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 See next page for details.

Table 1: Genes not appropriate for analysis utilizing short-read NGS technology

ATXN10	*CFC1*	*CSTB*	*CYP21A2*	*DIP2B*	*DMPK*
FXN	*HBA1*	*HBA2*	*HYDIN*	*IKBKG*	*SMN1*
SMN2	*STRC*				

Table 2: Genes not appropriate for complete analysis utilizing short-read NGS technology and on rTAG-I v1.2, will be noted on the gene list with a †

AFF2	*ATN1*	*FMR1*	*HOXA13*	*PHOX2B*	*PKD1*
TCF4	*TNXB*	*XYLT1*	*ZIC2*		

rTAG-I v1.2 Targeted Gene List:

A2ML1, AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA12, ABCA2, ABCA3, ABCB11, ABCB4*, ABCB6, ABCB7, ABCC6, ABCC8, ABCC9, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ABL1, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAN, ACAT1, ACBD5, ACD, ACE, ACER3, ACMSD, ACO2, ACOX1, ACP2, ACP5, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTG2, ACTL6B, ACTN1, ACTN2, ACTN4, ACVR1, ACVR2B, ACVRL1, ACY1, ADA, ADA2, ADAM17, ADAM22, ADAM28, ADAMTS10, ADAMTS13, ADAMTS17, ADAMTS2, ADAMTS9, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY1, ADCY5, ADCY6, ADD3, ADGRG1, ADGRG6, ADGRV1, ADK, ADNP, ADPRS, ADSL, AEBP1, AFF2†, AFF3, AFF4, AFG3L2, AGA, AGK, AGL, AGMO, AGO1, AGPAT2, AGPS, 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, AMH, AMHR2, AMMECR1, AMN, AMPD1, AMPD2, AMT, ANGPT2, ANGPTL6, ANK1, ANK2, ANK3, ANKH, ANKLE2, ANKRD1, ANKRD11, ANKRD17, ANKRD26, ANKS6, ANO10, ANO3, ANO5, ANO6, ANOS1, ANTXR1*, ANTXR2, AP1S1, AP1S2, AP2M1, AP2S1, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOA2, APOB, APOC2, APPL1, APRT, APTX, AQP2, AR, ARCN1, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGDIA, ARHGEF10, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3*, ARL6, ARL6IP1, ARMC9, ARNT2, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATCAY, ATIC, ATL1, ATM*, ATN1†, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B2, ATP2B3, ATP2C1, ATP5F1A, ATP5F1B, ATP5F1D, ATP5F1E, ATP5MK, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0A4, ATP6V0C, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP6V1E1, ATP7A, ATP7B, ATP8A2, ATP8B1, ATPAF2, ATR, ATRX, AUH, AUTS2, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAAT, BAG3, BAP1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCHE, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCORL1, BCS1L, BDP1, BGN, BHLHA9, BICD2, BICRA, BIN1, BLM, BLNK, BLOC1S6, BLTP1, BLVRA, BMP1, BMP2, BMP4, BMPER, BMPR1B, BMPR2, BOLA3, BPNT2, BPTF, BRAF, BRAT1, BRCA1*, BRCA2*, BRD4, BRF1, BRIP1*, BRPF1, BRWD3, BSCL2, BSND, BTD, BTK, BTRC, BUB1, BUB1B, C12orf4, C12orf57, C19orf12, C1QBP, C1R, C1S, C2CD3, 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, CAMTA1, CANT1, CAPN1, CAPN3, CARD11, CARMIL2, CARS1, CARS2, CASK, CASP10, CASP14, CASQ1, CASQ2, CASR, CASZ1, CAV1, CAV3, CAVIN1, CBL, CBLIF, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC115, CCDC134, CCDC174, CCDC22, CCDC39, CCDC40, CCDC47, CCDC50, CCDC65, CCDC78, CCDC8, CCDC88A, CCDC88C, CCM2, CCND2, CCNO, CCNQ, CCT5, CD151, CD164, CD19, CD247, CD27, CD320, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD70, CD79A, CD79B, CD81, CD96, CDAN1, CDC14A, CDC42, CDC6, CDH1, CDH11, CDH2, CDH23, CDH3, CDIN1, CDK10, CDK13, CDK19, CDK5, CDK5RAP2, CDK6, CDK8, CDKL5, CDKN1C, CDON, CDSN, CDT1, CEBPE, CEL, CELSR1, CENPE, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP55, CEP57, CEP63, CEP78, CEP83, CEP85L, CERS1, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFAP418, CFAP45, CFAP53, CFB, CFD, CFH, CFL2, CFP, CFTR, CHAMP1, CHAT, CHD1, CHD2, CHD3, CHD4, CHD7, CHD8, CHKB, CHMP1A, CHN1, CHRDL1, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST8, CHSY1, CHUK, CIB2, CIC, CIITA, CILK1*, CISD2, CIT, CITED2, CKAP2L, CLCF1, CLCN1, CLCN4, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN14, CLDN16, CLDN19, CLDN9, CLEC7A, CLIC2, CLIC5, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CLTC, CLUAP1, CNGB1, CNGB3, CNKSR2, CNNM2, CNOT1, CNOT3, CNPY3, CNTN1, CNTN2, CNTNAP1, CNTNAP2, COA3, COA5, COA6, COA7, COA8, COASY, COCH, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL17A1, COL18A1, COL1A1, COL1A2, COL25A1, COL2A1, COL3A1, COL4A1, COL4A2,*

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VPS13D, VPS33B, VPS45, VPS51, VPS53, VRK1, VWF, WAC, WARS2, WAS, WASF1, WASHC4, WASHC5, WBP11, WDFY3, WDPCP, WDR1, WDR11, WDR19, WDR26, WDR35, WDR37, WDR4, WDR45, WDR45B, WDR62, WDR73, WDR81, WFS1, WHRN, WIPF1, WIPI2, WNK1, WNT1, WNT10A, WNT10B, WNT2B, WNT3, WNT5A, WNT7A, WRAP53, WRN, WWOX, XDH, XIAP, XK, XPA, XPC, XPNPEP3, XPR1, XRCC1, XRCC2, XRCC4, XYLT1†, XYLT2, YAP1, YARS2, YME1L1, YWHAE, YWHAG, YWHAZ, YY1, ZAP70, ZBTB18, ZBTB20, ZBTB24, ZC3H14, ZC4H2, ZDHHC15, ZDHHC9, ZEB1, ZEB2, ZFHX4, ZFP57, ZFPM2, ZFYVE26, ZFYVE27, ZIC1, ZIC2†, ZIC3, ZMIZ1, ZMPSTE24, ZMYND10, ZMYND11, ZNF141, ZNF142, ZNF148, ZNF335, ZNF341, ZNF407, ZNF423, ZNF462, ZNF469, ZNHIT3, ZSWIM6*

rTAG-I v1.2 Targeted Cytogenetic Syndromes list for copy number variants

TAR syndrome deletion* (1q21.1), 1p36 deletion , 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