NEUROLOGY TEST REQUISITION FORM

GeneDx

PATIENT INFORMATION

First Name	Last Name	
Genetic Sex ○ Male ○ Female Gender Identification (optional): _____	Date of Birth (mm/dd/yy)	
Ancestry ○ White/Caucasian ○ Hispanic ○ Black/African American ○ Native American ○ East Asian ○ South Asian ○ Middle Eastern ○ Ashkenazi Jewish ○ Other: _____		
Email		
Address		
City	State	Zip Code
Primary Phone	Is this patient deceased? ○ Yes ○ No Deceased Date: _____	

SAMPLE INFORMATION

Date Sample Collected (mm/dd/yy) (required):	Medical Record #
○ Blood ○ Buccal Swab ○ Other (specify source): _____	
Patient has had a blood transfusion ○ Yes ○ No Date of Last Transfusion: _____ (2-4 weeks of wait time is required for some testing)	
Patient has had an allogenic bone marrow transplant ○ Yes ○ No Fibroblasts are recommended for patients who had an allogenic bone marrow transplant. See www.genedx.com/specimen-requirements for details.	
○ Treatment-Related **RUSH** Date: _____	

PATIENT CONSENTS

By signing this form, I acknowledge as the patient or relative being tested that I have read or have had read to me the GeneDx Informed Consent document at the end of this test requisition form, and understand the information regarding molecular genetics testing. I have had the opportunity to ask questions about the testing, the procedure, the risks, and the alternatives. By signing this form, I authorize GeneDx to perform genetic testing as ordered. I understand that, for tests that evaluate data from multiple family members concurrently, test results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their healthcare providers.

More information, including the GeneDx Notice of Privacy Policies, is available on GeneDx's website: www.genedx.com

○ By checking this box, I confirm that I am a New York state resident, and I give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing, and to be used as a de- identified sample for test development and improvement, internal validation, quality assurance, and training purposes. Otherwise, New York law requires GeneDx to destroy my sample after 60 days, and it cannot be used for test development studies.

○ Check this box if you wish to opt out of being contacted for research studies.

○ Check this box if you do not wish to receive ACMG secondary findings (Full Exome Sequencing and Genome Sequencing Tests ONLY; not for Xpanded® or Slice tests).

Signature of Patient/Legal Guardian (required)	**Date**
Signature of Relative A/Legal Guardian	**Date**
Signature of Relative B/Legal Guardian	**Date**

OPTIONAL AND FOR COMMERCIAL INSURANCE ONLY:

By entering my preferred contact information below, I give my permission to GeneDx to send me an email and/or text with a link to access my personalized Digital Patient Letter. Data rates may apply.

Mobile Number*	Email*

*Contact information provided must be for the individual authorizing the genetic testing.

ACCOUNT INFORMATION

GeneDx Account Number		**Account Name**
Phone		Fax
Address		City
State	Zip Code	Country
Ordering Provider Name		Role/Title
NPI		Phone Number
Send Report Via ○ Fax ○ Email ○ Portal Fax #/Email: _____		
Additional Reporting Provider's Name		
Send Report Via ○ Fax ○ Email ○ Portal Fax #/Email: _____		
SEND ADDITIONAL REPORT COPIES TO:		
Provider Name		GeneDx Acct#
Fax #/Email:		

ICD-10 CODES *(Required)*

ICD-10 Codes	
Clinical Diagnosis	Age of Onset

STATEMENT OF MEDICAL NECESSITY

By submission of this test requisition and accompanying sample(s), I: (i) authorize and direct GeneDx to perform the testing indicated; (ii) certify that the person listed as the ordering provider is authorized by law to order the test(s) requested; (iii) certify that any custom panel and/or ordered test(s) requested on this test requisition form are reasonable and medically necessary for the diagnosis and/or treatment of a disease, illness, impairment, symptom, syndrome or disorder; (iv) the test results will determine my patient's medical management and treatment decisions of this patient's condition on this date of service; (v) have obtained this patient's and relatives', when applicable, written informed consent to undergo any genetic testing requested; and (vi) that the full and appropriate diagnosis code(s) are indicated to the highest level of specificity.

