Test Name	Genes involved	Method
4H syndrome gene panel (2 genes)	POLR3A, POLR3B	CES/WES
Actionable Epilepsy and Channelopathies (34 genes)	SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, KCNQ2, KCNQ3, KCNMA1, KCNA1, KCNA2, KCNJ11, CACNA1H, CACNA1A, KCNT1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, CHRNA4, CHRNB2, CHRNA2, ALDH7A1, CSTB, EPM2A, FOLR1 , GAMT, GATM, EPM2B(NHLRC1), PNPO, POLG, SCN1A, SLC2A1, TSC1, TSC2	CES/WES
Adrenoleukodystrophy (ABCD1) gene analysis (1 genes)	ABCD1	CES/WES
Aicardi-Goutières syndrome gene panel (7 genes)	ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1	CES/WES
Alexander disease (GFAP) gene analysis (1 genes)	GFAP	CES/WES
Alkaptonuria (HGD) gene analysis (1 genes)	HGD	CES/WES
Alzheimer's Disease and Dementia (41 genes)	A2M, ACE, ANG, ALS2, APP, CHCHD10, CHMP2B, CSF1R, CYP2D6, DCTN1, CHMP2B, FUS, GBA, GPX1,GRN, HNRNPA2B1, MAPT, MATR3, MPO, NOTCH3, OPTN, PFN1, PRNP, PRPH, PSEN1, PSEN2, SETX, SQSTM1, SLC52A3, SNCA ,SOD1, SPG11, SQSTM1, TAF15, TARDBP, TBK1, TREM2, TYROBP, TUBA4A, UBQLN2, VAPB, VCP, APOE	CES/WES
Amyotrophic Lateral Sclerosis Spectrum Disorders Gene Curation Expert Panel (42 genes)	ALS2, ANG, ANXA11, **ARPP21***, C9orf72, CCNF, CHCHD10, CHMP2B, CYLD, DAO, DNAJC7, ERBB4, ERLIN1, EWSR1, FIG4, FUS, GLE1, GLT8D1, GRN, HNRNPA2B1, KIF5A, **LGALSL***, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SLC52A2, SLC52A3, SOD1, SPG11, SQSTM1, SS18L1, TAF15, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP.	CES/WES
Arthrogryposis & congenital myasthenic syndrome gene panel (46 genes)	ADCY6, ADGRG6, AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CNTNAP1, COL13A1, COLQ, DNM2, DOK7, DPAGT1, ECEL1, ERBB3, FBN2, GFPT1, GLE1, LAMB2, LRP4, MUSK, MYBPC1, MYH3, MYH8, NALCN, PI4KA, PIEZO2, PIP5K1C, RAPSN, SCN4A, SLC18A3, SLC35A3, SNAP25, SYT2, TNNI2, TNNT3, TPM2, UBA1, VIPAS39, VPS33B, ZBTB4	CES/WES
Ataxia-telangiectasia (ATM) gene analysis (1 genes)	ATM	CES/WES
ATRX gene analysis (1 genes)	ATRX	CES/WES
Benign infantile epilepsy (10 genes)	PRRT2, SCN2A, SCN8A, CHRNA2, KCNQ2, KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A	CES/WES
Brain Malformations Gene Curation Expert Panel (36 genes)	ACTB, AHI1, AKT3, APC2, B4GAT1, CDK5RAP2, CEP152, CNOT1, COL18A1, DCC, DCX, DISP1, DOCK6, FOXA2, GAS1, KIF21A, KIF5C, MACF1, MTOR, NEDD4L, NFIA, PIK3CA, PIK3R2, PROK2, PROKR2, QARS1, RAB18, RAB3GAP1, ROBO3, RXYLT1, SMARCC1, STIL, TUBB2B, TUBG1, VLDLR, WDR62	CES/WES

Brown Vialetto–Van Laere syndrome gene panel (2 genes)	SLC52A2 , SLC52A3	CES/WES
