Neurology Test Requisition Form

GeneDx an OPKO Health Company

Patient Information

First name Last name
Gender ❒ Male ❒ Female Date of birth (mm/dd/yy)_____
Ancestry ❒ Caucasian ❒ Eastern European ❒ Northern European
❒ Western European ❒ Native American ❒ Middle Eastern
❒ African American ❒ Asian ❒ Pacific Islander
❒ Caribbean ❒ Central/South American
❒ Ashkenazi Jewish ❒ Hispanic ❒ Other: _____

Mailing address

City State Zip code

Home phone Work phone

Email Patient's primary language if not English

Ordering Account Information

Acct # Account Name
Reporting Preference*. ❒ Care Evolve ❒ Fax ❒ Email
**If unmarked, we will use the account's default preferences or fax to new clients.*

Physician NPI #

Genetic Counselor

Street address 1

Street address 2

City State Zip code

Phone Fax (important)

Email Beeper

Send Additional Report Copies To:

Physician or GC/Acct # Fax#/Email/CE #

Physician or GC/Acct # Fax#/Email/CE #

Sample Information

Medical record # Specimen ID Date sample obtained (mm/dd/yy)
❒ Blood in EDTA (5-6 mL in lavender top tube)
❒ DNA (>20 ug): Tissue source _____ concentration ___ (ug/ml) Vol ___(ul)
❒ Oral Rinse (At least 30 mL of Scope oral rinse in a 50 mL centrifuge tube)
❒ Dried Blood Spots (2 cards) - Not accepted for any testing with a del/dup component
❒ Buccal Swab
❒ Other _____(Call lab)
Patient has had a blood transfusion ❒ Yes ❒ No Date of last transfusion __/__/__
(2-4 weeks of wait time is required for mtDNA testing only) Specimens are not accepted for patients who have had allogeneic bone marrow transplants
Clinical Diagnosis: _____ **ICD-10 Codes:** _____
Age at Initial Presentation: ____ **Add. ICD-10 Codes:** _____

Statement of Medical Necessity

This test is medically necessary for the diagnosis or detection of a disease, illness, impairment, symptom, syndrome or disorder. The results will determine my patient's medical management and treatment decisions. The person listed as the Ordering Physician is authorized by law to order the tests(s) requested herein. I confirm that I have provided genetic testing information to the patient and the patient has consented to genetic testing.

Medical Professional Signature (required) Date

Patient Consent (sign here or on the consent document)

I have read the Informed Consent document and I give permission to GeneDx to perform genetic testing as described. I also give permission for my specimen and clinical information to be used in de-identified studies at GeneDx to improve genetic testing and for publication, if appropriate. My name or other personal identifying information will not be used in or linked to the results of any studies and publications. I also give GeneDx permission to inform me or my health care provider in the future about research opportunities, including treatments for the condition in my family.

❒ Check this box if you wish to opt out of being contacted for research studies.
❒ Check this box if you are a New York state resident, and give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing.

Patient/Guardian Signature Date

Payment Options

❒ Insurance Bill

PATIENT STATUS – ONE MUST BE CHECKED ❒ Hospital Inpatient ❒ Hospital Outpatient ❒ Not a Hospital Patient

Referral/Prior Authorization # _____
Please attach copy of Referral/authorization

Insurance Carrier Policy Name ❒ Hold sample for Benefit Investigation (only if OOP cost is >$100) GeneDx Benefit Investigation #

Insurance ID # Group # Name of Insured Date of Birth Insurance Address City State Zip

Secondary Insurance Carrier Name Insurance ID# Group # Name of Insured Date of Birth
Relationship to Insured ❒ Child ❒ Spouse ❒ Self ❒ Other _____
Relationship to Insured ❒ Child ❒ Spouse ❒ Self ❒ Other _____

Please include a copy of the front and back of the patient's insurance card (include secondary when applicable)

If you would like to expedite an assessment of your possible eligibility for GeneDx's financial assistance program (FAP), please provide the number of your household members ____ and the annual income of your household $_____. GeneDx may require additional information from you to complete an application for GeneDx's financial assistance program.