Signature of Provider (required)	Date

PAYMENT OPTIONS *(Select One)*

○ **INSURANCE BILL** *(select all that applies)*

○ Commercial
○ Medicaid
○ Medicare
○ Tricare

FOR ALL INSURANCE CARDS PROVIDE FRONT AND BACK COPY OF CARD(S)

Patient Status
○ Hospital outpatient ○ Hospital inpatient; Date of Discharge _____
○ Not a hospital patient

Name of Insurance Carrier	Insurance ID#:
Relationship to Insured ○ Self ○ Spouse ○ Child ○ Other: _____	Policy Holder's Name Policy Holder's Date of Birth
Referral/Prior Authorization # (please attach)	GeneDx Benefit Investigation #

Secondary Insurance Type:

Insurance Carrier	Insurance ID #	Subscriber Name	Date of Birth

Relationship to Insured:
○ Self ○ Spouse ○ Child ○ Other: _____

○ **PATIENT BILL**

Amount Due: _____

If Patient Bill is selected, I am electing to be treated as a self-pay patient for this testing. I agree that neither GeneDx nor I will submit a claim to my insurance for this testing, if I have insurance. GeneDx will send an invoice to the patient listed above.

Authorized Patient/Guardian Signature

○ **INSTITUTIONAL BILL**

GeneDx Account #

Hospital/Lab Name

Place Sticker/Stamp Here

CLINICAL INFORMATION

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

CLINICAL INFORMATION

Relevant clinical records are required at the time of sample submission to ensure the information is included in data analysis.

Pre/Perinatal History
- ○ Growth delay
- ○ Increased body weight
- ○ Intrauterine growth retardation
- ○ Prematurity GA: _____

Structural Brain Abnormalities
- ○ Abnormal myelination
- ○ Abnormality of basal ganglia
- ○ Abnormality of brainstem
- ○ Abnormality of periventricular white matter
- ○ Abnormality of the corpus callosum
- ○ Aplasia/hypoplasia of cerebellar vermis
- ○ Aplasia/hypoplasia of cerebellum
- ○ Arnold Chiari malformation
- ○ Brain atrophy
- ○ Cerebellar atrophy
- ○ Cerebellar hypoplasia (Pontocerebellar hypoplasia)
- ○ CNS hypomyelination
- ○ Cortical dysplasia
- ○ Cortical tubers
- ○ Frontotemporal cerebral atrophy
- ○ Heterotopia (Periventricular nodular heterotopia)
- ○ Holoprosencephaly
- ○ Hydrocephalus
- ○ Leukodystrophy
- ○ Lissencephaly
- ○ Molar tooth sign on MRI
- ○ Pachygyria
- ○ Polymicrogyria
- ○ Pontocerebellar atrophy
- ○ Subcortical band heterotopia
- ○ Ventriculomegaly

Developmental/Behavioral Findings
- ○ Abnormal aggressive, impulsive or violent behavior
- ○ Abnormal social behavior
- ○ Absent speech
- ○ Aggressive behavior
- ○ Anxiety
- ○ Attention deficit hyperactivity disorder
- ○ Autistic behavior
- ○ Behavioral abnormality
- ○ Clumsiness
- ○ Cognitive impairment
- ○ Delayed fine motor development
- ○ Delayed gross motor development
- ○ Delayed speech & language development
- ○ Depression
- ○ Developmental regression
- ○ Dysarthria
- ○ Frequent falls
- ○ Gait disturbance
- ○ Global developmental delay
- ○ Hyperactivity
- ○ Incoordination
- ○ Intellectual disability
- ○ Memory impairment
- ○ OCD
- ○ Sleep disturbance
- ○ Specific learning disability
- ○ Speech articulation difficulties
- ○ Stereotypy

Neurological Findings
- ○ Abnormality of nervous system
- ○ Ataxia
- ○ Cerebral palsy
- ○ Chorea
- ○ Cortical visual impairment
- ○ Dementia
- ○ Dysarthria
- ○ Dyskinesia
- ○ Dysphasia
- ○ Dystonia
- ○ Encephalopathy
- ○ Epileptic encephalopathy
- ○ Familial or Sporadic hemiplegic migraine
- ○ Febrile seizures
- ○ Focal seizures
- ○ Frontotemporal dementia
- ○ Generalized Seizures
- ○ Headaches
- ○ Hyperreflexia
- ○ Infantile spasms
- ○ Myotonia
- ○ Myoclonus
- ○ Paresthesia
- ○ Parkinsonism
- ○ Peripheral neuropathy
- ○ Reduced tendon reflexes
- ○ Seizures
- ○ Sensory neuropathy
- ○ Spasticity
- ○ Status epilepticus
- ○ Stroke-like episode
- ○ Tremors
- ○ Upper motor neuron dysfunction
- ○ Vocal cord paresis

Craniofacial/Dysmorphism
- ○ Abnormal facial shape (Dysmorphic features)
- ○ Macrocephaly
- ○ Microcephaly

Eye Defects/Vision
- ○ Abnormality of Vision
- ○ Cataracts
- ○ Nystagmus
- ○ Optic Atrophy

Hearing Impairment
- ○ Abnormal newborn screen:

