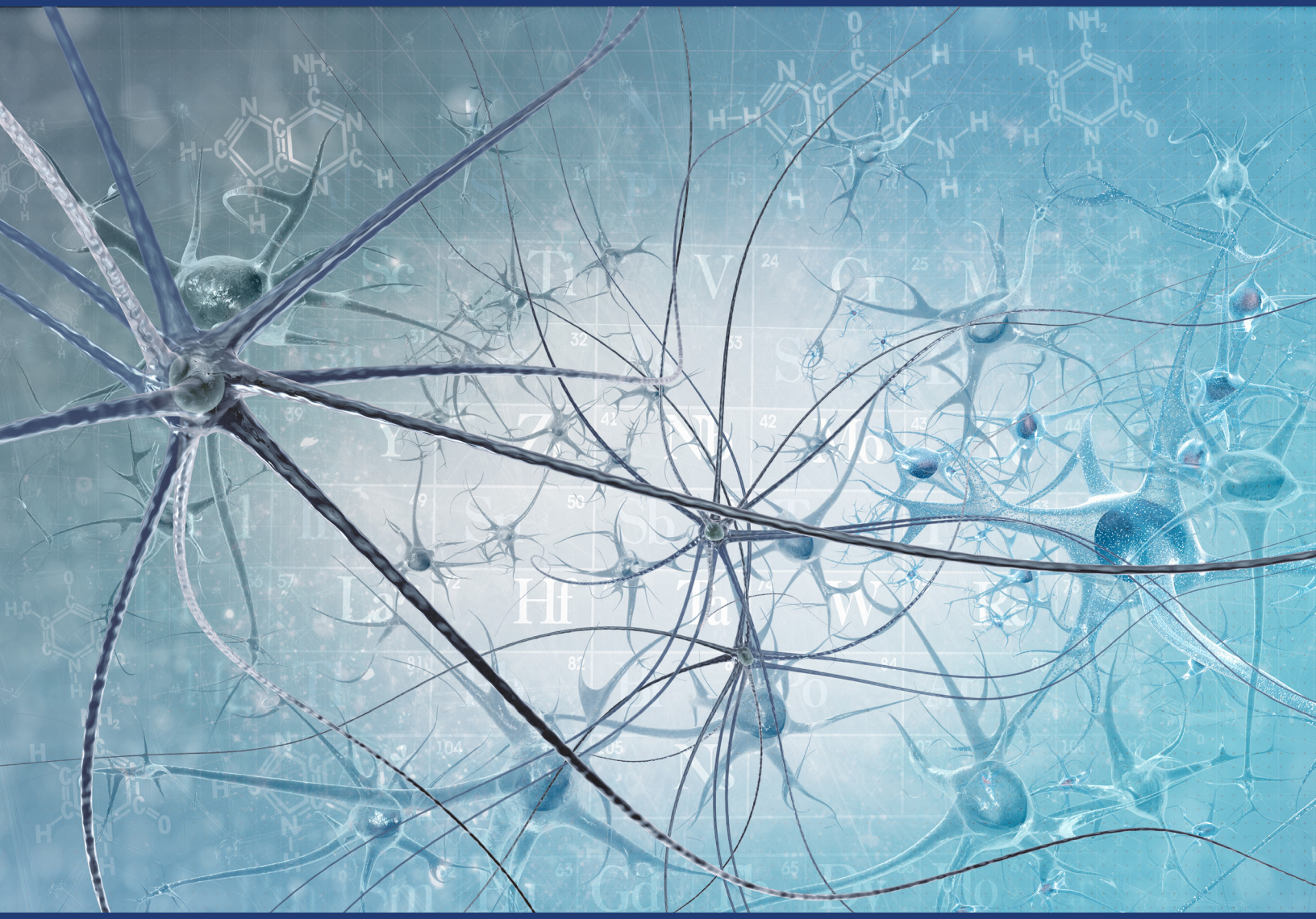
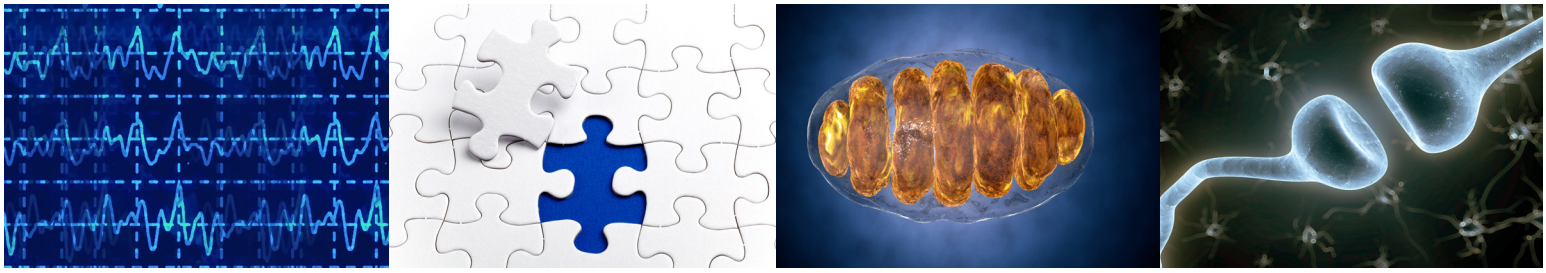
Greenwood Diagnostic Labs



NEUROLOGY

GENETIC TESTING



CONTACT A LABORATORY GENETIC COUNSELOR

LABGC@GGC.ORG
+1 (800) 473-9411
MONDAY-FRIDAY

SPECIMEN REQUIREMENTS

Please visit ggc.org/test-finder for specimen requirements for each test.

REQUISITION FORMS

Please visit ggc.org/labrequisition to access all requisition forms.