I represent that I am covered by insurance and authorize GeneDx, Inc. to give my designated insurance carrier, health plan, or third party administrator (collectively "Plan") the information on this form and other information provided by my health care provider necessary for reimbursement. I authorize Plan benefits to be payable to GeneDx. I understand that GeneDx will attempt to contact me if my out-of-pocket responsibility will be greater than $100 per test (for any reason, including co-insurance and deductible, or non-covered services). If GeneDx is unsuccessful in its attempts to contact me, I understand that it will be my responsibility to contact GeneDx to determine my out-of-pocket cost and to pay my out-of-pocket responsibility. I will cooperate fully with GeneDx by providing all necessary documents needed for Plan billing and appeals. I understand that I am responsible for sending GeneDx any and all of the money that I receive directly from my Plan in payment for this test. Reasonable collection and/or attorney's fees, including filing and service fees, shall be assessed if the account is sent to collection but said fees shall not exceed those permitted by state law. I permit a copy of this authorization to be used in place of the original.

Patient Signature (required)_____ Date _____

❒ Institutional Bill

GeneDx Account #

Hospital/Lab Name

Contact Name

Address

City State Zip Code

Phone Fax

❒ Patient Bill

Amount _____

I understand that my credit card will be charged the full amount for the testing.

Please bill my credit card (all major cards accepted)

❒ MasterCard ❒ Visa ❒ Discover ❒ American Express

Name as it appears on card

Account Number Expiration date CVC

Signature Date

For GeneDx Use Only

Neurology Test Options

Account # Account Name

First Name Last Name Date of Birth (mm/dd/yy)

GeneDx Neurology Genetic Testing Menu

Neurodevelopmental/Epilepsy Disorders

- ❒ 522 Fragile X syndrome (FMR1 repeat analysis)
  - ❒ Reflex to 952 Autism/ID Xpanded Panel, if 522 negative
- ❒ 910 Chromosomal Microarray (GenomeDx)*
  - ❒ Reflex to 952 Autism/ID Xpanded Panel, if 910 negative
- ❒ 523 Epilepsy Comprehensive Panel (seq & del/dup of 87 genes)
  - ❒ 814 STAT Epilepsy Panel (seq & del/dup of 22 genes)
  - ❒ 541 Infantile Epilepsy Panel (seq & del/dup of 75 genes)
  - ❒ 542 Childhood-Onset Epilepsy Panel (seq & del/dup of 58 genes)
  - ❒ 544 Progressive Myoclonic Epilepsy Panel (seq & del/dup of 17 genes)
  - ❒ 729 Rett/Angelman Related Disorders Panel (seq & del/dup of 12 genes)
  - ❒ 730 Tuberous Sclerosis Panel (TSC1 & TSC2 seq & del/dup)
  - ❒ 545 Rest of the Comprehensive Epilepsy Panel (if subpanel negative)
- ❒ 691 Brain Malformations Comprehensive Panel (seq & del/dup of 93 genes)
  - ❒ 698 Cortical Brain Malformations Panel (seq & del/dup of 56 genes)
  - ❒ 700 Pontocerebellar Hypoplasia Panel (seq & del/dup of 18 genes)
  - ❒ 701 Joubert Syndrome and Related Disorders Panel (seq & del/dup of 25 genes)
  - ❒ 946 Lissencephaly Panel (seq & del/dup of 24 genes)
  - ❒ 722 Rest of the Brain Malformations Panel (if subpanel negative)
- ❒ 689 Microcephaly Panel (seq & del/dup of 28 genes)
- ❒ 699 Syndromic Macrocephaly/Overgrowth Syndromes Panel (seq & del/dup of 11 genes)
- ❒ 566 Angelman syndrome methylation-MLPA (UPD, deletion)
- ❒ 546 Angelman syndrome (UBE3A seq & del/dup)
- ❒ 375 Angelman-like/Christianson syndromes (SLC9A6 seq)
- ❒ 548 Atypical Rett/Infantile spasms/West syndromes (CDKL5 seq & del/dup)
- ❒ 294 Biotinidase Deficiency (BTD seq)
- ❒ 239 Congenital insensitivity to pain and anhidrosis (NTRK1 seq)
- ❒ 584 Cornelia de Lange syndrome (NIPBL, SMC1A seq & del/dup)
- ❒ 205 Gorlin syndrome (PTCH1 seq & del/dup)
- ❒ 651 Paroxysmal kinesigenic dyskinesia w/infantile convulsions (PRRT2 seq)
- ❒ 195 PTEN-related disorders (PTEN seq & del/dup)
- ❒ 549 Rett/Atypical Rett syndromes (MECP2 seq & del/dup)
- ❒ 406 Sotos Syndrome (NSD1 seq & del/dup)
- ❒ 607 Neuronal ceroid-lipofuscinosis 2 (CLN2) (TPP1 seq)