  _____
- ○ Sensorineural hearing impairment/bilateral

Cardiac Findings
- ○ Cardiac rhabdomyoma

Respiratory Findings
- ○ Apnea
- ○ Hyperventilation
- ○ Hypoventilation
- ○ Respiratory distress
- ○ Respiratory insufficiency

Gastrointestinal Findings
- ○ Failure to thrive
- ○ Feeding difficulties

Musculoskeletal Findings
- ○ Arthrogryposis
- ○ Decreased muscle mass
- ○ Exercise intolerance
- ○ Fasciculations
- ○ Fatigue
- ○ Foot dorsiflexor weakness (foot drop)
- ○ Hypertonia
- ○ Hypotonia
- ○ Joint hypermobility
- ○ Muscle cramps
- ○ Muscle weakness
- ○ Myalgia
- ○ Myopathic facies
- ○ Myopathy
- ○ Pain
- ○ Pes cavus
- ○ Pes planus
- ○ Rhabdomyolysis
- ○ Scoliosis
- ○ Short stature

Skin/Hair Findings
- ○ Axillary freckling
- ○ Café-Au-Lait Macules
- ○ Hyperpigmentation of the skin
- ○ Hypopigmentation of the skin

Metabolic Issues/Mitochondrial
(Attach relevant lab reports/values)

- ○ Abnormal newborn screen result:

  _____
- ○ Elevated CPK:

  _____

Endocrine Findings
- ○ Delayed puberty

Vascular System
- ○ Arteriovenous malformation
- ○ Stroke

- ○ Other:

  _____

Signature of Provider (required)	Date

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GeneDx

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

TARGETED VARIANT TESTING AND SPECIAL SERVICES

Individual to be Tested: ○ Affected/Symptomatic ○ Unaffected/Asymptomatic

○ Known Familial Variant(s) in a Nuclear Gene
○ Known Familial Copy Number Variant(s)
○ Confirmation of Variant Identified in Research Lab
○ Targeted Mosaic Variant Testing
(Insurance Billing NOT Accepted; Patient Bill or Institutional Bill MUST be selected on page 1)
○ Known mtDNA Variant(s) Testing

Proband Name	Relationship to Proband	Proband GeneDx Accession #

If Proband Not Tested at GeneDx, SELECT ONE:
○ Family member test report included - Recommended if previous test was performed at another lab
○ Positive control included/will be sent - Recommended if previous test was performed at another lab
○ Positive control not available - Caveat language will be included on a negative report

VARIANT INFORMATION (please fill out the below information if family member report is not included) Number of Variants: _____

Gene	Coding DNA (c./m.)	Amino Acid (p.)	Transcript (NM#)
Gene	Coding DNA (c./m.)	Amino Acid (p.)	Transcript (NM#)

COPY NUMBER VARIANTS (CNV(s) require coordinates and genome build or transcript # and exon #) Number of Variants: _____

Gene(s)	Exon #	Coordinates	Genome Build
Gene(s)	Exon #	Coordinates	Genome Build

TESTING OPTIONS

CUSTOM DEL/DUP TESTING

○ 906 Deletion/Duplication Analysis of ONE Nuclear Gene
○ 703 Deletion/Duplication Analysis of 2-20 Nuclear Genes

Write-in Desired Gene(s) to be Tested: _____

FAMILY MEMBER TESTING (NO SEPARATE REPORT, ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS)

○ J767 Ataxia Xpanded®, Family Member Testing
○ 954 Autism/ID Xpanded®, Family Member Testing
○ T997 Cerebral Palsy Xpanded®, Family Member Testing
○ TG86 Congenital Hypotonia Xpanded®, Family Member Testing
○ 923 Epi Xpanded®, Family Member Testing
○ 725 Chromosomal Microarray Parental Testing
○ J854 Leukodystrophy Xpanded®, Family Member Testing
○ J513 Microcephaly Xpanded®, Family Member Testing
○ J820 Mito Xpanded®, Family Member Testing
○ TK80 Xpanded® Adult Movement Disorders Panel, Family Member Testing

	First Name	Last Name	DOB	
Mother				○ Asymptomatic ○ Symptomatic ○ Not Available ○ To be Sent Later**
Father				○ Asymptomatic ○ Symptomatic ○ Not Available ○ To be Sent Later**
Other	Relationship to Proband			
				○ Asymptomatic ○ Symptomatic ○ Not Available ○ To be Sent Later**

>> See next page for proband test selection

****Family member samples MUST BE RECEIVED WITHIN 3 WEEKS for inclusion in the proband's test.**

Write-in Test Selection: ○ Test Code: _____ ○ Test Name: _____

HISTORY

FAMILY HISTORY: ○ No Known Family History ○ Pedigree Attached ○ Adopted

Relationship	Maternal	Paternal	Relevant History	Age at Dx
1	○	○		
2	○	○		
3	○	○		

TESTING HISTORY: ○ Test Report Included (recommended)