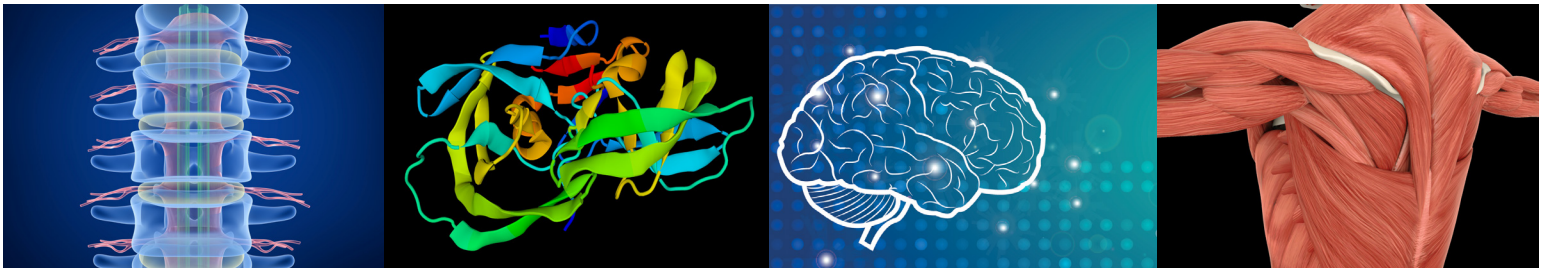
SHIPPING ADDRESS

Ship all specimens to:
106 Gregor Mendel Circle
Greenwood, SC 29646

BLOOD AND SALIVA SAMPLE KITS

Available by request
LABGC@GGC.ORG
+1 (800) 473-9411

Greenwood
Diagnostic Labs



The Greenwood Diagnostic Laboratories collectively offer extensive options of cytogenetic, biochemical, and molecular testing for a wide range of neurological disorders with underlying genetic etiologies. This subset of our complete test menu includes chromosomal whole genome microarray, next generation sequencing panels, and lysosomal enzyme testing as well as more traditional tests such as single gene testing and chromosome analysis among others. A diverse and heterogeneous group of neurological disorders with a genetic etiology can include the following conditions:

Ataxia
Autism
Congenital brain malformations
Developmental delay
Epilepsy
Intellectual disability
Muscular dystrophies
Movement disorders
Myotonic dystrophy
Neuropathies

While many of these disorders have multifactorial and complex etiologies, a proportion will have a specific genetic cause. A number of professional organizations, such as the American Academy of Neurology, the American Epilepsy Society, and the Child Neurology Society, have published guidelines outlining the application and clinical utility of genetic testing for various neurological conditions

Determining the exact genetic etiology for these phenotypes can facilitate appropriate medical management for the primary diagnosis as well as any secondary health issues associated with the syndrome. There are also specific examples, such as mutations in the *SCN1A* gene, where certain medications and therapies are contraindicated as they are known to worsen the clinical symptoms or cause adverse outcomes. Identification of a specific genetic change can have a significant impact on each patient's treatment and prognosis.

Given the overlapping clinical presentations for many of these conditions, multi-gene sequencing panels or tiered genetic testing is the most efficient option for determining the specific etiology. In addition, the growing number of number of targeted and FDA-approved therapies for specific neurological genetic diagnoses such as Duchenne muscular dystrophy (DMD) or spinal muscular atrophy (SMA) further validate the importance of establishing a genetic diagnosis.

In addition to our thorough test menu, Greenwood Diagnostic Laboratories offers parental testing at no charge to assist the appropriate classification of all copy number and sequencing variants as part of our mission of giving greater care to the patients and families we serve.

Molecular Testing

NGS Panels	CPT Code	Price
Charcot-Marie-Tooth Hereditary Neuropathy Sequencing Panel	81448	$3,000
Congenital Contractures Sequencing Panel	81479	$3,000
Early Infantile Epileptic Encephalopathy Sequencing Panel	81402x2, 81405x2, 81406x2, 81407, 81479	$3,500
Epilepsy/Seizure Sequencing Panel	81419	$3,500
Hereditary Spastic Paraplegia Sequencing Panel	81479	$3,500
Lysosomal Storage Disease Sequencing Panel	81479	$3,500
Mitochondrial Depletion Sequencing Panel	81479	$3,000
Neuromuscular Disorders Sequencing Panel	81479	$3,500
Neuronal Ceroid Lipofuscinoses Sequencing Panel	81479	$2,500
Overgrowth/Macrocephaly Sequencing Panel	81479	$3,000
Peroxisomal Biogenesis Disorders Sequencing Panel	81479	$2,500
Rett/Angelman Syndrome Sequencing Panel	81479	$3,000
Rhabdomyolysis and Metabolic Myopathies Sequencing Panel	81479	$3,000
Syndromic Autism Sequencing Panel	81479	$3,500
X-Linked Intellectual Disability (XLID) Sequencing Panel	81470	$3,500

Molecular Testing

Focused NGS (Custom Testing)	Price	CPT Code
Single Gene	$1,500	Contact Lab
Multi-gene Panel (2-20 genes)	$3,000	Contact Lab
These customizable tests are next generation sequencing based assays utilizing our whole exome sequencing platform, the Agilent SureSelect Clinical Research Exome kit.		

Whole Exome Sequencing	Price	CPT Code
Whole Exome Sequencing	Contact Lab	Contact Lab
Greenwood's WES test captures the entire exome with additional coverage for genes with known Mendelian disease associations. The average read depth for each exome is typically greater than 150X. The analysis and curation of variants is driven by the patient's reported phenotype. The standard WES test includes trio analysis with parents. Samples from siblings can be submitted in place of a parental specimen or in addition to parental samples.		