Canavan disease (ASPA) gene analysis (1 genes)	ASPA	CES/WES
Cerebral Palsy Gene Curation Expert Panel (18 genes)	ADD3, AP5Z1, ARL6IP1, ATCAY, B4GALNT1, CACNA1B, CPT1C, CYP2U1, DDHD1, DSTYK, EMC1, EMC1, ENTPD1, FITM2, GSX2, SPART, TUBB2A, ZFYVE27	CES/WES
Charcot-Marie-Tooth and sensory neuropathies gene panel (76 genes)	AARS(AARS1), AIFM1, ATL1, ATL3, CCT5, COX6A1, CTDP1, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, FAM134B(RETREG1), FGD4, FIG4, GAN, GARS(GARS1), GDAP1, GJB1, GNB4, HARS(HARS1), HINT1, HK1, HOXD10, HSPB1, HSPB8, IGHMBP2, INF2, JPH1, KARS(KARS1), KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS(MARS1), MED25, MFN2, MME, MORC2, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, PRDM12, PRPS1, PRX, RAB7A, SBF1, SBF2, SCN11A, SCN9A, SH3TC2, SLC12A6, SLC25A46, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, VCP, WNK1, YARS(YARS1)	CES/WES
Comprehensive Epilepsy gene panel (119 genes)	AARS(AARS1), ADAM22, ALDH7A1, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATRX, CACNA1A, CACNA1H, CACNB4, CAD, CASR, CDKL5, CERS1, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLN1(PPT1), CNPY3, CNTNAP2, CPLX1, CSTB, CUX2, CYFIP2, DENND5A, DOCK7, **DRPLA#**, DYNC1H1, DYRK1A, EEF1A2, EFHC1, EPM2A, EPM2B(NHLRC1),FGF12, FOLR1, GABBR2, GABRA1,GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATAD2B, GATM, GNAO1, GOSR2, GRIN2B, GRIN2D, GUF1, HCN1, HNRNPU, IQSEC2, ITPA, KCNA1, KCNA2, KCNB1, KCNC1, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, LMNB2, MBD5, MDH2, MECP2, MEF2C, NECAP1, NM1(MYO1C), NRXN1, NTRK2, PACS2, PCDH19, PIGA, PIGP, PLCB1, PNKP,PNPO, POLG, PRDM8, PRICKLE(PRICKLE1), QARS(QUARS1), RHOBTB2, SCARB2, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12,SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC9A6, SPTAN1, ST3GAL3, STXBP1, SYNGAP1,SYNJ1, SZT2,TBC1D24, TCF4, TSC1, TSC2, UBA5, UBE2A, WDR45, WWOX, YWHAG, ZEB2	CES/WES
Congenital muscular dystrophy (28 genes)	RXYLT1 ,POMT1 ,B4GAT1 ,CHKB ,COL6A1 ,COL6A2 ,COL6A3 ,COL12A1 ,B3GALNT2 ,DAG1 ,DMD ,EMD ,FKTN ,GMPPB ,ITGA7 ,LAMA2 ,LMNA ,DPM3 ,POMGNT1 ,SELENON ,CRPPA ,FKRP ,POMK ,POMGNT2 ,TCAP ,DPM1 ,DPM2 ,LARGE1	CES/WES
Congenital Myopathies Gene Curation Expert Panel (37 genes)	ACTA1, BIN1, CCDC78, CFL2, COL6A1, COL6A2, COL6A3, DNM2, HACD1, KBTBD13, KLHL40, KLHL41, LAMA2, LMOD3, MAP3K20, MEGF10, MTM1, MYH2, MYH7, MYL1, MYO18B, MYPN, NEB, ORAI1, PYROXD1, RYR1, RYR3, SCN4A, SELENON, SPEG, STAC3, STIM1, TNNT1, TNNT3, TPM2, TPM3, TTN	CES/WES