Other relevant results (clinical, laboratory/biochemical or research): _____

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TEST MENU

NEURODEVELOPMENTAL DISORDERS AND EPILEPSY

- ○ 522 Fragile X Syndrome (*FMR1* repeat analysis)
- ○ 910 Chromosomal Microarray
- ○ T395 Autism/ID Panel (seq & del/dup of 103 genes)

Order of Reflex Testing:

- ○ Concurrent analysis of 522 & 910, if non-diagnostic activate T395
- ○ Start with 522, if non-diagnostic activate 910, if non-diagnostic activate T395

- ○ 952 Autism/ID Xpanded® Panel (2600+ genes, trios preferred)
- ○ 195 *PTEN*-Related Disorders (*PTEN* seq & del/dup)
- ○ 729 Rett/Angelman Related Disorders Panel (seq & del/dup of 25 genes & methylation MLPA)
- ○ 549 Rett/Atypical Rett Syndromes (*MECP2* seq & del/dup)
- ○ TJ27 Angelman Syndrome/Prader-Willi Syndrome Methylation MLPA (UPD, deletion)
- ○ 546 Angelman Syndrome (*UBE3A* seq & del/dup)
- ○ 523 Comprehensive Epilepsy Panel (seq & del/dup of 144 genes)
  - ○ 814 STAT Epilepsy Panel (seq & del/dup of 27 genes)
  - ○ 541 Infantile Epilepsy Panel (seq & del/dup of 126 genes)
  - ○ 542 Childhood-Onset Epilepsy Panel (seq & del/dup of 87 genes)
- ○ 921 Epi Xpanded® Panel (1300+ genes, trios preferred)
- ○ 953 Epilepsy Del/Dup Panel (128 genes) (not a trio based test)
- ○ 651 PRRT2 Sequencing
- ○ T400 Hemiplegic Migraine Panel (seq & del/dup of 4 genes)
- ○ 730 Tuberous Sclerosis Panel (*TSC1* & *TSC2* seq & del/dup)

CNS MALFORMATIONS AND DISORDERS

- ○ 691 Comprehensive Brain Malformations Panel (seq & del/dup of 103 genes)
  - ○ 698 Cortical Brain Malformations Panel (seq & del/dup of 61 genes)
  - ○ 700 Pontocerebellar Hypoplasia Panel (seq & del/dup of 19 genes)
  - ○ 701 Joubert Syndrome and Related Disorders Panel (seq & del/dup of 29 genes)
  - ○ 946 Lissencephaly Panel (seq & del/dup of 26 genes)
- ○ 689 Microcephaly Panel (seq & del/dup of 65 genes)
- ○ J511 Microcephaly Xpanded® Panel (800+ genes, trios preferred)
- ○ 699 Syndromic Macrocephaly/Overgrowth Syndromes Panel (seq & del/dup of 29 genes)
- ○ J853 Leukodystrophy Xpanded® Panel (300+ genes, trios preferred)
- ○ 552 X-linked Hydrocephalus/X-linked Spastic Paraplegia/MASA/CRASH Syndrome (LICAM seq & del/dup)
- ○ TB51 Comprehensive Holoprosencephaly Panel (seq & del/dup of 17 genes)
  - ○ 2371 Holoprosencephaly (*SHH, ZIC2, SIX3,TGIF* seq & del/dup)
- ○ 526 Cerebral Cavernous Malformations (*KRIT1, CCM2, PDCD10* seq & del/dup)
- ○ T844 Dementia Panel (seq only of 11 genes, for patients 18 years and older)

