2, CASR, CAST, CASZ1, CAV1, CAV3, CAVIN1, CBL, CBLB, CBLIF, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC115, CCDC134, CCDC174, CCDC22, CCDC32, CCDC39, CCDC40, CCDC47, CCDC50, CCDC65, CCDC78, CCDC8, CCDC88A, CCDC88C, CCM2, CCND2, CCNO, CCNQ, CCT3, CCT5, CD151, CD164, CD19, CD247, CD27, CD320, CD36, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CD96, CDAN1, CDC14A, CDC42, CDC42BPB, CDC45, CDC6, CDCA7, CDH1, CDH11, CDH2, CDH23*, CDH3, CDIN1, CDK10, CDK13, CDK19, CDK5, CDK5RAP2, CDK6, CDK8, CDKL5, CDKN1C, CDON, CDSN, CDT1, CEBPE, CENPE, CENPF, CEP104, CEP120, CEP135, CEP152, CEP164, CEP250, CEP290, CEP295, CEP41, CEP55, CEP57, CEP63, CEP78, CEP83, CEP85L, CERS1, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFAP418, CFAP45, CFAP52, CFAP53, CFB, CFD, CFH, CFHR5, CFL2, CFP, CFTR*, CHAMP1, CHASERR, CHAT, CHD1, CHD2, CHD3, CHD4, CHD7, CHD8, CHKA, CHKB, CHMP1A, CHN1, CHRDL1, CHRM3, CHRNA1, CHRNA2, CHRNA3, CHRNA4, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST8, CHSY1, CHUK, CIB2, CIC, CIITA, CILK1*, CIROP, CISD2, CIT, CITED2, CKAP2L, CLCF1, CLCN1, CLCN4, CLCN5, CLCN7, CLCNKA†, CLCNKB, CLDN1, CLDN10, CLDN11, CLDN14, CLDN16, CLDN19, CLDN9, CLEC3B, CLEC7A, CLIC2, CLIC5, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CLTC, CLUAP1, CLXN, CNGA3, CNGB1, CNGB3, CNKSR2, CNNM2, CNOT1, CNOT2,*

