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 Provider 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.

Signature of Physician or Other Authorized NPI Provider (required) Date

Patient Consent (sign here)

I have read the attached 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. **More information is available on our website: www.genedx.com**

❒ 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

PATIENT STATUS – ONE MUST BE CHECKED: ❒ Hospital Inpatient ❒ Hospital Outpatient ❒ Not a Hospital Patient Hospital Patient Date of Discharge:_____

Payment Options

❒ Insurance Bill

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

Insurance Carrier Policy Name ❒ Hold sample for Estimated 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

Relationship to Insured ❒ Child ❒ Spouse ❒ Self ❒ Other _____

Secondary Insurance Carrier Name Insurance ID# Group # Name of Insured Date of Birth

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)

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 estimated 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 _____

If I have insurance coverage for this testing, I am electing to be treated as a self-pay patient for this testing. As such, I agree that neither GeneDx nor I will submit a claim to my insurance for this testing.

Please bill my credit card for the full amount stated above (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)

Family History of Disorder/Symptoms

❒ No Known Family History
❒ Pedigree Attached
❒ Adopted

Relationship	Maternal	Paternal	Disorder/Symptoms	Age at Dx
	❒	❒		
	❒	❒		
	❒	❒		
	❒	❒		

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 information

Family Member/Carrier 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) (heteroplasmy detection range: 1.5%-100%)
❒ 9017 Testing for ONE mtDNA variant (heteroplasmy detection range: 25%-100%)
❒ 9020 Testing for TWO mtDNA variants (heteroplasmy detection range: 25%-100%)

Mosaic Carrier Test

❒ J829 Testing for ONE known familial variant in a nuclear gene (please see website for complete list of applicable genes; insurance billing not accepted)

*Please select the repeat expansion analysis code for repeat expansion carrier testing.

Please fill out this information if selecting a test from the family member/carrier testing section:

Gene(s): _____ Variant(s): _____
Proband Name: _____
Proband GeneDx Acc#: _____
Relationship to proband: _____

❒ Parent/Carrier testing: Asymptomatic / Symptomatic (Circle one)
❒ Positive control included - **Positive control is required if previous test was performed at another lab.**
❒ Positive control not available. Please initial to acknowledge acceptance of caveat language on a negative report_____
❒ 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.

Family Member Testing (no separate report)

Mother: ❒ At GeneDx ❒ Not Available ❒ To be sent later**
First Name: _____ Last Name: _____ DOB: _____
❒ Asymptomatic ❒ Symptomatic

Father: ❒ At GeneDx ❒ Not Available ❒ To be sent later**
First Name: _____ Last Name: _____ DOB: _____
❒ Asymptomatic ❒ Symptomatic

Other: ❒ At GeneDx ❒ Not Available ❒ To be sent later**
Relationship to Proband: _____
First Name: _____ Last Name: _____ DOB: _____
❒ Asymptomatic ❒ Symptomatic

❒ J767 Ataxia Xpanded, Family Member Testing
❒ 954 Autism/ID Xpanded, Family Member Testing
❒ T997 Cerebral Palsy Xpanded, Family Member Testing
❒ 923 EpiXpanded, Family Member Testing
❒ 910 GenomeDx, Parental Testing
❒ J854 Leukodystrophy Xpanded, Family Member Testing
❒ J513 Microcephaly Xpanded, Family Member Testing
❒ J820 MitoXpanded, Family Member Testing

>> See next page for proband test selection

**** ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS**

Single Gene Analysis/Write-in Test Selection

❒ 906 Deletion/Duplication Analysis of ONE nuclear gene

Write in desired gene: _____

Test Code: _____ Test Name: _____

Test Code: _____ Test Name: _____

Please see our website (www.genedx.com) or other requisitions for additional tests not included on this requisition.

Neurology Test Options

Account # Account Name

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

GeneDx Neurology Genetic Testing Menu

Neurodevelopmental Disorders and Epilepsy

- ❒ 522 Fragile X syndrome (FMR1 repeat analysis)
- ❒ 910 Chromosomal Microarray (GenomeDx)
- ❒ T395 Autism/ID Panel (seq & del/dup of 104 genes)

Order of Reflex Testing:

- ❒ Concurrent analysis of 522 & 910, if negative activate T395
- ❒ Start with 522, if negative activate 910, if negative 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)
- ❒ 595 Prader-Willi syndrome methylation-MLPA (UPD, deletion)
- ❒ 566 Angelman 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 negative)
- ❒ 921 EpiXpanded Panel (1300+ genes, trios preferred)
- ❒ 953 Epilepsy Del/Dup Panel (128 genes) (not a trio based test)
- ❒ 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 negative)
- ❒ 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)
- ❒ 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 negative)
- ❒ 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)
- ❒ J762 Ataxia Xpanded Panel (950+ genes, trios preferred)
- ❒ 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 negative)
- ❒ 527 Dopa-responsive dystonia (GCH1 seq & del/dup)
- ❒ 359 Dopa-responsive dystonia/Infantile Parkinsonism/TH deficiency (TH seq)
- ❒ 218 Alexander disease (GFAP seq)
- ❒ 581 Niemann-Pick C disease (NPC1, NPC2 seq)