MOVEMENT DISORDERS

- ○ 941 Comprehensive Hereditary Spastic Paraplegia Panel (seq & del/dup of 83 genes)
- ○ T851 Cerebral Palsy Xpanded® Panel (1100+ genes, trios preferred)
- ○ TH97 Dentatorubral-Pallidoluysian Atrophy Repeat Analysis (*ATN1* repeat)
- ○ TH95 Friedreich Ataxia Repeat Analysis (*FXN* repeat)
- ○ TH94 Friedreich Ataxia Sequencing & Del/Dup (*FXN* sanger seq & del/dup)
- ○ J762 Ataxia Xpanded® Panel (1300+ genes, trios preferred)
- ○ 218 Alexander Disease (*GFAP* seq)
- ○ 581 Niemann-Pick C Disease (*NPC1, NPC2* seq & del/dup)
- ○ 527 Dopa-Responsive Dystonia (*GCH1* seq & del/dup)
- ○ TA78 Dopa-Responsive Dystonia/Infantile Parkinsonism/*TH* Deficiency (*TH* seq & del/dup)
- ○ T402 Dystonia and Parkinsonism Panel (seq & del/dup of 103 genes)
  - ○ T403 Dystonia Panel (seq & del/dup of 83 genes)
  - ○ T401 Parkinson Disease Panel (seq & del/dup of 44 genes)
- ○ TH83 Spinocerebellar Ataxia Repeat Expansion Analysis (*ATXN1, ATXN2, ATXN3, ATXN7, ATXN8, CACNA1A* repeat)
  - ○ TH84 Spinocerebellar Ataxia Type 1 Repeat Analysis (*ATXN1* repeat)
  - ○ TH85 Spinocerebellar Ataxia Type 2 Repeat Analysis (*ATXN2* repeat)
  - ○ TH86 Spinocerebellar Ataxia Type 3 Repeat Analysis (*ATXN3* repeat)
  - ○ TH88 Spinocerebellar Ataxia Type 7 Repeat Analysis (*ATXN7* repeat)
  - ○ TH89 Spinocerebellar Ataxia Type 8 Repeat Analysis (*ATXN8* repeat)
  - ○ TH87 Spinocerebellar Ataxia Type 6 Repeat Analysis (*CACNA1A* repeat)
- ○ TK79 Xpanded® Adult Movement Disorders Panel (500+ genes, trio preferred)
- ○ TL12-9 Spinocerebellar Ataxia and Related Disorders Panel (seq & del/dup of 56 genes)

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TEST MENU *(Continued)*

NEUROMUSCULAR DISORDERS

- ○ 737 Hereditary Neuropathy Panel (seq & del/dup of 89 genes)
  - ○ J778 CMT Panel (seq & del/dup of 54 genes)
  - ○ T399 Hereditary Sensory and Autonomic Neuropathy Panel (seq del/dup of 16 genes)
- ○ 742 *CMT1A/HNPP* (PMP22 del/dup)
- ○ 888 *HNPP/CMT1E* (PMP22 seq)
- ○ TB12 Erythermalgia/Paroxysmal Extreme Pain Disorder/Small Fiber Neuropathy/Congenital Insensitivity to Pain (*SCN9A* seq & del/dup)
- ○ 363 Familial Amyloid Polyneuropathy (TTR seq)
- ○ 820 Spinal & Bulbar Muscular Atrophy (AR repeat analysis)
- ○ 889 Neuromuscular Disorders Panel (115 genes)
  - ○ 890 Limb-Girdle Muscular Dystrophy Panel (seq & del/dup of 33 genes)
  - ○ 892 Congenital Myopathy & Muscular Dystrophy Panel (41 genes)
- ○ TG81 Periodic Paralysis Panel (seq & del/dup of 14 genes)
- ○ 787 Duchenne/Becker MD (*DMD* del/dup)
- ○ 786 Duchenne/Becker MD (*DMD* seq)
- ○ TG80 Arthrogryposis Panel (seq & del/dup of 90 genes)
  - ○ TG79 Distal Arthrogryposis Panel (seq & del/dup of 12 genes)
  - ○ TG76 Focused Arthrogryposis (seq & del/dup of 14 genes)
  - ○ 945 Congenital Myasthenia Syndromes Panel (seq & del/dup of 19 genes)
- ○ TG85 Prenatal Akinesia/Arthrogryposis (seq & del/dup of 27 genes)
- ○ TG78 Congenital Hypotonia Evaluation (*SMN1, SMN2, DMPK*, 15q11.2-q13.1)
- ○ TG77 Congenital Hypotonia Xpanded® Panel (1400+ genes; trios preferred)
- ○ T406 Spinal Muscular Atrophy Panel (seq & del/dup of 26 genes plus *SMN1/2* Dosage Analysis)
- ○ T789 *SMN1/2* Dosage Analysis
- ○ TG82 Myotonia Panel (*CNBP* and *DMPK* repeat analysis, seq & del/dup of 8 genes)
- ○ 818 Myotonic Dystrophy 1 (DM1) (*DMPK* repeat analysis)
- ○ 819 Myotonic Dystrophy 2 (DM2) (*CNBP* repeat analysis)
- ○ 743 Oculopharyngeal Muscular Dystrophy (*PABPN1* repeat analysis)
- ○ T815 Juvenile ALS Panel (seq & del/dup of 16 genes)
- ○ J805 Amyotrophic Lateral Sclerosis/Frontotemporal Lobar Degeneration (C9orf72 repeat analysis, for patients 18 years and older)
- ○ T404 Amyotrophic Lateral Sclerosis/Frontotemporal Lobar Degeneration Panel (seq & del/ dup of 24 genes, for patients 18 years and older)

