NEUROLOGY TEST REQUISITION FORM

GeneDx an OPKO Health Company

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 CONSENT FOR GENETIC TESTING, FINANCIAL AGREEMENT AND GUARANTEE:

By signing this form, I acknowledge as the patient or relative being tested that I have read the GeneDx Informed Consent document available from my healthcare provider or at genedx.com/forms, and I authorize GeneDx to perform genetic testing as ordered. I understand that, for tests that evaluate data from multiple family members concurrently, 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. By my signature below, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider. 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 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 and agree to be financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. I am aware that my insurance provider may send 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. 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 and I agree to pay any associated collection costs, including attorney fees.

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

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

○ 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 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 the studies listed above.

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

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

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 NECESSISTY

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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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 (heteroplasmy detection range 25%-100%)
○ Known mtDNA Variant(s) Testing by NGS (heteroplasmy detection range 1.5%-100%) - Test Code 453
○ Known mtDNA Variant(s) Testing by NGS - URINE (heteroplasmy detection range 5-100%) - Test Code T822

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
○ 910 Chromosomal Microarray Parental Testing
○ J854 Leukodystrophy Xpanded®, Family Member Testing
○ J513 Microcephaly Xpanded®, Family Member Testing
○ J820 Mito Xpanded®, Family Member Testing

	First Name	Last Name	DOB		
Mother	First Name	Last Name	DOB	○ Asymptomatic ○ Not Available	○ Symptomatic ○ To be Sent Later**
Father	First Name	Last Name	DOB	○ Asymptomatic ○ Not Available	○ Symptomatic ○ To be Sent Later**
Other	Relationship to Proband				
	First Name	Last Name	DOB	○ Asymptomatic ○ Not Available	○ Symptomatic ○ 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	Relavant 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 (2300+ genes, trios preferred)
- ○ 195 *PTEN*-Related Disorders (*PTEN* seq & del/dup)
- ○ 729 Rett/Angelman Related Disorders Panel (seq & del/dup of 20 genes)
- ○ 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 127 genes)
  - ○ 814 STAT Epilepsy Panel (seq & del/dup of 26 genes)
  - ○ 541 Infantile Epilepsy Panel (seq & del/dup of 111 genes)
  - ○ 542 Childhood-Onset Epilepsy Panel (seq & del/dup of 84 genes)
  - ○ 544 Progressive Myoclonic Epilepsy Panel (seq & del/dup of 18 genes)
  - ○ 545 Rest of the Comprehensive Epilepsy Panel (if subpanel non-diagnostic)
- ○ 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)
  - ○ 722 Rest of the Brain Malformations Panel (if subpanel non-diagnostic)
- ○ 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 42 genes)
  - ○ 942 Uncomplicated Hereditary Spastic Paraplegia Panel (seq & del/dup of 14 genes)
  - ○ 943 Rest of Comprehensive Hereditary Spastic Paraplegia Panel (if subpanel non-diagnostic)
- ○ 944 Hereditary Spastic Paraplegia Related Inborn Error of Metabolism Panel (seq & del/dup of 15 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)
- ○ 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 73 genes)
  - ○ T403 Dystonia Panel (seq & del/dup of 53 genes)
  - ○ T401 Parkinson Disease Panel (seq & del/dup of 29 genes)
  - ○ T919 Rest of Dystonia and Parkinsonism Panel (if subpanel non-diagnostic)
- ○ 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)

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TEST MENU *(Continued)*

NEUROMUSCULAR DISORDERS

- ○ 737 Hereditary Neuropathy Panel (seq & del/dup of 64 genes)
  - ○ 884 Core CMT Panel (seq & del/dup of 6 genes)
  - ○ 885 Axonal CMT Panel (seq & del/dup of 32 genes)
  - ○ 886 Demyelinating CMT Panel (seq & del/dup of 23 genes)
  - ○ J778 CMT Panel (seq & del/dup of 43 genes)
  - ○ T399 Hereditary Sensory and Autonomic Neuropathy Panel (seq del/dup of 14 genes)
  - ○ 887 Rest of the Hereditary Neuropathy Panel (if subpanel non-diagnostic)
- ○ 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 (seq & del/dup of 99 genes)
  - ○ 890 Limb-Girdle Muscular Dystrophy Panel (seq & del/dup of 30 genes)
  - ○ 891 Syndromic Congenital Muscular Dystrophy Panel (seq & del/dup of 19 genes)
  - ○ 892 Congenital Myopathy & Muscular Dystrophy Panel (seq & del/dup of 34 genes)
  - ○ 893 Myofibrillar Myopathy Panel (seq & del/dup of 8 genes)
  - ○ TG81 Periodic Paralysis Panel (seq & del/dup of 14 genes)
  - ○ 894 Rest of Neuromuscular Disorders Panel (if subpanel non-diagnostic)
- ○ 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 18 genes)
  - ○ TG84 Rest of Arthrogryposis Panel (if subpanel non-diagnostic)
- ○ TG85 Prenatal Akinesia/Arthrogryposis (seq & del/dup of 28 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 18 genes plus *SMN1/2* Dosage Analysis)
- ○ T789 *SMN1/2* Dosage Analysis
- ○ TG82 Myotonia Panel
- ○ 818 Myotonic Dystrophy 1 (DM1) (*DMPK* repeat analysis)
  - ○ 900* Reflex to DM1 Southern Blot, if 818 is positive (blood sample is required)
- ○ 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

** Samples from New York state cannot be accepted for the Southern Blot test.*

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
- ○ 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)
- ○ 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

INFORMED CONSENT

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

General Information About Genetic Testing

What is genetic testing?