Cystic megalocephaly (MLC1) gene analysis (1 genes)	MLC1	CES/WES
Dravet syndrome (SCN1A) gene analysis (1 genes)	SCN1A	CES/WES
Duchenne Muscular Dystrophy (DMD) gene sequencing (1 genes)	DMD	CES/WES
Dystonia gene panel (83 genes)	ACTB, AFG3L2, ANO3, ARSA, ATM, ATP1A3, ATP6AP2, ATP7B, AUH, BCAP31, C19orf12, CACNA1A, CACNA1B, CACNA1G, CCDC88C, CIZ1, COASY, COL6A3, DDC, DNAJC13, DNAJC6, DRD2, EEF2, ELOVL4, ELOVL5, FBXO7, FGF14, FTL, GBA, GCDH, GCH1, GNAL, HEXA, HPCA, HTRA2, ITPR1, KCNC3, KCND3, KCNMA1, KCTD17, KMT2B, LRRK2, MAPT, MECP2, MR1, NKX2-1, NPC1, NPC2, PANK2, PARK2(PRKN), PARK7, PDYN, PINK1, PLA2G6, PNKD, PPP2R2B, PRKAG2, PRKCG, PRKRA, PRRT2, SGCE, SLC2A1, SLC30A10, SLC6A3, SMPD1, SNCA, SPR, SPTBN2,	CES/WES

	SYNJ1, TAF1, TGM6, TH, THAP1, TIMM8A, TMEM240, TOR1A, TRPC3, TTBK2, TUBB4A, UCHL1, VPS13A, VPS35, WDR45	
Early Infantile Epileptic Encephalopathy Panel (125 genes)	GPHN, ADARB1, ARFGEF2, FARS2, GNB5, CNPY3, CPLX1, CHD2, WDR45, PLPBP, PRRT2, PNKP, RNF13, EFHC1, CLCN4, SIK1, AARS1, ARX, DNM1, DYRK1A, EEF1A2, PHACTR1, BRAT1, FGF12, ARHGEF15, TRAK1, CDK19, RHOBTB2, ARHGEF9, PLCB1, PACS2, DENND5A, DMXL2, CUX2, SZT2, ETHE1, PIGN, FRRS1L, RALGAPA1, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, PARS2, NECAP1, CYFIP2, KCNH5, GLS, GNAO1, GOT2, SLC13A5, GRIN2A, GRIN2B, GRIN2D, HNRNPU, KCNT2, HCN1, ITPA, KCNA2, KCNB1, KCNQ2, KCNQ3, MDH1, MDH2, MECP2, NEUROD2, NSF, NTRK2, ALDH7A1, ACTL6B, WWOX, PIGA, ADAM22, POLG, DALRD3, PNPO, PPP3CA, TBC1D24, SLC12A5, PCDH19, KCNT1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, ARV1, ST3GAL3, SLC1A2, SLC1A4, SLC2A1, SPTAN1, CDKL5, STXBP1, SLC35A2, UGDH, UGP2, YWHAG, CACNA1A, CACNA1E, CAD, SLC25A22, ALG13, UBA5, AP3B2, NPRL3, SMC1A, DOCK7, CASK, SLC25A12, SYNGAP1, SYNJ1, ST3GAL5, PIGQ, CACNA2D2, TIMM50, PIGB, GABBR2, SETD1A, ZEB2, MAGI2.	CES/WES
Epilepsy Advanced Sequencing and CNV Evaluation (232 genes)	HCN4, PQBP1, ATP6AP2, BCKDK, SLC9A6, DEAF1, RNASEH2A, ARFGEF2, POMT1, RAI1, LIAS, CHD2, WDR45, PRRT2, STX1B, TREX1, PNKP, CHRNA2, CHRNA4, CHRNA7, CHRNB2, EFHC1, RAB39B, TPP1, CLN3, CLN5, COL4A1, CPT2, CRH, PRICKLE1, PRIMA1, CSTB, SIK1, CTSD, KCTD7, VPS13B, ADSL, CYP27A1, DCX, PRICKLE2, AFG2A(SPATA5), ARX, DNM1, DYNC1H1, ABAT, DPYD, DYRK1A, EEF1A2, EMX2, CLN8, FKTN, BRAT1, FGD1, FGFR3, FOXG1, RAB3GAP1, IQSEC2, FLNA, ARHGEF9, VPS13A, PLCB1, SZT2, ATP13A2, FOLR1, ATP6V0A2, PIGN, ALPL, GABRA1, GABRB2, GABRB3, GABRD, MFSD8, GABRG2, NIPBL, ASPM, GAMT, SAMHD1, SETBP1, CNTNAP2, KIFBP, PHGDH, GATM, GFAP, GPC3, SRPX2, GLDC, AMT, GNAO1, TSEN54, KANSL1, SLC13A5