Order of Reflex Testing:

- ○ Activate J805, if non-diagnostic activate T404

MITOCHONDRIAL DISORDERS

- ○ J809 Mito Xpanded® Panel (1800+ genes, trios preferred)
- ○ 554 Concurrent full sequence analysis & deletion testing of the mito genome (not a trio based test)
- ○ 554 Full sequence analysis and deletion testing of the mitochondrial genome
- ○ 704 mtDNA Point Variants Plus Large Deletions Panel
- ○ TH12 Leber Hereditary Optic Neuropathy (LHON) Panel
- ○ TB60 Deletion analysis of mito genome
- ○ 394 *POLG* gene sequencing & del/dup
- ○ 615 Combined Mito Genome Plus Mito Focused Nuclear Gene Panel (seq & del/dup of mito genome and 202 nuclear genes)
- ○ 573 Mitochondrial Focused Nuclear Gene Panel (seq & del/dup of 202 genes)
- ○ 575 Mitochondrial Encephalopathy/Leigh Syndrome Nuclear Gene Panel (seq & del/dup of 134 genes)
- ○ 576 Lactic Acidosis/Pyruvate Metabolism Nuclear Gene Panel (seq & del/dup of 130 genes)
- ○ 577 Progressive External Ophthalmoplegia (PEO)/Optic Atrophy Nuclear Gene Panel (seq & del/dup of 44 genes)
- ○ 578 Methylglutaconic Acidura Nuclear Panel (seq & del/dup of 14 genes)

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TEST MENU *(Continued)*

NEUROMETABOLIC DISORDERS

- ○ J979 Combined Lysosomal and Peroxisomal Disorders Panel (seq & del/dup of 82 genes)
  - ○ T013 Lysosomal Disorders Panel (seq & del/dup of 57 genes)
  - ○ J978 Peroxisomal Disorders Panel (seq & del/dup of 25 genes)
- ○ J977 Congenital Disorders of Glycosylation Panel (seq & del/dup of 108 genes)
- ○ J976 Creatine Deficiency Syndromes Panel (seq & del/dup of 3 genes)
- ○ J995 Disorders of Hyperphenylalanimenia and Biopterin Metabolism Panel (seq & del/dup of 7 genes)
- ○ T382 Fatty Acid Oxidation Disorders Panel (seq & del/dup of 15 genes)
- ○ T010 Hyperammonemia, Urea Cycle and Transporter Defects Panel (seq & del/dup of 48 genes)
- ○ T012 Metabolic Myopathy Panel (seq & del/dup of 30 genes)
- ○ T011 Methylmalonic Acidemia, Disorders of Cobalamin Metabolism and Related Disorders Panel (seq & del/dup of 19 genes)
- ○ J981 Riboflavin Transporter Deficiency and Related Disorders (seq & del/dup of 9 genes)
- ○ 334 Carnitine Palmitoyltransferase II Deficiency (*CPT2* seq & del/dup)
- ○ 2321 Fabry Disease (*GLA* seq & del/dup)
- ○ TG94 Gaucher Disease (*GBA* seq)
- ○ 507 Krabbe Disease (*GALC* seq & del/dup)
- ○ TH08 Pompe Disease/Glycogen Storage Disease Type II (*GAA* seq and del/dup)
- ○ TG92 Wilson Disease (*ATP7B* seq & del/dup)
- ○ J975 X-linked Adrenoleukodystrophy (*ABCD1* seq & del/dup)

NEUROFIBROMATOSIS

- ○ 961 Comprehensive NF Panel: *NF1, SPRED1, NF2* and *SMARCB1* sequencing and deletion/duplication testing
- ○ 962 NF1 Panel: *NF1* and *SPRED1* sequencing and deletion/duplication testing
- ○ TA06 Reflex to Noonan Syndrome and RASopathies panel (sequencing of 25 genes) if 962 is non-diagnostic
- ○ 963 NF2 Panel: *LZTR1, NF2* and *SMARCB1* sequencing and deletion/duplication testing

GeneDx tests are frequently updated and improved based upon the most recent scientific evidence. The test codes, genes, and gene quantities listed on this test requisition are subject to change by GeneDx at any time. The most current test menu and list of genes included for a specific test panel may be found on our website, genedx.com. Please note that GeneDx reserves the right to modify and upgrade any ordered panel to the version currently listed on our website.

INFORMED CONSENT

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

For the purposes of this consent, "I", "my", and "your" will refer to me or to my child, including my unborn child, if my child is the person for whom the healthcare provider has ordered testing.