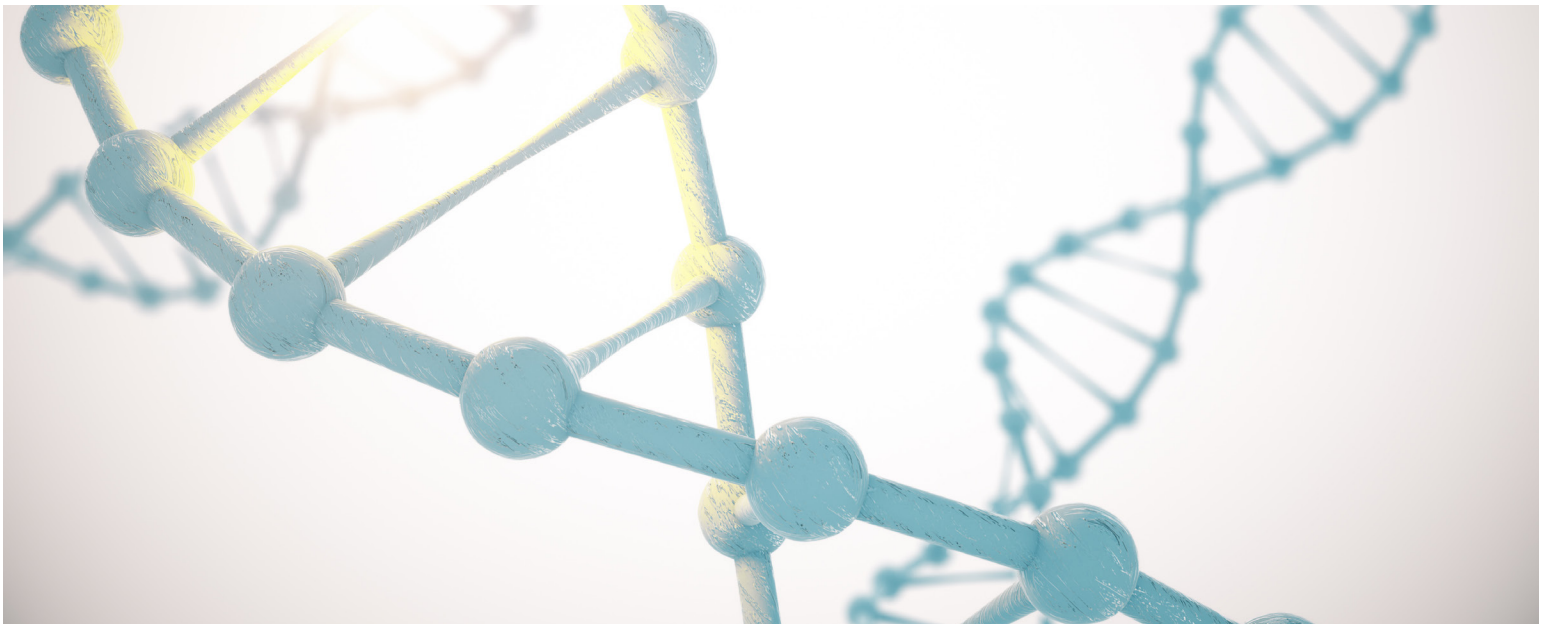
QUICK Analysis	Price	CPT Code
Quickly Uncovering Important Clinical Knowledge Analysis	$0	N/A
The QUICK Analysis is Greenwood's free NGS-reflex analysis that rapidly screens full exome data for pathogenic alterations when panel results are negative. The QUICK Analysis increases the yield of cost-effective gene panel tests. While the QUICK Analysis is not as thorough as a whole exome analysis, it has proven effective in expediting a diagnosis for a growing number of patients.		

Reflex analysis done at no charge following next generation sequencing test.

Quickly Uncovering Important Clinical Knowledge

ANALYSIS

NGS PANELS



Charcot-Marie-Tooth Hereditary Neuropathy Sequencing Panel — 81448

AARS1	*AIFM1*	*BSCL2*	*COX6A1*	*DHTKD1*	*DNAJB2*	*DNM2*	*DYNC1H1*	*EGR2*	*FGD4*	*FIG4*	*GAN*	*GARS1*	*GDAP1*
GJB1	*GNB4*	*HARS1*	*HINT1*	*HSPB1*	*HSPB8*	*IGHMBP2*	*INF2*	*KARS*	*KIF1B*	*LITAF*	*LMNA*	*LRSAM1*	*MARS1*
MED25	*MFN2*	*MME*	*MORC2*	*MPZ*	*MTMR2*	*NDRG1*	*NEFH*	*NEFL*	*PDK3*	*PLEKHG5*	*PMP22*	*PRPS1*	*PRX*
RAB7A	*SBF1*	*SBF2*	*SH3TC2*	*SLC12A6*	*SPG11*	*SURF1*	*TFG*	*TRIM2*	*TRPV4*	*VCP*	*YARS*		

Congenital Contractures Sequencing Panel — 81479

ACTA1	*ADCY6*	*ADGRG6*	*ALG2*	*ANTXR2*	*CHAT*	*CHMP1A*	*CHRNA1*	*CHRNB1*	*CHRND*	*CHRNE*	*CHRNG*	*CHST14*	*CNTN1*
CNTNAP1	*COL3A1*	*DNM2*	*DOK7*	*ECEL1*	*ERBB3*	*ERCC6*	*FBN1*	*FBN2*	*FKBP10*	*GLDN*	*GLE1*	*KLHL41*	*LMNA*
MUSK	*MYBPC1*	*MYH2*	*MYH3*	*MYH8*	*NALCN*	*NEB*	*NEK9*	*PIEZO2*	*PIP5K1C*	*PITX1*	*PLOD2*	*PLOD3*	*PSMB8*
RAPSN	*RIPK4*	*SCARF2*	*SKI*	*SLC18A3*	*SLC39A13*	*SLC5A7*	*TNNI2*	*TNNT3*	*TPM2*	*TPM3*	*UBA1*	*ZBTB42*	*ZC4H2*
ZMPSTE24													

Early Infantile Epileptic Encephalopathy Sequencing Panel — 81402x2, 81405x2, 81406x2, 81407, 81479

AARS	*ACO2*	*ADAM22*	*ALDH7A1*	*ALG13*	*AP3B2*	*ARHGEF9*	*ARV1*	*ARX*	*BRAT1*	*CACNA1A*	*CACNA2D2*	*CAD*	*CDKL5*
CHD2	*CLCN4*	*CNPY3*	*COQ4*	*CPLX1*	*CUX2*	*CYFIP2*	*DENND5A*	*DNM1*	*DOCK7*	*EEF1A2*	*FGF12*	*FRRS1L*	*GABBR2*
GABRA1	*GABRB1*	*GABRB2*	*GABRB3*	*GABRG2*	*GLS*	*GNAO1*	*GRIN2B*	*GRIN2D*	*GUF1*	*HCN1*	*HNRNPU*	*ITPA*	*KCNA2*
KCNB1	*KCNQ2*	*KCNQ3*	*KCNT1*	*KCNT2*	*MDH2*	*NECAP1*	*NTRK2*	*PACS2*	*PCDH19*	*PHACTR1*	*PIGA*	*PIGP*	*PLCB1*
PNKP	*PNPO*	*PURA*	*RHOBTB2*	*SCN1A*	*SCN1B*	*SCN2A*	*SCN3A*	*SCN8A*	*SIK1*	*SLC12A5*	*SLC13A5*	*SLC1A2*	*SLC25A12*
SLC25A22	*SLC2A1*	*SLC35A2*	*SPATA5*	*SPTAN1*	*ST3GAL3*	*STXBP1*	*SYNGAP1*	*SYNJ1*	*SZT2*	*TBC1D24*	*TRAK1*	*UBA5*	*WDR45*
WWOX	*YWHAG*												