Xpanded Panels (Trio required)*

- ❒ 952 Autism/ID Xpanded Panel (2300+ genes)
- ❒ 921 EpiXpanded Panel (1300+ genes)
  - ❒ 953 Epilepsy Del/Dup Panel (95 genes)
  (If EpiXpanded panel negative, not a trio based test)
- ❒ J762 Ataxia Xpanded Panel (~1000 Genes)
- ❒ J511 Microcephaly Xpanded Panel (~800 genes)
- ❒ J809 MitoXpanded Panel (~1800 genes)
  - ❒ 554 Concurrent full sequence analysis & deletion testing of the mito genome (not a trio based test)
- ❒ J853 Leukodystrophy Xpanded Panel (~300 genes)

*Please contact GeneDx if samples are not available for both parents.

Xpanded Family Member Testing (no separate report)

- ❒ 923 EpiXpanded, Family Member Testing
- ❒ 954 Autism/ID Xpanded, Family Member Testing
- ❒ J767 Ataxia Xpanded, Family Member Testing
- ❒ J513 Microcephaly Xpanded, Family Member Testing
- ❒ J820 MitoXpanded, Family Member Testing
- ❒ J854 Leukodystrophy Xpanded, Family Member Testing

Biological Parent Sample Information

Mother: ❒ Not available ❒ To be sent later**

First name _____ Last name _____ DOB_____

❒ Asymptomatic ❒ Symptomatic

Father: ❒ Not available ❒ To be sent later**

First name _____ Last name _____ DOB_____

❒ Asymptomatic ❒ Symptomatic

Other: ❒ To be sent later** Relationship to Proband:_____

First name _____ Last name _____ DOB_____

**Additional samples must be received within 3 weeks

Neuromuscular Disorders

- ❒ 737 Hereditary Neuropathy Panel (seq & del/dup of 53 genes)
  - ❒ 742 CMT1A/HNPP (PMP22 del/dup)
  - ❒ 888 HNPP/CMT1E (PMP22 seq) (if del/dup negative)
  - ❒ 884 Core CMT Panel (seq & del/dup of 4 genes)
  - ❒ 885 Axonal CMT Panel (seq & del/dup of 21 genes)
  - ❒ 886 Demyelinating CMT Panel (seq & del/dup of 19 genes)
  - ❒ J778 CMT Panel (seq & del/dup of 36 genes)
  - ❒ 887 Rest of the Hereditary Neuropathy Panel (if subpanel negative)
- ❒ 889 Neuromuscular Disorders Panel (seq & del/dup of 80 genes)
  - ❒ 890 Limb-Girdle Muscular Dystrophy Panel (seq & del/dup of 24 genes)
  - ❒ 891 Syndromic Congenital Muscular Dystrophy Panel (seq & del/dup of 18 genes)
  - ❒ 892 Congenital Myopathy & Muscular Dystrophy Panel (seq & del/dup of 22 genes)
  - ❒ 893 Myofibrillar Myopathy Panel (seq & del/dup of 8 genes)
  - ❒ 894 Rest of Neuromuscular Disorders Panel (if subpanel negative)
- ❒ 787 Duchenne/Becker MD (DMD del/dup)
- ❒ 786 Duchenne/Becker MD (DMD seq if del/dup negative)