PURPOSE OF THIS TEST

The purpose of this test is (a) to see if I may have a genetic variant or chromosome rearrangement causing a genetic disorder; or (b) to evaluate the chance that I will develop or pass on a genetic disorder in the future. If I already know the specific gene variant(s) or chromosome rearrangement that causes the genetic disorder in my family, I agree to inform the laboratory of this information.

WHAT TYPE OF TEST RESULTS CAN I EXPECT FROM GENETIC TESTING?

1. Positive: A change in your DNA was found, which is very likely the cause of your features/symptoms. This is the most straightforward test result, which can be used as the basis to test other family members to determine their chances of having either the disease or a child with the disease.
2. Negative: No variants were found to explain your symptoms. This does not mean that you do not have a genetic condition. It is still possible that there is a genetic variant not found by the test that was ordered. Your healthcare provider or genetic counselor may discuss more testing either now or in the future.
3. Variant of Uncertain Significance (VUS): A change in a gene was found. However, we are not sure whether this variant is the cause of your symptoms/features. More information is needed. We may suggest testing other family members to help figure out the meaning of the test result.
4. Unexpected Results: In rare instances, this test may reveal an important genetic change that is not directly related to the reason for ordering this test. For example, this test may find you are at risk for another genetic condition I am not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. We may disclose this information to the ordering healthcare provider if it likely affects medical care.

Because medical and scientific knowledge is constantly changing, new information that becomes available may supplement the information GeneDx used to interpret my results. Healthcare providers can contact GeneDx at any time to discuss the classification of an identified variant.

WHAT IS TRIO/DUO-BASED GENETIC TESTING?

For some genetic tests, including samples from the biological parents and/or other biological relatives along with the patient's sample can help with the interpretation of the test results. These tests are often referred to as "trio tests" since they typically include samples from the patient and both parents.
Samples from relatives should be submitted with the patient's sample. Clinical information must be provided for the patient and any relative who submits a sample.
I understand that GeneDx will use the relative sample(s) when needed for the interpretation of my test results and that my test report may include clinical and genetic information about a relative when it is relevant to the interpretation of the test results. I further understand that relatives will not receive an independent analysis of data nor a separate report.

RISKS AND LIMITATIONS OF GENETIC TESTING

1. In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology.
2. Accurate interpretation of test results may require knowing the true biological relationships in a family. I understand that if I fail to accurately state the biological relationships in my family, it could lead to incorrect interpretation of the test results, incorrect diagnoses, and/or inconclusive test results. If genetic testing reveals that the true biological relationships in a family are not as I reported them, including non-paternity (the reported father is not the biological father) and consanguinity (the parents are related by blood), I agree to have these findings reported to the healthcare provider who ordered the test.
3. Although genetic testing is highly accurate, inaccurate results may occur. These reasons include, but are not limited to mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or other reasons.
4. I understand that this test may not detect all of the long-term medical risks that I might experience. The result of this test does not guarantee my health and that additional diagnostic tests may still need to be done.
5. I agree to provide an additional sample if the initial sample is not adequate.

PATIENT CONFIDENTIALITY AND GENETIC COUNSELING

It is recommended that I receive genetic counseling before and after having this genetic test. I can find a genetic counselor in my area at *www.nsgc.org*. Further testing or additional consultations with a healthcare provider may be necessary.
To maintain confidentiality, test results will only be released to the referring healthcare provider, the ordering laboratory, to me, to other healthcare providers involved in my care, diagnosis and treatment, or to others with my consent or as permitted or required by law. Federal laws prohibit unauthorized disclosure of this information. More information can be found at: *www.genome.gov/10002077*

INTERNATIONAL SAMPLES

If I reside outside the United States, I attest that by providing a sample for testing, I am not knowingly violating any export ban or other legal restriction in the country of my residence.

SAMPLE RETENTION

After testing is complete, my sample may be de-identified and be used for test development and improvement, internal validation, quality assurance, and training purposes. GeneDx will not return DNA samples to you or to referring healthcare providers, unless specific prior arrangements have been made.
I understand that samples from residents of New York State will not be included in the de-identified research studies described in this authorization and GeneDx will not retain them for more than 60 days after test completion, unless specifically authorized by my selection. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language. GeneDx will not perform any tests on the biological sample other than those specifically authorized.

DATABASE PARTICIPATION

De-identified health history and genetic information can help healthcare providers and scientists understand how genes affect human health. Sharing this de-identified information helps healthcare providers to provide better care for their patients and researchers to make new discoveries. GeneDx shares this type of information with healthcare providers, scientists, and healthcare databases. GeneDx will not share any personally identifying information and will replace the identifying information with a unique code not derived from any personally identifying information. Even with a unique code, there is a risk that I could be identified based on the genetic and health information that is shared. GeneDx believes that this is unlikely, though the risk is greater if I have already shared my genetic or health information with public resources, such as genealogy websites.