Epilepsy/Seizure Sequencing Panel — 81479

ABAT	*ADSL*	*ALDH5A1*	*ALDH7A1*	*ALG13*	*ANKRD11*	*ARFGEF2*	*ARHGEF9*	*ARID1B*	*ARX*	*ATP1A2*	*ATP6AP2*	*BRAT1*	*CACNA1A*
CACNA1E	*CACNB4*	*CASK*	*CASR*	*CDKL5*	*CHD2*	*CHRNA2*	*CHRNA4*	*CHRNB2*	*CLCN4*	*CLN3*	*CLN5*	*CLN6*	*CLN8*
CLTC	*CNTNAP2*	*CSTB*	*CTSD*	*CUL4B*	*CUX2*	*DCX*	*DDX3X*	*DEPDC5*	*DNM1*	*DNM1L*	*DOCK7*	*DYRK1A*	*EEF1A2*
EHMT1	*EPM2A*	*FGF12*	*FLNA*	*FOLR1*	*FOXG1*	*GABBR2*	*GABRA1*	*GABRB1*	*GABRB2*	*GABRB3*	*GABRG2*	*GAMT*	*GATM*
GNAO1	*GNB1*	*GOSR2*	*GRIN1*	*GRIN2A*	*GRIN2B*	*HCN1*	*HECW2*	*HNRNPU*	*IQSEC2*	*IRF2BPL*	*KANSL1*	*KCNA1*	*KCNA2*
KCNAB1	*KCNB1*	*KCNC1*	*KCNH1*	*KCNJ10*	*KCNQ2*	*KCNQ3*	*KCNT1*	*KCNT2*	*KCTD7*	*KIF5C*	*LGI1*	*LIAS*	*MBD5*
MECP2	*MEF2C*	*MFSD8*	*MOCS1*	*MOCS2*	*MTOR*	*NALCN*	*NECAP1*	*NEDD4L*	*NEXMIF*	*NHLRC1*	*NPRL2*	*NPRL3*	*NRXN1*
OPHN1	*PACS1*	*PACS2*	*PAFAH1B1*	*PCDH19*	*PHF6*	*PHGDH*	*PIGA*	*PIGN*	*PIGO*	*PIGT*	*PLCB1*	*PLPBP*	*PNKP*
PNPO	*POLG*	*PPP2CA*	*PPP3CA*	*PPT1*	*PRICKLE1*	*PRICKLE2*	*PRRT2*	*PURA*	*QARS1*	*RELN*	*RHOBTB2*	*ROGD1*	*SCARB2*
SCN1A	*SCN1B*	*SCN2A*	*SCN3A*	*SCN8A*	*SIK1*	*SLC13A5*	*SLC25A19*	*SLC25A22*	*SLC2A1*	*SLC35A2*	*SLC6A1*	*SLC9A6*	*SMC1A*
SMS	*SNAP25*	*SPATA5*	*SPTAN1*	*ST3GAL3*	*ST3GAL5*	*STX1B*	*STXBP1*	*SYN1*	*SYNGAP1*	*SYNJ1*	*SZT2*	*TBC1D24*	*TCF4*
TPP1	*TSC1*	*TSC2*	*TUBB2A*	*UBA5*	*UBE3A*	*USP9X*	*WDR45*	*WDR62*	*WWOX*	*ZEB2*			

Hereditary Spastic Paraplegia Sequencing Panel — 81479

ABCD1	*ACOX1*	*ADAR*	*ALDH18A1*	*ALS2*	*AP4B1*	*AP4E1*	*AP4M1*	*AP4S1*	*AP5Z1*	*ATL1*	*ATP13A2*	*B4GALNT1*	*BICD2*
BSCL2	*C12orf65*	*C19orf12*	*CAPN1*	*CPT1C*	*CYP2U1*	*CYP7B1*	*DDHD1*	*DDHD2*	*ENTPD1*	*ERLIN1*	*ERLIN2*	*EXOSC3*	*FA2H*
FARS2	*GBA2*	*GJC2*	*HACE1*	*HSPD1*	*IFIH1*	*KDM5C*	*KIDINS220*	*KIF1A*	*KIF1C*	*KIF5A*	*KLC2*	*L1CAM*	*MAG*
MARS	*MECP2*	*NIPA1*	*NT5C2*	*OPA3*	*PGAP1*	*PLA2G6*	*PLP1*	*PNPLA6*	*PQBP1*	*RAB3GAP2*	*REEP1*	*REEP2*	*RNASEH2B*
RTN2	*SACS*	*SAMHD1*	*SLC16A2*	*SLC2A1*	*SLC33A1*	*SPAST*	*SPG11*	*SPG20*	*SPG21*	*SPG7*	*TECPR2*	*TFG*	*TREX1*
TUBB3	*TUBB4A*	*UCHL1*	*VAMP1*	*VPS37A*	*WASHC5*	*WDR45*	*ZFYVE26*	*ZFYVE27*					

Lysosomal Storage Disorders Sequencing Panel — 81479

ADAMTSL2	*AGA*	*ANTXR2*	*ARSA*	*ARSB*	*ASAH1*	*ATP13A2*	*CLN3*	*CLN4*	*CLN5*	*CLN6*	*CLN8*	*COL11A2*	*COL2A1*
CTNS	*CTSA*	*CTSC*	*CTSD*	*CTSK*	*DHCR7*	*DYM*	*FUCA1*	*GAA*	*GALC*	*GALNS*	*GBA*	*GLA*	*GLB1*
GM2A	*GNE*	*GNPTAB*	*GNS*	*GPC3*	*GUSB*	*HEXA*	*HEXB*	*HGSNAT*	*HRAS*	*HYAL1*	*IDS*	*IDUA*	*KMT2D*
LAMP2	*LIPA*	*MAN2B1*	*MANBA*	*MCOLN1*	*MFSD8*	*NAGA*	*NAGLU*	*NEU1*	*NPC1*	*NPC2*	*PEX1*	*PEX10*	*PEX12*
PEX13	*PEX14*	*PEX16*	*PEX19*	*PEX2*	*PEX26*	*PEX3*	*PEX5*	*PEX6*	*PHYH*	*PPT1*	*PSAP*	*RAI1*	*SGSH*
SLC17A5	*SMPD1*	*SUMF1*	*TCF4*	*TPP1*									

Mitochondrial Depletion Sequencing Panel — 81479

ABAT	*AGK*	*APTX*	*C10ORF2*	*DGUOK*	*DNA2*	*FBXL4*	*GFER*	*MFN2*	*MGME1*	*MPV17*	*OPA1*	*OPA3*	*POLG*
POLG2	*RRM2B*	*SLC25A4*	*SPG7*	*SUCCLA2*	*SUCLG1*	*TFAM*	*TK2*	*TYMP*					