Neuromuscular Disorders

- ❒ 737 Hereditary Neuropathy Panel (seq & del/dup of 64 genes)
  - ❒ 884 Core CMT Panel (seq & del/dup of 4 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 negative)
- ❒ 742 CMT1A/HNPP (PMP22 del/dup)
- ❒ 888 HNPP/CMT1E (PMP22 seq)
- ❒ 363 Familial Amyloid Polyneuropathy (TTR seq)
- ❒ 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 negative activate T404

- ❒ 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)
  - ❒ 894 Rest of Neuromuscular Disorders Panel (if subpanel negative)
- ❒ 787 Duchenne/Becker MD (DMD del/dup)
- ❒ 786 Duchenne/Becker MD (DMD seq)
- ❒ T406 Spinal Muscular Atrophy Panel (seq & del/dup of 18 genes plus SMN1/2 Dosage Analysis)
- ❒ T789 SMN1/2 Dosage Analysis
- ❒ 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)
- ❒ 945 Congenital Myasthenia Syndromes Panel (seq & del/dup of 18 genes)

* Samples from New York state cannot be accepted for the Southern Blot test. A 2-5 mL blood sample is required for Southern Blot analysis.

Neurology Test Options

Account # Account Name

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

GeneDx Neurology Genetic Testing Menu

Mitochondrial Disorders

- ❒ J809 MitoXpanded Panel (1800+ genes, trios preferred)
  - ❒ 554 Concurrent full sequence analysis & deletion testing of the mito genome (not a trio based test)
- ❒ 615 Combined Mito Genome Plus Mito Focused Nuclear Gene Panel (seq & del/dup of mito genome and 202 nuclear genes)
- ❒ 554 Full sequence analysis and deletion testing of the mitochondrial genome
- ❒ 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 152 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)
- ❒ 704 65 mtDNA Point Variants Plus Large Deletions Panel
- ❒ 444 Deletion/duplication analysis of mito genome
- ❒ 394 POLG gene sequencing

Neurometabolic Disorders

- ❒ J979 Combined Lysosomal and Peroxisomal Disorders Panel (seq & del/dup of 83 genes)
  - ❒ T013 Lysosomal Disorders Panel (seq & del/dup of 25 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)
- ❒ 507 Krabbe Disease (GALC seq & del/dup)
- ❒ 287 Pompe disease/glycogen storage disease type II (GAA seq)
- ❒ J975 X-linked adrenoleukodystrophy (ABCD1 seq & del/dup)

Neurofibromatosis

- ❒ 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 negetive
- ❒ 963 NF2 panel: NF2 and SMARCB1 sequencing and deletion/duplication testing
- ❒ 961 Comprehensive NF panel: NF1, SPRED1, NF2 and SMARCB1 sequencing and deletion/duplication testing

Neurology Clinical Information

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:**____ ❒ **Unaffected/asymptomatic Parent or Carrier testing**

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

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 IQ: _____
- ❒ Learning disability
- ❒ Speech delay

Behavioral
- ❒ Autism spectrum disorder
- ❒ Autistic features
- ❒ Behavioral/Psychiatric abnormalities (circle all that apply)
- ❒ Obsessive-compulsive disorder
- ❒ Stereotypic behaviors
- ❒ Dementia (early/late) onset (circle if applies)

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 all that apply)
- ❒ 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
- ❒ Cerebellar atrophy
- ❒ Cortical dysplasia
- ❒ Frontotemporal lobar degeneration
- ❒ Lissencephaly
- ❒ Molar tooth sign
- ❒ Periventricular nodular heterotopia
- ❒ Polymicrogyria
- ❒ Pontocerebellar hypoplasia
- ❒ Subcortical band hetertopia
- ❒ Imaging abnormalities: _____

_____

Muscular
- ❒ Abnormal electromyography (EMG)
- ❒ Dysphagia
- ❒ Dysarthria
- ❒ 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)
- ❒ Myopathic facies
- ❒ Myotonia

Movement
- ❒ Ataxia
- ❒ Chorea
- ❒ Dystonia
- ❒ Dyskinesia
- ❒ Spasticity
- ❒ Tremor/Parkinsonism (circle all that apply)

Neurological
- ❒ Nerve conduction studies: _____
- ❒ Congenital neuropathy
- ❒ Distal motor neuropathy
- ❒ Episodic apnea (sudden)
- ❒ Foot drop
- ❒ Hypomyelination
- ❒ Motor neuron dysfunction: ❒ Upper ❒ Lower
- ❒ Pes cavus
- ❒ Pressure palsy
- ❒ Recurrent headache/migraine
- ❒ Reduced/absent deep tendon reflexes
- ❒ Sensory neuropathy
  - ❒ Hyperesthesia ❒ Paresthesia
- ❒ Sleep apnea
- ❒ Stroke/stroke-like episodes
- ❒ Vocal cord paresis

Autonomic
- ❒ Abnormal sweating
- ❒ Abnormal temperature regulation

Endocrine
- ❒ Adrenal insufficiency
- ❒ Diabetes mellitus: ❒ Type I ❒ Type II
- ❒ Gynecomastia
- ❒ Hypoparathyroidism
- ❒ Hypothyroidism
- ❒ Pheochromocytoma/paraganglioma
- ❒ Other endocrine dysfunction _____

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
- ❒ Positive newborn screen: _____

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
  - ❒ Ragged red fibers
  - ❒ Respiratory enzymes: _____
  - ❒ Ultrastructure (EM): _____
- ❒ Nerve biopsy
  - ❒ Histology:_____
  - ❒ Ultrastructure (EM): _____

Informed Consent

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.

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.

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.

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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