- ❒ 945 Congenital Myasthenia Syndromes Panel (seq & del/dup of 14 genes)
- ❒ 551 Nemaline myopathy (ACTA1 seq and AJ founder mutation in NEB gene)
- ❒ 342 Spinal muscular atrophy w/respiratory distress, type 1 (IGHMBP2 seq)
- ❒ 941 Comprehensive Hereditary Spastic Paraplegia Panel (seq & del/dup of 36 genes)
  - ❒ 942 Uncomplicated Hereditary Spastic Paraplegia Panel (seq & del/dup of 11 genes)
  - ❒ 943 Rest of Comprehensive Hereditary Spastic Paraplegia Panel (if subpanel negative)
- ❒ 944 Hereditary Spastic Paraplegia Related Inborn Error of Metabolism Panel (seq & del/dup of 14 genes)
- ❒ 650 Erythromelalgia, small fiber neuropathy (SCN9A seq)
- ❒ 363 Familial Amyloid Polyneuropathy (TTR seq)
- ❒ 238 Hereditary inclusion body myopathy (M743T [aka M712T] mut. in GNE)
- ❒ 818 Myotonic Dystrophy 1 (DM1) (DMPK repeat analysis)
  - ❒ 900* Reflex to DM1 Southern blot, if 818 is positive
- ❒ 819 Myotonic Dystrophy 2 (DM2) (CNBP repeat analysis)
- ❒ 743 Oculopharyngeal Muscular Dystrophy (PABPN1 repeat analysis)
- ❒ 820 Spinal & Bulbar Muscular Atrophy (AR repeat analysis)
- ❒ J805 Amyotyrophic lateral sclerosis/Frontotemporal lobar degeneration (C9orf72 repeat analysis)
- ❒ 706 ALS XomeDxSlice (seq-only of 24 genes; Slice ID S1498158236)

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

Mitochondrial Disorders

- ❒ 615 Combined Mito Genome Plus Mito Nuclear Gene Panel (seq & del/dup of mito genome and 319 nuclear genes)
- ❒ 554 Full sequence analysis and deletion testing of the mitochondrial genome
- ❒ 573 Comprehensive Mitochondrial Nuclear Gene Panel (seq & del/dup of 319 genes)
- ❒ 575 Mitochondrial Encephalopathy/Leigh Syndrome Nuclear Gene Panel (seq & del/dup of 146 genes)
- ❒ 576 Lactic Acidosis/Pyruvate Metabolism Nuclear Gene Panel (seq & del/dup of 153 genes)
- ❒ 577 Progressive External Ophthalmoplegia (PEO)/ Optic Atrophy Nuclear Gene Panel (seq & del/dup of 55 genes)
- ❒ 578 Methylglutaconic Aciduria Nuclear Gene Panel (seq & del/dup of 13 genes)
- ❒ 704 65 mtDNA Point Variants Plus Large Deletions Panel
- ❒ 444 Deletion/duplication analysis of mito genome
- ❒ 906 Deletion/duplication analysis of a single nuclear gene: **write in desired gene** _____
- ❒ 394 POLG gene sequencing
- ❒ 557 PUS1 Gene sequencing
- ❒ 582 SDHA Gene sequencing

Neurology Test Options

Account # Account Name

First Name Last Name Date of Birth (mm/dd/yy)

GeneDx Neurology Genetic Testing Menu (continued)