Neuromuscular Disorders Sequencing Panel — 81479

ACTA1	*AGK*	*AMPD1*	*ANO5*	*ATP2A1*	*ATP7A*	*B3GALNT2*	*B4GAT1*	*BAG3*	*BICD2*	*BIN1*	*BSCL2*	*CACNA1S*	*CAPN3*
CAV3	*CAVIN1*	*CCDC78*	*CFL2*	*CHAT*	*CHKB*	*CHRNA1*	*CHRNB1*	*CHRND*	*CHRNE*	*CHRNG*	*CLCN1*	*CNTN1*	*COL12A1*
COL6A1	*COL6A2*	*COL6A3*	*COLQ*	*CRPPA*	*CRYAB*	*DAG1*	*DCTN1*	*DES*	*DMD*	*DNAJB6*	*DNM2*	*DNMT1*	*DOK7*
DPM1	*DPM2*	*DPM3*	*DYNC1H*	*DYSF*	*EMD*	*FBXL4*	*FHL1*	*FIG4*	*FKRP*	*FKTN*	*FLNC*	*GAA*	*GAN*
GARS	*GLE1*	*GMPPB*	*GNE*	*HINT1*	*HNRNPDL*	*HSPB1*	*HSPB8*	*IGHMBP2*	*ITGA7*	*KBTBD13*	*KLHL40*	*KLHL41*	*LAMA2*
LAMP2	*LARGE1*	*LAS1L*	*LDB3*	*LIMS2*	*LMNA*	*LMOD3*	*MATR3*	*MEF10*	*MGME1*	*MTM1*	*MTMR14*	*MUSK*	*MYF6*
MYH2	*MYH7*	*MYOT*	*NEB*	*PABPN1*	*PHKA1*	*PLEC*	*PLEKHG5*	*PMM2*	*PNPLA2*	*POLG*	*POLG2*	*POMGNT1*	*POMGNT2*
POMK	*POMT1*	*POMT2*	*PYGM*	*RAPSN*	*REEP1*	*RRM2B*	*RYR1*	*SCN4A*	*SELENON*	*SETX*	*SGCA*	*SGCB*	*SGCD*
SGCE	*SGCG*	*SIL1*	*SLC25A4*	*SLC5A7*	*SMCHD1*	*SPEG*	*STAC3*	*SUCLA2*	*SUCLG1*	*SYNE1*	*SYNE2*	*TCAP*	*TIA1*
TK2	*TMEM43*	*TMEM5*	*TNNI2*	*TNNT1*	*TNPO3*	*TOR1AIP1*	*TPM2*	*TPM3*	*TRAPPC11*	*TRIM32*	*TRPV4*	*TTN*	*TWNK*
TYMP	*UBA1*	*VCP*	*VRK1*										

Neuronal Ceroid Lipofuscinoses Sequencing Panel — 81479

CLN3	*CLN4*	*CLN5*	*CLN6*	*CLN8*	*CTSD*	*MFSD8*	*PPT1*	*TPP1*

Overgrowth/Macrocephaly Sequencing Panel — 81479

BRWD3	*CDKN1C*	*CUL4B*	*DNMT3A*	*EED*	*EZH2*	*GLI3*	*GPC3*	*MED12*	*NFIX*	*NSD1*	*PHF6*	*PTCH1*	*PTEN*
RNF135	*UPF3B*												

Peroxisomal Biogenesis Sequencing Panel — 81479

BRWD3	*CDKN1C*	*CUL4B*	*DNMT3A*	*EED*	*EZH2*	*GLI3*	*GPC3*	*MED12*	*NFIX*	*NSD1*	*PHF6*	*PTCH1*	*PTEN*
RNF135	*UPF3B*												

Rett/Angelman Syndrome Sequencing Panel — 81479

ARX	ATRX	CDKL5	CNTNAP2	CTNNB1	EHMT1	FOLR1	FOXG1	MBD5	MECP2	MEF2C	NRXN1	OPHN1	PCDH19
PNKP	PQBP1	SLC2A1	SLC9A6	TCF4	UBE3A	ZEB2							

Rhabdomylysis & Metabolic Myopathies Sequencing Panel — 81479

ACADM	ACADVL	AGL	ALDOA	AMPD1	ANO5	ATP2A1	C10ORF2	CASQ1	CAV3	CPT2	CTDP1	CYTB	DGUOK
DMD	DYSF	ENO3	ETFA	ETFB	ETFDH	FDX1L	FKRP	FKTN	GAA	HADHA	HADHB	ISCU	LDHA
LPIN1	MT-CYB	PFKM	PGAM2	PGK1	PGM1	PHKA1	PHKB	POLG	PYGM	RRM2B	RYR1	SCN4A	SIL1
SLC16A1	SLC25A20	SUCLA2	TK2	TSEN54	TSFM								

Syndromic Autism Sequencing Panel — 81479

ADNP	*ALDH5A1*	*AMT*	*AP1S2*	*ARID1B*	*ARX*	*ATRX*	*BCKDK*	*BRAF*	*CACNA1C*	*CASK*	*CDKL5*	*CHD7*	*CHD8*
CNTNAP2	*CREBBP*	*CTNNB1*	*DHCR7*	*DYRK1A*	*EHMT1*	*FGD1*	*FMR1*	*FOLR1*	*FOXG1*	*FOXP1*	*FOXP2*	*GABRB3*	*GRIN2B*
HDAC8	*HOXA1*	*HPRT1*	*KDM5C*	*KIRREL3*	*L1CAM*	*LAMC3*	*MBD5*	*MECP2*	*MED12*	*MEF2C*	*MID1*	*NHS*	*NIPBL*
NLGN3	*NLGN4X*	*NRXN1*	*NSD1*	*NTNG1*	*OPHN1*	*PAFAH1B1*	*PCDH19*	*PHF6*	*PNKP*	*PQBPQ*	*PTCHD1*	*PTEN*	*PTPN11*
RAB39B	*RAD21*	*RAI1*	*RELN*	*SCN1A*	*SCN2A*	*SETBP1*	*SETD2*	*SHANK3*	*SLC2A1*	*SLC9A6*	*SMC1A*	*SMC3*	*STXBP1*
SYNE1	*TBL1XR1*	*TBR1*	*TCF4*	*TMEM231*	*TMLHE*	*TSC1*	*TSC2*	*TUBA1A*	*UBE3A*	*UBE3C*	*VPS13B*	*ZEB2*	