, WDR62, GRIA3, GRN, GRIN1, GRIN2A, GRIN2B, SETD2, ANKRD11, POMT2, HSD17B10, HNRNPU, HPRT1, NEXMIF, TUBB2B, HCN1, KCNA1, KCNA2, KCNB1, KCNC1, KCNH2, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, NHLRC1, LAMA2, LBR, MECP2, MEF2C, ASAH1, NDUFA1, ATP1A2, ATP1A3, NOTCH3, ATP2A2, OPHN1, ALDH7A1, PAFAH1B1, PAK3, PAX6, WWOX, TUBA8, PEX7, SERPINI1, PIGA, PLP1, POLG, ATRX, RBFOX1, NDE1, CLN6, PNPO, POMGNT1, PIGV, MBD5, CENPJ, RELN, CPA6, SLC4A10, TBC1D24, PCDH19, KCNT1, PURA, QARS1, SLC25A19, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SHH, STIL, SIX3, SLC2A1, SLC6A1, SLC6A8, SMS, SNAP25, SPTAN1, CDKL5, STXBP1, SYN1, SYP, TBX1, TCF4, NR2F1, TSC1, TSC2, UBE3A, SLC35A2, CACNA1A, CACNA2D1, TUBA1A, CACNB4, FKRP, EPM2A, RNASEH2B, ROGDI, MCPH1, TBL1XR1, SLC25A22, ALG9, ALG13, L2HGDH, PANK2, DNAJC5, SLC19A3, KMT2D, KDM5C, SMC1A, PLA2G6, ADGRV1, RNASEH2C, PHF6, CUL4B, CASR, PIGO, OFD1, LMNB2, DOCK7, CASK, CTSF, SUCLA2, SYNGAP1, SYNJ1, ST3GAL5, CACNA1H, SMC3, LGI1, LARGE1, CACNA2D2, ADGRG1, NRXN1, SCARB2, GOSR2, DEPDC5, ZEB2, MAGI2.	CES/WES
Early-onset juvenile parkinsonism gene panel (13 genes)	ATP13A2, DNAJC6, FBXO7, LRRK2, PARK2(PRKN), PARK7, PINK1, PLA2G6, RAB39B, SLC6A3, SNCA, SYNJ1, VPS13C	CES/WES
Epilepsy Expert Panel (92 gene)	ABAT, ALG13, ARX, ATP1A2, BRAT1, BRAT1, CACNA1H, CACNB4, CASR, CHD2, CHRNA2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CPA6, CPA6, CRH, CSTB, CSTB, CTSD, CTSF, CUX2, DEPDC5, DNAJC5, DNM1, DOCK7, EEF1A2, EPM2A, GABRA1, GABRB3, GABRD, GABRD, GABRG2, GNAO1, GNAO1, GRIN1, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, HCN1, KCNA1, KCNA2, KCNA2, KCNB1, KCNC1, KCNC1, KCNH5, KCNMA1, KCNMA1, KCNQ2, KCNQ2, KCNQ2, KCNT1, KCTD7, LGI1, MFSD8, NECAP1, NHLRC1, NPRL3, OTUD7A, PCDH19, PIGA, PIGO, PLCB1, PNKP, PNPO, PPT1, PRICKLE1, PRICKLE1, PRICKLE2, PRRT2, PURA, ROGDI, RORB, RYR3, SCARB2, SCN1A, SCN1A, SCN1A, SCN1A, SCN1B, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SERPINI1, SLC12A5, SLC25A22, SLC35A2, SPTAN1, ST3GAL5, STX1B, STXBP1, SYNGAP1, SYNJ1, SZT2, TPP1, WDR45, WWOX	CES/WES

Epileptic encephalopathy gene panel (111 genes)	AARS(AARS1),ABAT, ABCC8, ALDH7A1, ALG13, AMT, AP3B2, ARHGEF9, ARV1, ARX, ATP13A2, ATP7A, CACNA1A, CACNA1H, CACNB4, CAD, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLN3, CLN5, CLN6, CLN8, CLTC, CTSD, CTSF, D2HGDH, DENND5A, DHDDS, DNAJC5, DNM1, DOCK7, EEF1A2, FGF12, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GLDC, GNAO1, GRIN2B, GRIN2D, GRN, GUF1, HCN1, HNRNPU, HSD17B4, IDH2, ITPA, KCNA2, KCNB1, KCNJ11, KCNQ2, KCNQ3, KCNT1, KCTD7, MBD5, MDH2, MECP2, MFSD8, NECAP1, NTRK2, NUS1, PC, PCDH19, PEX1, PEX10, PEX12, PEX26, PEX6, PIGA, PLCB1, PNKP, PNPO, PPT1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A22, SLC2A1, SLC35A2, SNAP25, SPTAN1, ST3GAL3, STXBP1, SUOX, SYNJ1, SZT2, TBC1D24, TCF4, TPP1, TSC1, TSC2, UBA5, UBE3A, WWOX, ZEB2	CES/WES