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RECQL4, REEP1, REEP2, REL, RELA, RELB, RELN, REN, RERE, RET, RETREG1, RFT1, RFWD3, RFX5, RFX6, RFX7, RFXANK, RFXAP, RHAG, RHOBTB2, RIMS2, RIN2, RINT1, RIPK1, RIPK4, RIPOR2, RIPPLY2, RIT1, RLBP1, RLIM, RMND1, RMRP, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF13, RNF168, RNF170, RNF216, RNF220, RNF31, RNPC3, RNU12, RNU4-2, RNU4ATAC, RNU7-1, ROBO1, ROBO2, ROBO3, ROGDI, ROR2, RORA, RORB, RORC, RPA1, RPE65, RPGR, RPGRIP1, RPGRIP1L, RPIA, RPL10, RPL11, RPL13, RPL15, RPL19, RPL26, RPL27, RPL35A, RPL3L, RPL5, RPS10, RPS17, RPS19, RPS23, RPS24, RPS26, RPS28, RPS29, RPS6KA3, RPS7, RPSA, RRAGC, RRAGD, RRAS, RRAS2, RREB1, RRM2B*, RS1, RSPH1, RSPH3, RSPH4A, RSPH9, RSPO1, RSPO2, RSPRY1, RSRC1, RTEL1, RTN2*, RTN4IP1, RTTN, RUBCN, RUNX1, RUNX2, RUSC2, RXYLT1, RYR1, RYR2, S1PR2, SACS, SAG, SALL1, SALL4, SAMD9, SAMD9L, SAMHD1*, SAR1B, SARS1, SARS2, SASH3, SASS6, SATB1, SATB2, SBDS†, SBF1, SBF2, SC5D, SCAF4, SCARB2, SCARF2, SCLT1, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN4B, SCN5A, SCN8A, SCN9A, SCNM1, SCNN1A, SCNN1B, SCNN1G, SCO1, SCO2, SCP2, SCUBE3, SCYL1, SCYL2, SDCCAG8, SDHA†, SDHAF1, SDR9C7, SEC23B*, SEC24D, SEC61A1, SECISBP2, SELENOI, SELENON, SEMA4A, SEMA6B, SEPSECS, SERAC1, SERPINA1, SERPINB7, SERPINB8, SERPINC1*, SERPINE1, SERPINF1, SERPINF2, SERPING1, SERPINH1*, SERPINI1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SF3B2, SF3B4, SFRP4, SFTPB, SFTPC, SFXN4, SGCA, SGCB, SGCD, SGCE, SGCG, SGMS2, SGPL1, SGSH, SH2B1, SH2D1A, SH3BP2, SH3PXD2B, SH3TC2, SHANK1, SHANK2, SHANK3, SHARPIN, SHH, SHMT2, SHOC2, SHOX*, SHQ1, SI, SIAH1, SIGMAR1, SIL1, SIM1, SIN3A, SIX1, SIX2, SIX3, SIX5, SIX6, SKI, SKIC2, SKIC3, SLC10A1, SLC10A2, SLC10A7, SLC11A2, SLC12A1, SLC12A2, SLC12A3, SLC12A5*, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC17A8, SLC18A2, SLC18A3, SLC19A2, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC24A1, SLC24A5, SLC25A1, SLC25A10, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A36, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC26A1, SLC26A2, SLC26A3, SLC26A4, SLC27A4, SLC29A3, SLC2A1, SLC2A10, SLC2A2*, SLC30A10, SLC30A7, SLC30A9, SLC31A1, SLC32A1, SLC33A1*, SLC34A1, SLC34A2, SLC34A3, SLC35A1, SLC35A2, SLC35A3, SLC35B2, SLC35C1, SLC35D1, SLC37A4, SLC38A3, SLC38A8, SLC39A13, SLC39A14, SLC39A4, SLC39A7, SLC39A8, SLC40A1, SLC45A1, SLC45A2, SLC46A1, SLC4A1, SLC4A10, SLC4A11, SLC4A4, SLC51B, SLC52A1, SLC52A2, SLC52A3, SLC5A1, SLC5A5, SLC5A6, SLC5A7, SLC6A1, SLC6A17, SLC6A19*, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A2, SLC7A7, SLC7A9*†, SLC9A1, SLC9A3, SLC9A6, SLC9A7, SLFN14, SLITRK6, SLURP1, SLX4, SMAD2, SMAD3, SMAD4, SMAD6, SMAD9, SMARCA2, SMARCA4, SMARCAD1, SMARCAL1, SMARCB1, SMARCC2, SMARCD1, SMARCD2, SMARCE1, SMC1A, SMC3, SMC5, SMCHD1, SMG8, SMG9, SMO, SMOC1, SMPD1, SMPD4†, SMPX, SMS, SNAI2, SNAP25, SNAP29, SNAPC4, SNIP1, SNORD118, SNRPB, SNRPN, SNTA1, SNX10, SNX14, SNX27, SOD1, SON, SORD†, SOS1, SOS2, SOST, SOX10, SOX11, SOX17, SOX18, SOX2, SOX3, SOX4, SOX5, SOX6, SOX9, SP110, SP7, SPAG1, SPARC, SPART, SPAST, SPATA7, SPECC1L, SPEG, SPEN, SPG11, SPI1, SPINK5, SPINT2, SPOP, SPPL2A, SPR, SPRED1, SPRY4, SPTA1, SPTAN1, SPTB, SPTBN1, SPTBN2, SPTBN4, SPTLC1, SPTSSA, SQOR, SRC, SRCAP, SRD5A2, SRD5A3, SREBF1, SRP54, SRPX2, SRRM2, SRSF1, SRY, SSR4, ST14, ST3GAL3, ST3GAL5, STAC3, STAG1, STAG2, STAMBP, STAR, STAT1, STAT2, STAT3, STAT5A, STAT5B†, STEEP1, STIL, STIM1, STING1, STK4, STRA6, STRADA, STS, STT3A, STT3B, STUB1, STX11, STX16, STX1B, STX3, STXBP1, STXBP2, STXBP3, SUCLA2, SUCLG1, SUFU*, SULF1, SULT2B1, SUMF1, SUOX, SUPT16H, SUPT5H, SURF1, SUZ12†, SV2A, SVBP, SYN1, SYNE1, SYNE2, SYNE4, SYNGAP1, SYNJ1, SYP, SYT1, SYT14, SYT2, SZT2, TAB2, TAC3, TACO1, TACR3, TAF1, TAF13, TAF2, TAF4, TAF6, TAF8, TAFAZZIN, TALDO1, TAMM41, TANC2, TANGO2, TAOK1, TAP1, TAP2, TAPT1, TARS2, TASP1, TAT, TBC1D20, TBC1D23, TBC1D24*, TBC1D2B, TBC1D7, TBCD, TBCE, TBCK, TBK1*, TBL1XR1, TBR1, TBX1, TBX15, TBX18, TBX19, TBX2, TBX20†, TBX22, TBX3, TBX4, TBX5, TBX6, TBXAS1, TBXT*, TCAP, TCEAL1, TCF12, TCF20, TCF3, TCF4†, TCIRG1, TCN2, TCOF1, TCP1, TCTN1, TCTN2, TCTN3, TDP2, TECPR2, TECR, TECRL, TECTA, TEFM, TEK, TELO2, TENM3, TENT5A, TERC, TERT, TET2, TET3, TF, TFAM, TFAP2A, TFAP2B, TFE3, TFG, TG, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM1, TGM5, TGM6, TH, THAP1, THAP11, THBD, THOC2, THOC6, THPO, THRA, THRB, THUMPD1, TIAM1, TIMM50, TIMM8A, TIMMDC1, TINF2, TJP2, TK2, TKT, TLK2†, TLL1, TLR8, TMC1, TMCO1, TMEM106B, TMEM107, TMEM126A, TMEM126B, TMEM132E, TMEM138, TMEM147, TMEM163, TMEM165, TMEM199, TMEM216, TMEM222, TMEM231†, TMEM237, TMEM240, TMEM260, TMEM38B, TMEM43, TMEM63A, TMEM67, TMEM70, TMEM94, TMIE, TMPRSS15, TMPRSS3, TMPRSS6, TMTC3, TMX2,*