X-Linked Intellectual Disability (XLID) Sequencing Panel — 81470

ABCD1	*ACSL4*	*AFF2*	*AGTR2*	*AIFM1*	*AP1S2*	*ARHGEF6*	*ARHGEF9*	*ARX*	*ATP6AP2*	*ATP7A*	*ATR-X*	*BCOR*	*BRWD3*
CASK	*CCDC22*	*CDK16*	*CDKL5*	*CLCN4*	*CLIC2*	*CNKSR2*	*CUL4B*	*DCX*	*DKC1*	*DLG3*	*DMD*	*EBP*	*EIF2S3*
FAAH2	*FANCB*	*FGD1*	*FLNA*	*FMR1*	*FRMPD4*	*FTSJ1*	*GDI1*	*GK*	*GPC3*	*GRIA3*	*GSPT2*	*HCCS*	*HCFC1*
HDAC8	*HPRT1*	*HSD17B10*	*HUWE1*	*IDS*	*IGBP1*	*IL1RAPL1*	*IQSEC2*	*KDM5C*	*KIAA2022*	*KLF8*	*L1CAM*	*LAMP2*	*LAS1L*
MAGT1	*MAOA*	*MBTPS2*	*MECP2*	*MED12*	*MID1*	*MTM1*	*NAA10*	*NDP*	*NDUFA1*	*NHS*	*NLGN3*	*NLGN4X*	*NSDHL*
OCRL	*OFD1*	*OGT*	*OPHN1*	*OTC*	*PAK3*	*PCDH19*	*PHF6*	*PHF8*	*PLP1*	*PORCN*	*PQBP1*	*PRPS1*	*PTCHD1*
RAB39B	*RAB40AL*	*RBM10*	*RPL10*	*RPS6KA3*	*SHROOM4*	*SLC16A2*	*SLC9A6*	*SMC1A*	*SMS*	*SOX3*	*SRPX2*	*SYN1*	*SYP*
TAF1	*THOC2*	*TIMM8A*	*TSPAN7*	*UBE2A*	*UPF3B*	*USP9X*	*WDR13*	*ZC4H2*	*ZCCHC12*	*ZDHHC15*	*ZDHHC9*	*ZMYM3*	*ZNF41*
ZNF711	*ZNF81*												

ENZYME PANELS

DBS Lysosomal Storage Disease Enzyme Panel
Price: $800 CPT Code: 82657 (x4) Sample Type: D

Disorders	Enzymes
α-mannosidosis	α-mannosidase
β-mannosidosis	β-mannosidase
Aspartylglucosaminuria	Aspartylglucosaminidase
Fabry Disease	α-galactosidase
Fucosidosis	α-fucosidase
Gaucher Disease	β-glucosidase
Krabbe Disease	Galactocerebrosidase
MPS IVB, GM1-Gangliosidosis	β-galactosidase
Neuronal Ceroid Lipofuscinosis	Tripeptidyl-peptidase 1
Niemannn-Pick Disease A/B	Acid Sphingomyelinase
Pompe Disease	α-glucosidase
Schindler/Kanzaki Disease	N-acetyl-alpha galactosaminidase

DBS Mucopolysaccharidosis (MPS) Enzyme Panel
Price: $800 CPT Code: 82657 (x4) Sample Type: D, WB

Disorders	Enzymes
Hunter syndrome (MPS II)	Iduronate-2-sulfatase
Hurler syndrome (MPS I)	α-iduronidase
Sanfilippo syndrome B (MPS IIIB)	N-acetyl-α-glucosaminidase
Maroteaux-Lamy syndrome (MPS VI)	Arylsulfatase B
Morquio syndrome A (MPS IVA)	N-acetyl galactosamine-6-sulfatase
Morquio syndrome B (MPS IVB)	β-galactosidase
Sly syndrome (MPS VII)	β-glucuronidase

Hydrops Enzyme Panel
Price: $800 CPT Code: 82657 (x4) Sample Type: F

Disorders	Enzymes
Gaucher Disease	β-glucosidase
GM1-Gangliosidosis	β-galactosidase
Sialidosis	α-neuraminidase (sialidase)
Sly syndrome (MPS VII)	β-glucronidase

Lysosomal Storage Disease Enzyme Panel
Price: $1,000 CPT Code: 82657 (x5) Sample Type: WB

Disorders	Enzymes
α-mannosidosis	α-mannosidase
β-mannosidosis	β-mannosidase
Aspartylglucosaminuria	Aspartylglucosaminidase
Fabry Disease	α-galactosidase
Fucosidosis	α-fucosidase
Gaucher Disease	β-glucosidase
Hurler Syndrome (MPS I)	α-iduronidase
Krabbe Disease	Galactocerebrosidase
Metachromatic Luekodystrophy	Arylsulfatase A
MPS IVB, GM1-Gangliosidosis	β-galactosidase
Niemann-Pick Disease A/B	Acid sphingomyelinase
Schindler/Kanzaki Disease	N-acetyl-alpha galactosaminidase
Tay-Sachs/Sandhoff Disease	β-hexosaminidase

Morquio Syndrome (MPS IV) Enzyme Panel, Type A & B
Price: $400 CPT Code: 82657 (x2) Sample Type: F, L, WB

Disorders	Enzymes
Morquio A syndrome (MPS IVA)	N-acetyl galactosamine-6-sulfatase
Morquio B syndrome (MPS IVB)	β-galactosidase

ENZYME PANELS

Mucopolysaccharidosis (MPS) Enzyme Panel
Price: $1,000 CPT Code: 82657 (x5) Sample Type: F, WB

Disorders	Enzymes
Hunter syndrome (MPS II)	Iduronate-2-sulfatase
Hurler syndrome (MPS I)	α-iduronidase
Maroteaux-Lamy syndrome (MPS VI)	Arylsulfatase B
Morquio syndrome A (MPS IVA)	N-acetyl galactosamine-6-sulfatase
Morquio syndrome B (MPS IVB)	β-galactosidase
Sanfilippo syndrome A (MPS IIIA)	Heparan-N-sulfatase
Sanfilippo syndrome B (MPS IIIB)	N-acetyl-alpha-D-glucosaminidase
Sanfilippo syndrome C (MPS IIIC)	Acetyl CoAglucosamine N acetyl trasnferase
Sanfilippo syndrome D (MPS IIID)	N-acetyl glucosamine-6-sulfatase
Sly syndrome (MPS VII)	β-glucuronidase

Mucolipidosis II/III Screen
Price: $400 CPT Code: 82657 (x2) Sample Type: DBS, P

Three or four enzymes will be measured in plasma or DBS to detect elevated activity