Neurometabolic & Other Neurogenetic Disorders

- ❒ 665 Hyperammonemia, Urea Cycle and Transporter Defects Panel (seq of 44 genes)
  - ❒ 684 Reflex del/dup panel for hyperammonemia, urea cycle & transporter defect panel
- ❒ 667 Methylmalonic Acidemia, Disorders of Cobalamin Metabolism and Related Disorders Panel (seq of 16 genes)
  - ❒ 685 Reflex del/dup panel for methylmalonic academia & related disorders panel
- ❒ 664 Fatty Acid Oxidation Disorders Panel (seq of 15 genes)
  - ❒ 683 Reflex del/dup panel for fatty acid oxidation disorders panel
- ❒ J544 Metabolic Myopathy Panel (seq of 24 genes)
  - ❒ J698 Reflex del/dup panel for metabolic myopathy panel
- ❒ 547 Aicardi-Goutieres syndrome (TREX1, RNASEH2A, RNASEH2B, RNASEH2C seq)
- ❒ 218 Alexander disease (GFAP seq)
- ❒ 219 Allgrove (Triple-A) syndrome (AAAS seq)
- ❒ 334 Carnitine palmitoyltransferase type II (CPT2) Deficiency (CPT2 seq)
- ❒ 526 Cerebral cavernous malformations (KRIT1, CCM2, PDCD10 seq & del/dup)
- ❒ 274 Cobalamin C deficiency (MMACHC seq)
- ❒ 550 Coffin-Lowry syndrome (RPS6KA3 seq & del/dup)
- ❒ 227 Cohen syndrome (VPS13B seq)
- ❒ 527 Dopa-responsive dystonia (GCH1 seq & del/dup)
- ❒ 359 Dopa-responsive dystonia/Infantile Parkinsonism/TH deficiency (TH seq)
- ❒ 2321 Fabry disease (GLA seq)
- ❒ 399 Glutaric Aciduria Type I (GCDH seq)
- ❒ 2371 Holoprosencephaly (SHH, ZIC2, SIX3,TGIF seq & del/dup)
- ❒ 553 Incontinentia pigmenti (IKBKG [NEMO] common deletion assay and seq; for females only)
- ❒ 583 Kabuki syndrome (MLL2 seq)
- ❒ 673 KBG syndrome (ANKRD11 seq)
- ❒ 507 Krabbe disease (GALC seq & del/dup)
- ❒ 2432 Mucolipidosis IV (MCOLN1 seq)
- ❒ 2631 Niemann-Pick A/B disease (SMPD1 seq)
- ❒ 581 Niemann-Pick C disease (NPC1, NPC2 seq)
- ❒ 313 Ornithine transcarbamylase deficiency (OTC seq)
- ❒ 313E Ornithine transcarbamylase deficiency (OTC seq & del/dup in females)
- ❒ 595 Prader-Willi syndrome methylation-MLPA (UPD, deletion)
- ❒ 2923 Rubinstein-Taybi syndrome (CREBBP seq & del/dup)
- ❒ 474 Septo-optic dysplasia (HESX1 seq)
- ❒ 415 Simpson-Golabi-Behmel syndrome (GPC3 seq in males)
- ❒ 415E Simpson-Golabi-Behmel syndrome (GPC3 seq & del/dup in females)
- ❒ 2502 Smith-Lemli-Opitz syndrome (DHCR7 seq)
- ❒ 2511 Smith-Magenis syndrome (RAI1 seq & intragenic del/dup)
- ❒ 519 Tay-Sachs disease (HEXA seq)
- ❒ 532 Trifunctional protein deficiency (HADHA, HADHB seq)
- ❒ 270 VLCAD deficiency (ACADVL seq)
- ❒ 552 X-linked hydrocephalus/X-linked spastic paraplegia/ MASA/CRASH syndrome (L1CAM seq & del/dup)

Neurofibromatosis (NF)

- ❒ 962 NF type 1 Panel: NF1 and SPRED1 (seq & del/dup)
- ❒ 534 Noonan syndrome and RASopathies panel (seq of 15 genes) (reflex if 962 is negative)
- ❒ 963 NF type 2 Panel: NF2 and SMARCB1 (seq & del/dup)
- ❒ 961 Comprehensive NF Panel (seq & del/dup of 4 genes)
- ❒ J660 NF type 1 (NF1 seq & del/dup only)
- ❒ 816 Legius syndrome (SPRED1 seq only)

Additional Test

Test Code _____ Test Name _____

Family History of Neurological Disorders

- ❒ No Known Family History
- ❒ Pedigree Attached
- ❒ Adopted

Relationship	Maternal	Paternal	Neurological Disorder	Age at Dx
	❒	❒		
	❒	❒		
	❒	❒		
	❒	❒		
	❒	❒		
	❒	❒		
	❒	❒		
	❒	❒		