INFORMED CONSENT

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

EXOME/GENOME SEQUENCING SECONDARY FINDINGS

- Applicable Only for Full Exome Sequencing and Genome Sequencing Tests.
- Does not pertain to Xpanded® or Slice tests

As many different genes and conditions are analyzed in an exome or genome sequencing test, these tests may reveal some findings not directly related to the reason for ordering the test. Such findings are called "incidental" or "secondary" and can provide information that was not anticipated.

Secondary findings are variants, identified by an exome or genome sequencing test, in genes that are unrelated to the individual's reported clinical features.

The American College of Medical Genetics and Genomics (ACMG) has recommended that secondary findings identified in a specific subset of medically actionable genes associated with various inherited disorders be reported for all probands undergoing exome or genome sequencing. Please refer to the latest version of the ACMG recommendations for reporting of secondary findings in clinical exome and genome sequencing for complete details of the genes and associated genetic disorders. Reportable secondary findings will be confirmed by an alternate test method when needed.

WHAT WILL BE REPORTED FOR THE PATIENT?
All pathogenic and likely pathogenic variants associated with specific genotypes identified in the genes (for which a minimum of 10X coverage was achieved by exome sequencing or a minimum of 15X coverage was achieved by genome sequencing), as recommended by the ACMG.

WHAT WILL BE REPORTED FOR RELATIVES?
The presence or absence of any secondary finding(s) reported for the proband will be provided for all relatives analyzed by an exome or genome sequencing test.

LIMITATIONS
Pathogenic and/or likely pathogenic variants may be present in a portion of the gene not covered by this test and therefore are not reported. The absence of reportable secondary findings for any particular gene does not mean there are no pathogenic and/or likely pathogenic variants in that gene. Pathogenic variants and/or likely pathogenic variants that may be present in a relative, but are not present in the proband, will not be identified, or reported. Only changes at the sequence level will be reported in the secondary findings report. Larger deletions/duplications, abnormal methylation, triplet repeat or other expansion variants, or other variants not routinely identified by clinical exome and genome sequencing will not be reported.

FINANCIAL AGREEMENT AND GUARANTEE
For insurance billing, I understand and authorize GeneDx to bill my health insurance plan on my behalf, to release any information required for billing, and to be my designated representative for purposes of appealing any denial of benefits. I irrevocably assign to and direct that payment be made directly to GeneDx.
I understand that my out-of-pocket costs may be different than the estimated amount indicated to me by GeneDx as part of a benefit investigation. I agree to be financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. If my insurance provider sends a payment directly to me for services performed by GeneDx on my behalf, I agree to endorse the insurance check and forward it to GeneDx within 30 days of receipt as payment towards GeneDx's claim for services rendered.
If I do not have health insurance, I agree to pay for the full cost of the genetic testing that was ordered by my healthcare provider and billed to me by GeneDx. I further understand and agree that, if I fail to make payment for genetic testing, in accordance with the payment policies of GeneDx, my account may be turned over to an external collection agency for non-payment. I agree to pay any associated collection costs, including attorney fees. By my signature on the GeneDx Test Requisition Form or at the bottom of this form, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider.

MEDICARE
A completed Advance Beneficiary Notice (ABN) is required for Medicare patients. Please visit our website, *www.genedx.com/billing* for more information.

DIGITAL PATIENT LETTER CONSENT

- Applicable Only for Commercial Insurance
- Estimate is provided by your health insurance company and therefore NO estimate will be sent for any orders placed with federal or state-funded insurance plans (e.g. Medicare, Medicaid, Tricare, etc.), institutional bill, or patient bill (self-pay).

To provide you with the estimated out-of-pocket expenses related to your test, GeneDx will send you an email and/or text with the link to access your personalized Digital Patient Letter. In order to send this information, we need your consent and agreement to the following items:

1. GeneDx can use your email address or mobile phone number solely for the purpose of GeneDx sending your estimated financial obligation. Text message data rates may apply. GeneDx is not responsible for undelivered messages due to incorrect or illegible contact information.
2. GeneDx will send you an email and/or text message containing a link to view your personalized Patient Letter that includes the test out-of-pocket estimate. The link is time-sensitive and will only be available for 72 hours from the time the message is sent. In order to view the estimate, you must click the link in the message.
3. If you take no action, GeneDx will assume that you agree to move ahead with testing and will bill your health insurance. You can approve testing with insurance, switch to self-pay, or cancel the test via the link within the given 72-hour window. In turn, if GeneDx receives your sample(s) and the billing method hasn't been changed, or the test hasn't been cancelled, we will move ahead with testing as ordered, and you will be responsible for any out-of-pocket costs for the completion of the test(s).