DNA provides instructions for our body's growth and development. Genes are distinct sequences of DNA, and are arranged on chromosomes. The DNA in a gene contains instructions for making proteins, which determine things like growth and metabolism as well as traits like eye color and blood type. Genetic disorders are caused by certain changes in DNA affecting the structure or number of chromosomes. Genetic testing is a laboratory test that tries to identify these changes in chromosomes or the DNA. Genetic testing can be a diagnostic test, which is used to identify or rule out a specific genetic condition. Genetic screening tests are used to assess the chance for a person to develop or have a child with a genetic condition. Genetic screening tests are not typically diagnostic and results may require additional testing.

The purpose of this test is to see if I, or my child, may have a genetic variant or chromosome rearrangement causing a genetic disorder or to determine the chance that I, or my child, will develop or pass on a genetic disorder in the future. 'My child' can also mean my unborn child, for the purposes of this consent.

If I/my child already know the specific gene variant(s) or chromosome rearrangement that causes the genetic disorder in my family, I will inform the laboratory of this information.

What could I learn from this genetic test?

The following describes the possible results from the test:

1) Positive: A positive result indicates that a genetic variant has been identified that explains the cause of my/my child's genetic disorder or indicates that I/my child am at increased risk to develop the disorder in the future. It is possible to test positive for more than one genetic variant.

2) Negative: A negative result indicates that no disease-causing genetic variant was identified by the test performed. It does not guarantee that I/my child will be healthy or free from genetic disorders or medical conditions. If I/my child test negative for a variant known to cause the genetic disorder in other members of my/my child's family, this result rules out a diagnosis of the same genetic disorder in me/my child due to this specific change.

3) Inconclusive/Variant of Uncertain Significance (VUS): A finding of a variant of uncertain significance indicates that a genetic change was detected, but it is currently unknown whether that change is associated with a genetic disorder either now or in the future. A variant of uncertain significance is not the same as a positive result and does not clarify whether I/my child is at increased risk to develop a genetic disorder. The change could be a normal genetic variant or it could be disease-causing. Further analysis may be recommended, including testing parents and other family members. Detailed medical records or information from other family members also may be needed to help clarify results.

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 tell me about the risk for another genetic condition I/my child is not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. This information may be disclosed to the ordering health care provider if it likely impacts medical care.

Result interpretation is based on currently available information in the medical literature, research and scientific databases. Because the literature, medical and scientific knowledge are constantly changing, new information that becomes available in the future may replace or add to the information GeneDx used to interpret my/my child's results. Providers can contact GeneDx at any time to discuss the classification of an identified variant. In addition, I or my/my child's health care providers may monitor publicly available resources used by the medical community, such as ClinVar (www.clinvar.com), to find current information about the clinical interpretation of my/my child's variant(s).

For tests that evaluate data from multiple family members, my spouse, or partner concurrently, results may be included in a single comprehensive report.

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 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/my child's test results. The patient report may include clinical and genetic information about a relative when it is relevant to the interpretation of the results. Relatives do not receive an independent analysis of data nor a separate report.

What are the risks and limitations of this genetic test?

- Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer. 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.
- Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in my/my child's family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results. In some cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. This includes non-paternity (the stated father of an individual is not the biological father) and consanguinity (the parents of an individual are related by blood). It may be necessary to report these findings to the health care provider who ordered the test.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to: mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, or the presence of change(s) in such a small percentage of cells that the change(s) may not be detectable by the test (mosaicism).
- This test does not have the ability to detect all of the long-term medical risks that I/my child might experience. The result of this test does not guarantee my health or the health of my child/fetus. Other diagnostic tests may still need to be done, especially when only a genetic screening test has been performed previously.
- Occasionally, an additional sample may be needed if the initial specimen 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 here: www.nsgc.org. Further testing or additional consultations with a health care provider may be necessary.

To maintain confidentiality, the test results will only be released to the referring health care provider, to the ordering laboratory, to me, to other health care providers involved in my/my child's diagnosis and treatment, or to others as entitled by law. The United States Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, I understand that I can visit www.genome.gov/10002077.

International Specimens

If I/my child 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/my child's residence.

Additional information about the specific test being ordered is available from my health care provider or I can go to the GeneDx website, www.genedx.com.This information includes the complete gene lists, the specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, the limitations of genetic testing, as well as information about how specimens and information are stored and used.

Specimen Retention

After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring heath care 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 will not be retained 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. No tests other than those authorized shall be performed on the biological sample.

Database Participation

De-identified heath history and genetic information can help health care providers and scientists understand how genes affect human health. Though {I/my child} may not personally benefit, sharing this information helps health care providers to provide better care for their patients and researchers to make discoveries. GeneDx shares this type of information with health care providers, scientists and health care databases. No personal identifying information will be shared, as it will be replaced with a unique code.

Even though only a code is used for the reporting to the database, there is a risk that {I/my child} 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/my child's} genetic or health information with public resources, such as genealogy websites.

Recontact for Research Participation

Separate from the above, GeneDx may collaborate with scientists, researchers and drug developers to advance knowledge of genetic diseases and to develop new treatments. If there are opportunities to participate in research relevant to the disorder in {my/my child's} family, and if I have consented for recontact, GeneDx may allow my healthcare provider to be recontacted for research purposes, such as the development of new testing, drug development, or other treatment modalities. In some situations, such as if my health care provider is not available, I may be contacted directly.

Any research that results in medical advances, including new products, tests or discoveries, may have potential commercial value and may be developed and owned by GeneDx or the collaborating researchers. If any individuals or corporations benefit financially from these studies, no compensation will be provided to {my/my child} or {my/my child's} heirs.