Family Member Testing

Testing for known familial variant in a nuclear gene

- ❒ 9011 Testing for ONE known familial variant in a nuclear gene
- ❒ 9012 Testing for TWO known familial variants in a nuclear gene
- ❒ 905 Testing for ONE known familial exon-level del/dup or chromosomal microarray del/dup

Testing for known mtDNA variant(s)

- ❒ 453 Testing for ONE to THREE mtDNA variant(s) (with heteroplasmy level)
- ❒ 9017 Testing for ONE mtDNA variant (with estimated heteroplasmy level)
- ❒ 9020 Testing for TWO mtDNA variants (with estimated heteroplasmy level)

Gene(s): _____ Variant(s): _____

Proband Name: _____

Proband GeneDx Acc#:_____ Relationship to proband: _____

- ❒ Positive control included - **Positive control is required if previous test was performed at another lab.**
- ❒ Family Member Test Report included - A clear copy of the test report on the variant positive family member is recommended if previous test was performed at another lab.

Neurology Clinical Information

GeneDx an OPKO Health Company

Account # Account Name

First Name Last Name Date of Birth (mm/dd/yy)

DETAILED MEDICAL RECORDS MUST BE ATTACHED

Clinical Diagnosis:_____ **ICD-10 Codes:**_____ **Age at Initial Presentation:**____ ❒ **Parent/Carrier testing-unaffected/no symptoms**

Perinatal History
- ❒ Cystic hygroma/increased NT
- ❒ IUGR
- ❒ Oligohydramnios/polyhydramnios (circle if applies)
- ❒ Prematurity

Growth
- ❒ Failure to thrive
- ❒ Macrocephaly Head circumference: _____
- ❒ Microcephaly Head circumference:_____
- ❒ Overgrowth
- ❒ Short stature

Physical/Cognitive Development
- ❒ Developmental regression
- ❒ Fine motor delay
- ❒ Gross motor delay
- ❒ Intellectual disability/MR IQ:_____
- ❒ Learning disability
- ❒ Speech delay

Behavioral
- ❒ Autism spectrum disorder
- ❒ Autistic features
- ❒ Behavioral/Psychiatric abnormalities (circle if applies)
- ❒ Obsessive-compulsive disorder
- ❒ Stereotypic behaviors

Craniofacial/Ophthalmalogic/Auditory
- ❒ Blindness
- ❒ Cataracts
- ❒ Cleft lip/palate
- ❒ Coloboma of eye
- ❒ CPEO (Ophthalmoplegia)
- ❒ External ear malformation
- ❒ Eye movement disorder
- ❒ Facial dysmorphism - please describe:

  _____

  _____
- ❒ Optic atrophy
- ❒ Ototoxicity (aminoglycoside-induced)
- ❒ Ptosis
- ❒ Retinitis pigmentosa
- ❒ Sensorineural hearing loss
- ❒ Other visual abnormality:_____

Cardiac/Congenital Heart Malformations
- ❒ Arrhythmia/conduction defect
- ❒ ASD/VSD (circle if applies)
- ❒ Cardiomegaly
- ❒ Cardiomyopathy
- ❒ Coarctation of aorta
- ❒ Hypoplastic left heart
- ❒ Tetralogy of Fallot

Gastrointestinal
- ❒ Chronic diarrhea
- ❒ Constipation
- ❒ Delayed gastric emptying
- ❒ Gastrointestinal reflux
- ❒ Gastroschisis/omphalocele
- ❒ Hepatic failure
- ❒ Nausea
- ❒ Pyloric stenosis
- ❒ Recurrent vomiting
- ❒ Tracheoesophageal fistula