Episodic ataxia gene panel (4 genes)	CACNA1A, CACNB4, KCNA1, SLC1A3	CES/WES
Familial female mental retardation/epilepsy gene panel (31 genes)	ALDH7A1, ARX, ATRX, CASK, CDKL5, CUL4B, FOLR1, GRIA3, HSD17B10, IQSEC2, KCNQ2, KCNQ3, KDM5C, MECP2, MED12, MEF2C, OPHN1, PCDH19, PGK1, PHF6, PNPO, POLG, SCN1A, SCN1B, SCN2A, SCN8A, SLC2A1, SLC9A6, SPTAN1, STXBP1, UBE2A	CES/WES
Familial hemiplegic migraine gene panel (4 genes)	ATP1A2, CACNA1A, PRRT2, SCN1A	CES/WES
Fragile-X Syndrome (1 genes)	FMR1	CES/WES
Friedreich's ataxia (1 genes)	FXN	CES/WES
Fukuyama Congenital Muscular Dystrophy (FKTN) gene sequencing (does not include repeat expansions) (1 genes)	FKTN	CES/WES
Hereditary sensory and autonomic neuropathy (14 genes)	SPTLC2, ATL1, DNMT1, ATL3, WNK1, FAM134B(RETREG1), KIF1A, SCN9A, IKBKAP(ELP1),NTRK1 , NGF , DST , SCN11A ,PRDM12	CES/WES
Idiopathic, generalized epilepsy (10 genes)	CACNA1H, CASR, CACNB4, GABRD, CLCN2, SLC2A1, GABRA1, SLC12A5, EFHC1, GABRA1, **ICK***	CES/WES
Intellectual Disability and Autism Gene Curation Expert Panel (267 genes)	ABCD1, ACSL4, ADNP, ADSL, AFF2, AGTR2, AHDC1, ALDH5A1, ALG12, ALG3, ALG6, ANK2, ANK3, ANKRD11, ANKRD17, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARX, ASH1L, ASXL1, ASXL2, ASXL3, ATP13A2, ATP6AP2, ATP7A, ATRX, AUTS2, **AVPR1A*, BAZ2B*,** BCAP31, BCL11A, BPTF, BRD4, BRSK2, BRWD3, CACNG2, CAMTA1, CASK, CC2D1A, CCDC22, CDH15, CDK13, **CDK16***, CHD3, CHD7, CHD8, CLCN4, CLIC2, CNKSR2, CNOT1, CNOT3**, CNTN4*,** CNTN6, CNTNAP2, CRADD, CRBN, CREBBP, CSNK2A1, CTCF, CTNNB1, CUL3, CUL4B, DDX3X, DHCR7, DKC1, DLG3, DLG4, DLGAP2, DNMT3A, DPP6, DYRK1A, EBF3, EBP, EED, EFTUD2, EHMT1, EIF2S3, EN2, EPB41L1, **EXOC2***, EXOC7, EZH2, FBN1, FGD1, FLNA, FMR1, FOLR1, FOXP1, FOXP2, FRMPD4, FTSJ1, GATAD2B, GDI1, GK, GNAI1, GPC3, GRIA3, GRIK2, H1-4, HCFC1, HDAC8, HECW2, HEPACAM, HNRNPK, HNRNPR, HNRNPU, HOXA1, HPRT1, HUWE1, IDS, IGBP1, IL1RAPL1, IQSEC2, ITPR1, KANSL1, KAT6A, KAT6B, KATNAL2, KCNH1, KDM5B, KDM5C, KDM6B, KIF1A, KMT2B, KMT2C, KMT2E, L1CAM, LAMC3, LAS1L, LINS1, MAGEL2, MAGT1, MAN1B1, MAOA, MBD5, MBTPS2, MCPH1, MED12, MED13, MED13L, MED23, MEF2C, MEIS2, MET, MID1, MSL3, MYT1L, NAA10, NAA15, NBEA, NDP, NEXMIF