Multiple Sulfatase Deficiency Enzyme Panel
Price: $400 CPT Code: 82657 (x2) Sample Type: F, DBS, WB

This panel contains 3 sulfastase enzymes: Arysulfatase B, Iduronate-2-sulfatase, and N-actyl-galactosamine-6-sulfatase

Neurological (Sphingolipidoses) Panel
Price: $600 CPT Code: 82657 (x3) Sample Type: WB

Disorders	Enzyme
Fabry Disease	α-galactosidase
Gaucher Disease	β-galactosidase
Krabbe Disease	Galactocerebrosidase
Metachromatic Luekodystrophy	Arylsulfatase A
MPS IVB, GM1-Gangliosidosis	β-galactosidase
Neuronal Ceroid Lipofuscinosis 1	Palmitoyl-protein thioesterase 1
Neuronal Ceroid Lipofuscinosis 2	Tripeptidyl peptidase 1
Niemann Pick Disease A/B	Acid Spingomyelinase
Tay-Sachs/Sandhoff Disease	β-hexosaminidase

Oligosaccharidoses Panel
Price: $600 CPT Code: 82657 (x3) Sample Type: D, F*, L

Disorders	Enzyme
α-mannosidosis	α-mannosidase
β-mannosidosise	β-mannosidase
Aspartylglucosaminuria	Aspartylglucosaminidase
Fucosidosis	α-fucosidase
MPS IVB, GM1-Gangliosidosis	β-galactosidase
Schindler Disease	α-N-acetyl galactosaminidase
Sialidosis*	α-neurominidase-sialidase

* Only fibroblasts are accepted for Sialidosis

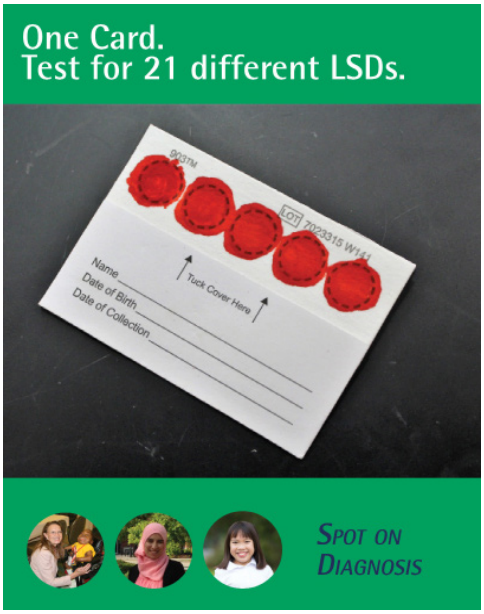
Sanfilippo Syndrome (MPS III) Enzyme Panel
Price: $800 CPT Code: 82657 (x4) Sample Type: F, WB

Disorders	Enzymes
Sanfilippo syndrome A (MPS IIIA)	Heparan-N-sulfatase
Sanfilippo syndrome B (MPS IIIB)	N-acetyl-alpha-D-glucosaminidase
Sanfilippo syndrome C (MPS IIIC)	Acetyl CoAglucosamine N acetyl transferase
Sanfilippo syndrome D (MPS IIID)	N-acetyl glucosamine-6-sulfatase

BIOCHEMICAL TESTING

ANALYTE PANELS	Price	CPT Code
Biochemical Genetics Profile (P, U, WB) Acylcarnitine Profile Amino acid Analysis Carnitine Analysis (Total and Free) Organic Acid Analysis	$821	82017 82139 82379 83919
Storage Disease: Analyte Panel (Urine) Mucopolysaccharidosis Urine Analysis Oligosaccharide Urine Analysis Siaclic Acid Analysis	$750	83684 (X2) 84377 84275

All enzymes are available for individual analysis for $200. CPT Codes may vary.



DRIED BLOOD SPOT CARD

Enzyme Analysis AND Gene Sequencing for more than 20 Lysosomal Storage Diseases all from ONE DBS Sample!

Confirmatory molecular testing is available at a 20% discount when enzyme testing performed at Greenwood is abnormal.

BIOCHEMICAL SAMPLE & SHIPPING REQUIREMENTS

Dried Blood Spot (D) : Completely fill at least 3 circles with a single layer of blood for each circle. Dry spots 3-4 hours prior to sending.

Plasma (P) : Sodium heparin tube (green top) tube. Ship whole blood overnight at room temperature, OR spin down, remove plasma and send plasma frozen.

Leukocytes (L) : Ship whole blood in sodium heparin (green top) tube overnight. Samples should arrive within 24-48 hours of draw, OR isolated leukocyte pellet can be shipped frozen overnight.

Fibroblasts (F) : Fresh tissue should be placed in transport media (preferred) or sterile saline and shipped overnight. For cultured tissue, please send two T25 flasks overnight. If cultured tissue is being sent, a control flask is requested in addition to the patient sample.

Whole Blood (WB) : Ship whole blood in sodium heparin (green top) tube. Samples should arrive within 24-48 hours of draw. Ship overnight at room temperature.

Urine (U) : Send frozen via overnight shipping.

Single Tests (Molecular)