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rTAG-I v2.0 Targeted Cytogenetic Syndromes list for copy number variants

TAR syndrome deletion* (1q21.1), 1p36 deletion,1p36.33 duplication† (*ATAD3* fusion cluster), 3q29 deletion, Wolf-Hirschhorn (4p16.3), Cri du Chat (5p15), *NIPBL* duplication (5p13.2), Sotos (5q35), Paternal UPD6 imprinting center (6q24.2), Williams (7q11.23), distal 7q11.23 deletion, Russell-Silver imprinting center (7q32.2), 8p inverted duplication deletion, 8p23.3 deletion, Langer-Giedion (8q23.3q24.11), Kleefstra (9q34.3), 10p terminal deletion (10p15.3), Beckwith-Wiedemann imprinting centers (11p15,5), Potocki-Shaffer (11p11.2), Jacobsen (11q24.3), Pallister-Killian (12p), Trisomy 13, UPD14 imprinting center (14q32), Prader-Willi and Angelman critical regions and imprinting center (15q11.2), 15q24 deletion, 16p11.2 proximal deletion and duplication, Miller-Dieker lissencephaly (17p13.3), Charcot-Marie Tooth (17p12), Smith-Magenis (17p11.2), Potocki-Lupski (17p11.2), Neurofibromatosis type 1 (17q11.2), Renal cysts and diabetes (RCAD) (17q12), Koolen-De Vries (17q21.31), Trisomy 18, Tetrasomy 18p, Trisomy 21, 22q11 deletion/duplication (including DiGeorge, Cat Eye, and Emmanuel critical regions), Phelan-McDermid (22q13.3), Xp11.23p11.22 duplication, Pelizaeus-Merzbacher (Xq22.2), *MECP2* duplication (Xq28), Xq28 inth22h1/int22h2 mediated duplication, sex chromosome aneuploidies (X, XXX, XXY)

*Analysis includes variant assessment for relevant autosomal recessive phenotypes associated with this gene/copy number change; autosomal dominant phenotypes are outside the reporting scope for this test

†Analysis of this gene may not be complete using short-read next generation sequencing and additional analyses using alternative methodologies may need to be considered if clinically indicated