, NHS, NIPBL, NLGN3, NLGN4X, NONO, NR2F1, NR4A2, NRXN1, NSD1, NSD2, NSDHL, NSUN2, **NTNG1*,** OCRL, OFD1, OPHN1, PACS1, PAK3, PGAP3, PHF21A, PHF6, PHF8, PHIP, PIGL, PIGN, PLP1, POGZ, PORCN, PPM1D, PPP2R1A, PPP2R5D, PQBP1, PRPS1, PRSS12, PTCHD1, PUF60, QRICH1, RAB39B, RAB40AL, RAD21, RAI1, RELN, RERE, RPL10, RPS6KA3, SARS1, SATB2, SET, SETBP1, SETD5, SHANK2, SHANK3, SHROOM4, SIN3A, SLC16A2, SLC2A1, SLC6A4,	CES/WES

	SLC9A9, SMAD4, SMARCA1, SMARCA2, SMARCA4, SMARCC2, SMC1A, SMC3, SMS, SNAP25, SOBP, SON, SOX5, ST3GAL3, STAG1, SYP, TANC2, TAOK1, TBL1XR1, TBR1, TCF7L2, TECR, TELO2, TMLHE, TNRC6B, TRAPPC9, TRIM8, TRIO, TRIP12, TRRAP, TSPAN7, TUSC3, UBE2A, UPF3B, USP27X, USP7, USP9X, VPS13B, WAC, WDFY3, ZC3H14, ZC4H2, ZDHHC15, ZDHHC9, ZEB2, ZMIZ1, ZNF292, ZNF41, ZNF674, ZNF711, ZNF81	
Lissencephaly Sequencing panel (50 genes)	ACTB, ACTG1, ARX, ATP6V0A2, B3GALNT2, B3GNT1(B4GAT1), CDK5, DCX, DYNC1H1, FKRP, FKTN, FTO, GPR56(ADGRG1, ISPD(CRPPA), KATNB1, KIAA1279(KIFBP), KIF2A, KIF5C, LAMA2, LAMB1, LAMC3, LARGE(LARGE1), NDE1, OCLN, PAFAH1B1, PHGDH, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SNAP29, TBC1D20, TMEM5(RXYLT1), TUBA1A, TUBA8, TUBB, TUBB2A, TUBB3, TUBB4A, TUBG1, VIPAS39, VLDLR, VPS33B, WDR62	CES/WES
Limb-Girdle Muscular Dystrophy(39 genes)	DNAJB6 ,POMT1 ,BVES ,DAG1 ,DES ,DMD ,ANO5 ,FKTN ,TRIM32 ,SYNE1 ,SMCHD1 ,GAA ,TOR1AIP1 ,DOK7 ,GMPPB ,POMT2 ,LAMA2 ,LMNA ,PLEC ,POMGNT1 ,LIMS2 ,PNPLA2 ,SELENON ,TRAPPC11 ,SGCA ,SGCB ,SGCD ,SGCG ,TTN ,CRPPA ,FKRP ,CAPN3 ,DYSF ,POMK ,POMGNT2 ,TCAP ,CAV3 ,MYOT ,HNRNPDL	CES/WES
Microcephaly Sequencing (59 genes)	ARFGEF2, ASPM, ASXL3, ATR, ATRX, CASC5(KNL1), CASK, CDC6, CDK5RAP2, CDK6, CDKL5, CDT1, CENPE, CENPJ, CEP135, CEP152, CEP63, DYRK1A, FOXG1, IER3IP1, KIF11, LIG4, MCPH1, MECP2, MED17, MFSD2A, NBN, NDE1, NHEJ1, NIN, ORC1, ORC4, ORC6, PCNT, PHC1, PNKP, RAB18, RAB3GAP1, RAB3GAP2, RAD50, RBBP8, SASS6, SLC25A19, **SLC2A1#,** SLC9A6, STAMBP, STIL, TBC1D20, TCF4, TRAPPC9, TRMT10A, TSEN2, TSEN34, TSEN54, TUBGCP6, UBE3A, WDR62, ZEB2, ZNF335	CES/WES
Muscular Dystrophies and Myopathies Gene Curation Expert Panel (30 genes)	ANO5, BVES, CAPN3, CAV3, COL6A1, COL6A2, COL6A3, CRPPA, DMD, DNAJB6, DYSF, FKRP, FKTN, GMPPB, HNRNPDL, LAMA2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TRAPPC11, TRIM32, TTN.	CES/WES
Myotonic dystrophy (1 genes)	CNBP	CES/WES
Neurofibromatosis (9 genes)	SPRED1 ,NF1 ,NF2 ,CCND1 ,SMARCB1 ,TSC1 ,TSC2 ,VHL ,LZTR1	