Seizures/Epilepsy
- ❒ Epileptic encephalopathy
- ❒ Febrile seizures
  - ❒ Dravet syndrome
- ❒ Focal seizures
- ❒ Generalized seizures
  - ❒ Absence ❒ Clonic
  - ❒ Myoclonic ❒ Tonic-clonic
- ❒ Infantile/epileptic spasms
  - ❒ Ohtahara syndrome ❒ West syndrome
- ❒ Status epilepticus

Brain Malformations/Abnormal Imaging
- ❒ Abnormalities of basal ganglia
- ❒ Agenesis of the corpus callosum
- ❒ Brain atrophy
- ❒ Cortical dysplasia
- ❒ Frontotemporal lobar degeneration
- ❒ Hemimegalencephaly
- ❒ Holoprosencephaly
- ❒ Hydrocephalus
- ❒ Lissencephaly
- ❒ Molar tooth sign
- ❒ Periventricular leukomalacia
- ❒ Periventricular nodular heterotopia
- ❒ Polymicrogyria
- ❒ Pontocerebellar hypoplasia
- ❒ Subcortical band hetertopia
- ❒ Tumor (type: _____)

Muscular
- ❒ Abnormal electromyography (EMG)
- ❒ Dysphagia
- ❒ Easy fatigue
- ❒ Exercise intolerance
- ❒ Hypertonia
- ❒ Hypotonia
- ❒ Joint hypermobility
- ❒ Muscle fasciculations
- ❒ Muscle stiffness
- ❒ Muscle wasting
- ❒ Muscle weakness: proximal/distal/ upper limb/lower limb (circle all that apply)
- ❒ Myotonia
- ❒ Respiratory insufficiency

Neurological
- ❒ Nerve conduction studies: _____
- ❒ Ataxia
- ❒ Bulbar signs
- ❒ Chorea
- ❒ Congenital Neuropathy
- ❒ Distal motor neuropathy
- ❒ Dystonia
- ❒ Episodic apnea (sudden)
- ❒ Foot drop
- ❒ Hypomyelination
- ❒ Lower extremity weakness
- ❒ Motor neuron dysfunction: ❒ Upper ❒ Lower
- ❒ Pes cavus
- ❒ Pressure palsy
- ❒ Recurrent headache/migraine
- ❒ Reduced/absent deep tendon reflexes
- ❒ Sensory Neuropathy
  - ❒ Hyperesthesia ❒ Paresthesia
- ❒ Sleep apnea
- ❒ Spasticity
- ❒ Stroke/stroke-like episodes
- ❒ Tremor/Parkinsonism (circle all that apply)
- ❒ Vocal cord paresis

Autonomic
- ❒ Abnormal sweating
- ❒ Abnormal temperature regulation

Endocrine
- ❒ Diabetes mellitus: ❒ Type I ❒ Type II
- ❒ Gynecomastia
- ❒ Hypoparathyroidism
- ❒ Hypothyroidism
- ❒ Pheochromocytoma/paraganglioma

Skeletal/Limb Abnormalities
- ❒ Club foot
- ❒ Contractures
- ❒ Hammer toe
- ❒ Hip dysplasia
- ❒ Osteomyelitis/necrosis
- ❒ Polydactyly
- ❒ Scoliosis
- ❒ Syndactyly
- ❒ Vertebral anomaly

Genitourinary Abnormalities
- ❒ Ambiguous genitalia
- ❒ Hydronephrosis
- ❒ Hypospadias
- ❒ Kidney malformation
- ❒ Neurogenic bladder
- ❒ Renal tubulopathy
- ❒ Undescended testis

Metabolic
- ❒ CPK abnormalities (value: _____)
- ❒ Elevated alanine
- ❒ Elevated pyruvate
- ❒ Hyperammonemia
- ❒ Hypoglycemia
- ❒ Ketosis
- ❒ Lactic acidemia/high CSF lactate
- ❒ Low plasma carnitine
- ❒ Organic aciduria

Skin Abnormalities
- ❒ Axillary and/or inguinal freckling
- ❒ Hypopigmentation/hyperpigmentation type: _____
- ❒ Other skin abnormality:_____