Individual Gene Sequencing	CPT Code	Price
3-Methylcrotonylglycinuria I/II : *MCCC1/MCCC2* Sequencing	81406x2	$2,000
Adrenoleukodystrophy, X-linked : *ABCD1* Sequencing	81405	$1,000
Alpha-mannosidosis : *MAN2B1* Sequencing	81479	$1,500
Angelman syndrome : *UBE3A* Sequencing	81406	$1,500
Aspartylglucosaminuria : *AGA* Sequencing	81479	$1,000
Beta-mannosidosis : *MANBA* Sequencing	81479	$1,000
Biotinidase Deficiency : *BTD* Sequencing	81404	$1,000
Carnitine Palmitoyltransferase IA Deficiency : *CPT1A* Sequencing	81406	$1,500
Carnitine Palmitoyltransferase II Deficiency : *CPT2* Sequencing	81404	$1,000
Citrullinemia, Type 1 : *ASS1* Sequencing	81406	$1,500
Congenital Disorder of Glycosylation 1a : *PMM2* Sequencing	81479	$1,000
Congenital Disorder of Glycosylation 1b : *MPI* Sequencing	81405	$1,000
Congenital Disorder of Glycosylation 1c : *ALG6* Sequencing	81479	$1,000
Copper Transport Disorders : *ATP7A* Sequencing	81479	$1,500
Creatine Transporter Deficiency : *SLC6A8* Sequencing	81479	$1,500
Fabry Disease : *GLA* Sequencing	81405	$1,000
FLNA-Related Disorders : *FLNA* Sequencing	81479	$1,500
Fucosidosis : *FUCA1* Sequencing	81479	$1,000
Galactosemia : *GALT* Sequencing	81406	$1,000
Galactosialidosis : *CTSA* Sequencing	81479	$1,200
Glutaric Acidemia, Type I : *GCDH* Sequencing	81406	$1,000
Glycogen Storage Disease Type II, Pompe Disease : *GAA* Sequencing	81406	$1,000
Glycogen Storage Disease, Type 0 : *GYS2* Sequencing	81479	$1,200
Hunter Syndrome (MPSII) : *IDS* Sequencing	81405	$1,000
Hurler Syndrome (MPS I) : *IDUA* Sequencing	81406	$1,000
Krabbe Disease : *GALC* Sequencing	81406	$1,000
Maroteaux-Lamy Syndrome (MPSVI) : *ARSB* Sequencing	81479	$800
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency : *ACADM* Sequencing	81479	$1,000
Metachromatic Leukodystrophy : *ARSA* Sequencing	81405	$1,000
Morquio Syndrome A (MPSIVA) : *GALNS* Sequencing	81479	$1,000
Morquio Syndrome B (MPS IVB), GM1 Gangliosidosis : *GLB1* Sequencing	81479	$1,200
Neuronal Ceroid Lipofuscinosis Type 1 (CLN1) : *PPT1* Sequencing	81479	$800
Neuronal Ceroid Lipofuscinosis Type 2 (CLN2) : *TPP1* Sequencing	81479	$1,000
Neuronal Ceroid Lipofuscinosis Type 3, Batten Disease : *CLN3* Sequencing	81479	$1,000
Niemann-Pick Disease A/B : *SMPD1* Sequencing	81479	$800
Ornithine Transcarbamylase Deficiency : *OTC* Sequencing	81405	$1,000
Pelizaeus-Merzbacher Disease, Spastic Paraplegia : *PLP1* Sequencing	81405	$700
Phenylketonuria : *PAH* Sequencing	81406	$1,000
Primary Carnitine Deficiency, systemic : *SLC22A5* Sequencing	81405	$1,000
Rett Syndrome : *MECP2* Sequencing	81302	$900
Sandhoff Disease : *HEXB* Sequencing	81479	$900
Sanfilippo Syndrome A (MPS IIIA) : *SGSH* Sequencing	81479	$1,200
Sanfilippo Syndrome B (MPS IIIB), : *NAGLU* Sequencing	81479	$1,200
Sanfilippo Syndrome C (MPSIIIC) : *HGSNAT* Sequencing	81479	$1,500
Sanfilippo Syndrome D (MPS IIID : *GNS* Sequencing	81479	$1,000
Short-Chain Acyl-CoA Dehydrogenase Deficiency : *ACADS* Sequencing	81405	$1,000
Sialidosis : *NEU1* Sequencing	81479	$800
Sly Syndrome (MPS VII) : *GUSB* Sequencing	81479	$1,000
Spinal Muscular Atrophy : *SMN1* Sequencing	81405	$1,000

Single Tests (Molecular)

Sequencing	CPT Code	Price
Succinyl CoA : 3-oxoacid CoA transferase Deficiency : OXCT1 Sequencing	81479	$1,000
Tay-Sachs Disease : HEXA Sequencing	81406	$1,000
Very Long Chain Fatty Acid Deficiency : ACADVL Sequencing	81406	$1,500
X-Linked Hydrocephalus : L1CAM Sequencing	81407	$1,500
Deletion/Duplication	**CPT Code**	**Price**
Charcot-Marie-Tooth Disease, Type 1A : PMP22 Deletion/Duplication MLPA	81324	$500
Duchenne/Becker Muscular Dystrophy : DMD Deletion/Duplication (MLPA)	81161	$500
Hunter Syndrome (MPSII) : IDS Deletion/Duplication (MLPA)	81404	$500
Pelizaeus-Merzbacher Disease, Spastic Paraplegia : PLP1 Deletion/Duplication (MLPA)	81404	$500
Rett Syndrome : MECP2 Deletion/Duplication (MLPA)	81304	$500
Spinal Muscular Atrophy : SMN1/SMN2 Deletion/Duplication (MLPA)	81329	$600
Targeted Analysis	**CPT Code**	**Price**
Rett Syndrome : MECP2 Targeted Mutation Analysis	81303	$350
Trinucleotide Repeat Analysis	**CPT Code**	**Price**
Myotonic Dystrophy : DMPK Trinucleotide Repeat Analysis	81234	$350
Methylation	**CPT Code**	**Price**
Angelman/Prader-Willi Syndrome Methylation Analysis	81331	$350
EpiSign Complete	Contact Lab	$1,500
EpiSign Variant	Contact Lab	$1,200

Cytogenetic Testing

Chromosome Analysis	CPT Code	Price
Chromosome Analysis, High Resolution (Blood)	88230, 88262, 88289, 88291	$794
Chromosome Analysis, Routine (Blood)	88230, 88262,88291	$602
Chromosome Analysis, Routine, Rule out mosaic (Blood)	88230, 88263, 88285 (x5), 88291	$755
Microarray	**CPT Code**	**Price**
CytoScan Xon Microarray : Single Gene Analysis	Contact Lab	$700
CytoScan Xon Microarray : 2-10 Genes	Contact Lab	$1,200
CytoScan Xon Microarray : More than 10 Genes	Contact Lab	$1,950

FISH Analysis	Chromosomal Region	CPT Code	Price
Angelman Syndrome	15q11q13	88275, 88273 88271, 88291	$584
DiGeorge/VCF	22q11.2	88275, 88273, 88271, 88291	$584
Kallmann Syndrome	Xp22.3	88275, 88273, 88271, 88291	$584
Miller-Dieker Syndrome	17p13	88275, 88273, 88271, 88291	$584
Smith-Magenis Syndrome	17p11.2	88275, 88273, 88271, 88291	$584
Williams Syndrome	7q11.23	88275, 88273, 88271, 88291	$584
Wolf-Hirschhorn Syndrome	4p-	88275,88273, 88271, 88291	$584

Our team looks forward to working with you.






Ray Louie, PhD




Benjamin Hilton, PhD

Mission Statement

The Greenwood Genetic Center is a nonprofit institute organized to provide clinical genetic services, diagnostic laboratory testing, educational programs and resources and research in the field of medical genetics.

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