Neuronal migration disorders sequencing (53 genes)	ACTB, ACTG1, AKT3, ARFGEF2, ARX, B3GALNT2, B3GNT1(B4GAT1), CCND2, CDK5, COL4A1, COL4A2, DCX, DEPDC5, EMX2, ERMARD, FIG4, FKRP, FKTN, FLNA, GMPPB, GPR56(ADGRG1), ISPD(CRPPA), KATNB1, KIF2A, KIF5C, LAMB1, LARGE(LARGE1), MCPH1, NDE1, OCLN, PAFAH1B1, PI4KA, PIK3CA, PIK3R2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RELN, RTTN, SHH, SIX3, SRPX2, TMEM5(RXYLT1),TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, WDR(DAW1)	CES/WES
Non-Huntington chorea gene Pane (15 genes)	NKX21(NKX2-1), ADCY5, PDE10A, GNAO1, VPS13A, XK, JPH3, SLC20A2, PDGFB, PDGFRB, XPR1, PANK2, FLT(FLT1), CP, ATP7B	CES/WES
Parkinson disease (29 genes)	AFG3L2, ATP13A2, ATP6AP2, C10orf2(TWNK), C19orf12, COASY, CP, CYP27A1, DNAJC5, DNAJC6, FBXO7, GBA, LRRK2, PARK2(PRKN), PARK7, PINK1, PLA2G6, POLG, PRKRA, SLC16A2, SLC20A2, SLC6A3, SMPD1, SNCA, SYNJ1, VPS13A, VPS35, WDR45, XPR1	CES/WES
Pontocerebellar hypoplasia (14 genes)	AMPD2, CASK, CHMP1A, CLP1, EXOSC3, EXOSC8, PCLO, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VPS53, VRK1	CES/WES
Spinocerebellar ataxia (37 genes)	STUB1, NOP56 ,AFG3L2 ,TTBK2 ,BEAN1 ,DAB1 ,EEF2 ,FAT2 ,FGF14 ,PLD3 ,ATXN10 ,PLEKHG4 ,GRM1 ,TMEM240 ,TGM6 ,ITPR1 ,KCNC3 ,KCND3 ,ATXN3 ,MME ,CCDC88C ,PDYN ,PPP2R2B ,PRKCG ,ELOVL5 ,ATXN1 ,ATXN2 ,ATXN7 **,@ATXN8OS** ,SPTBN2 ,ELOVL4 ,TBP ,TRPC3 **,#ATXN8** ,CACNA1A ,CACNA1G ,PUM1	CES/WES

SMA panel (20 genes)	ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DNAJB2, DYNC1H1, FBXO38, GARS(GARS1), HSPB8, IGHMBP2, LAS1L, PLEKHG5, SIGMAR1, SLC5A7, TRPV4, TRIP4, UBA1, VAPB	CES/WES
Clinical Exome gene panel (6161 genes)	Covering 6161 clinically relevant genes	CES
Whole Exome Sequencing	Covering all the coding regions (~24383)	WES
Whole Genome sequencing	Covering Exons, Introns, Noncoding regions	WGS

Specimen Type	Peripheral blood/purified genomic DNA/chorionic villus sample (CVS)/amniotic fluid/ Dried Blood Spots (FTA Cards)/ Product of Conception (POC)
Container	EDTA anticoagulated peripheral blood; DNA in sealed eppendorf tube; amniotic fluid in a sterile falcon tube/cultured cells; CVS in a sterile 15ml falcon tube with RPMI1640+10% FBS+ 1% antibiotic. For Dried Blood Spots (FTA Cards) - Whatman FTA card in sealable plastic bag/Envelope cover (Add desiccant packets if available). For Product of conception (POC) - Wide mouth screw capped containers with plain RPMI, or sterile saline may be used for transportation of the specimen.

*** Genes which are not covered in CES but present in WES.**

 # Genes which are not covered in both CES and WES.

@ Genes which are not covered in WES but present in CES