Biopsy Abnormalities
- ❒ Muscle biopsy
  - ❒ COX deficiency
  - ❒ Histology:_____
  - ❒ Large mitochondria (mt)/mt proliferation
  - ❒ Positive newborn screen: _____
  - ❒ Ragged red fibers
  - ❒ Respiratory enzymes: _____
  - ❒ Ultrastructure (EM): _____
- ❒ Nerve biopsy
  - ❒ Histology:_____
  - ❒ Ultrastructure (EM): _____

Other clinical history or testing (summarize or attach reports):
- ❒ Array CGH: _____
- ❒ Chromosomes/FISH: _____
- ❒ Other relevant results (clinical or research):

  _____

  _____

Draw/attach pedigree and/or include additional clinical information _____

_____

_____

Informed Consent and Authorization Form

Account # Account Name

First Name Last Name Date of Birth (mm/dd/yy)

I understand that my health care provider has ordered the following genetic testing for {me/my child}:_____.

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 harmful changes in DNA or from changes in the structure or number of chromosomes. Genetic testing is a laboratory test that tries to identify these harmful 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 diagnostic 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.

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 specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, and the limitations of genetic testing.

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 for 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 both 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 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.

Informed Consent and Authorization Form

Account # Account Name

First Name Last Name Date of Birth (mm/dd/yy)

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.

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 health 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 below. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language.

Database Participation

De-identified health 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 databases, 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 health care 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 {me/my child} or {my/my child's} heirs.

Patient Consent (sign here or on page 1 of the test requisition form)

I have read the Informed Consent document and I give permission to GeneDx to perform genetic testing as described. I also give permission for my specimen and clinical information to be used in de-identified studies at GeneDx to improve genetic testing and for publication, if appropriate. My name or other personal identifying information will not be used in or linked to the results of any studies and publications. I also give GeneDx permission to inform me or my health care provider in the future about research opportunities, including treatments for the condition in my family.

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

❒ Check this box if you are New York state resident, and give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing.

Patient/Guardian Signature Date (mm/dd/yyyy)

If I wish to change my decisions or have any questions, I understand that I may contact the laboratory via email at genedx@genedx.com or by phone at +1-301-519-2100, or if I am located in the United States, toll free at +1-888-729-1206.

A. Notifier:

B. Patient Name: **C. Identification Number:**

Advance Beneficiary Notice of Noncoverage (ABN)

NOTE: If Medicare doesn't pay for **D.**_____below, you may have to pay. Medicare does not pay for everything, even some care that you or your health care provider have good reason to think you need. We expect Medicare may not pay for the **D.**_____below.

D.	E. Reason Medicare May Not Pay:	F. Estimated Cost

WHAT YOU NEED TO DO NOW:

- Read this notice, so you can make an informed decision about your care.
- Ask us any questions that you may have after you finish reading.
- Choose an option below about whether to receive the **D.**_____listed above.
  Note: If you choose Option 1 or 2, we may help you to use any other insurance that you might have, but Medicare cannot require us to do this.

G. OPTIONS: Check only one box. We cannot choose a box for you.

☐ **OPTION 1.** I want the **D.**_____listed above. You may ask to be paid now, but I also want Medicare billed for an official decision on payment, which is sent to me on a Medicare Summary Notice (MSN). I understand that if Medicare doesn't pay, I am responsible for payment, but **I can appeal to Medicare** by following the directions on the MSN. If Medicare does pay, you will refund any payments I made to you, less co-pays or deductibles.

☐ **OPTION 2.** I want the **D.**_____listed above, but do not bill Medicare. You may ask to be paid now as I am responsible for payment. **I cannot appeal if Medicare is not billed.**

☐ **OPTION 3.** I don't want the **D.**_____listed above. I understand with this choice I am **not** responsible for payment, and **I cannot appeal to see if Medicare would pay.**

H. Additional Information:

This notice gives our opinion, not an official Medicare decision. If you have other questions on this notice or Medicare billing, call **1-800-MEDICARE** (1-800-633-4227/**TTY:** 1-877-486-2048). Signing below means that you have received and understand this notice. You also receive a copy.

I. Signature:	J. Date:

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