NEUROLOGY TEST REQUISITION FORM

GeneDx an OPKO Health Company

PATIENT INFORMATION

First name | Last name

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: _____

ACCOUNT INFORMATION

Account number | **Account name**

Phone | Fax

Address | City

State | Zip code | Country

Ordering provider

Name | Role/Title

Phone | NPI

Email address (for report access)

Reporting Preference: ◯ Portal ◯ Fax ◯ Email ◯ Care Evolve *If unmarked, we will use the account's default preferences or fax to new clients.*

Additional Reporting Providers ◯ Same as ordering provider

Name | Role/Title

Phone | NPI

Email address (for report access)

Additional clinical or laboratory contact (optional)

Name | Email address (for report access)

SEND ADDITIONAL REPORT COPIES TO

Healthcare provider/Acct # | Fax #/Email

SAMPLE INFORMATION

Date sample obtained (mm/dd/yy) | Medical record #

◯ Blood ◯ Buccal swab
◯ Muscle (~50mg flash frozen; shipped on dry ice) ◯ Other: _____
◯ DNA: tissue 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: _____

ICD-10 codes (required): | **Clinical diagnosis:** | **Age at initial presentation:**

STATEMENT OF MEDICAL NECESSITY

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

Signature of provider (required) **Date**

PATIENT CONSENT

By signing this form I acknowledge as the patient that I have read the attached informed consent document and that I authorize GeneDx to perform genetic testing as described. For tests that evaluate data from multiple family members concurrently, such as me and my spouse or partner, results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their health care providers I have been informed that GeneDx may contact me or my healthcare provider about research opportunities in the future. For the insurance bill, I understand and authorize GeneDx to share information with the designated insurance carrier for reimbursement. I understand that GeneDx will attempt to contact me if my estimated out-of-pocket responsibility will be greater than $100 per test. If GeneDx is unsuccessful in its attempts to contact me, it will be my responsibility to contact GeneDx to determine and pay the out-of-pocket cost. More information, including the GeneDx Notice of Privacy Policies, is available on GeneDx's website: www.genedx.com

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

PATIENT STATUS – ONE MUST BE CHECKED: ◯ **Hospital outpatient** ◯ **Hospital inpatient** **Date of discharge:** _____ ◯ **Not a hospital patient**

PAYMENT OPTIONS

◯ **Insurance Bill** | ◯ Copy of front and back of insurance card(s) included | ◯ **Hold sample for estimated benefit investigation**

Insurance carrier | Referral/Prior authorization number (please attach) | GeneDx benefit investigation #

Insurance ID # | Relationship to insured: ◯ Self ◯ Spouse ◯ Child ◯ Other _____ | Policy holder name | Policy holder's date of birth

◯ **Patient Bill** Amount _____

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.

◯ **GeneDx Affiliate Code:**

◯ **Institutional Bill**

GeneDx account # | Hospital/Lab name

Place sticker/stamp here

CLINICAL INFORMATION

Account #	Account Name

First Name	Last Name	Date of Birth

CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

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
- ○ 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/Mito
(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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First Name	Last Name	Date of Birth

REASON FOR EXPEDITED TESTING (REQUIRED)

○ Pregnancy (gestational age____weeks) ○ Transplantation ○ Other:_____

TARGETED VARIANT TESTING AND SPECIAL SERVICES

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

○ Known Familial Variant(s) in a Nuclear Gene	○ Mosaic Carrier Testing
○ Known Familial Copy Number variant(s)	○ Known mtDNA Variant(s) Testing (heteroplasmy detection range 25%-100%)
○ Confirmation of Variant Identified in Research Lab	○ 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 #: _____

Non-GeneDx Test: ○ Family member test report included (recommended if previous test was performed at another lab)
○ Positive control included/will be sent - **Positive control is 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)

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

Mother: First Name: _____ Last Name: _____ DOB: _____	○ Asymptomatic ○ Symptomatic	○ Not Available ○ To be sent later**
Father: First Name: _____ Last Name: _____ DOB: _____	○ Asymptomatic ○ Symptomatic	○ Not Available ○ To be sent later**
Other: First Name: _____ Last Name: _____ DOB: _____ Relationship to Proband: _____	○ Asymptomatic ○ Symptomatic	○ Not Available ○ To be sent later**

>> See next page for proband test selection

** ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS

WRITE-IN TEST SELECTION

○ Test Code:_____ Test Name: _____

HISTORY

FAMILY HISTORY

○ No Known Family History ○ Pedigree Attached ○ Adopted

RELATIONSHIP	MATERNAL	PATERNAL	RELEVANT HISTORY	AGE AT DX
	○	○		
	○	○		
	○	○		

TESTING HISTORY

○ Test report included (recommended)

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

207 Perry Parkway, Gaithersburg, MD 20877 • T: (888) 729-1206 (Toll-Free), (301) 519-2100 • F: (201) 421-2010 • www.genedx.com

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TEST MENU

NEURODEVELOPMENTAL DISORDERS AND EPILEPSY

○ 522	Fragile X syndrome (*FMR1* repeat analysis)	○ 546	Angelman syndrome *(UBE3A* seq & del/dup)
○ 910	Chromosomal Microarray (GenomeDx)	○ 523	Comprehensive Epilepsy Panel (seq & del/dup of 127 genes)
○ T395	Autism/ID Panel (seq & del/dup of 104 genes)	○ 814	STAT Epilepsy Panel (seq & del/dup of 26 genes)
Order of Reflex Testing:		○ 541	Infantile Epilepsy Panel (seq & del/dup of 111 genes)
○ Concurrent analysis of 522 & 910, if negative activate T395		○ 542	Childhood-Onset Epilepsy Panel (seq & del/dup of 84 genes)
○ Start with 522, if negative activate 910, if negative activate T395		○ 544	Progressive Myoclonic Epilepsy Panel (seq & del/dup of 18 genes)
○ 952	Autism/ID Xpanded Panel (2300+ genes, trios preferred)	○ 545	Rest of the Comprehensive Epilepsy Panel (if subpanel negative)
○ 195	PTEN-related disorders *(PTEN* seq & del/dup)	○ 921	EpiXpanded Panel (1300+ genes, trios preferred)
○ 729	Rett/Angelman Related Disorders Panel (seq & del/dup of 20 genes)	○ 953	Epilepsy Del/Dup Panel (128 genes) (not a trio based test)
○ 549	Rett/Atypical Rett syndromes *(MECP2* seq & del/dup)	○ T400	Hemiplegic migraine panel (seq & del/dup of 4 genes)
○ 595	Prader-Willi syndrome methylation-MLPA (UPD, deletion)	○ 730	Tuberous Sclerosis Panel *(TSC1* & *TSC2* seq & del/dup)
○ 566	Angelman syndrome methylation-MLPA (UPD, deletion)		

CNS MALFORMATIONS AND DISORDERS

○ 691	Comprehensive Brain Malformations Panel (seq & del/dup of 103 genes)	○ J511	Microcephaly Xpanded Panel (800+ genes, trios preferred)
○ 698	Cortical Brain Malformations Panel (seq & del/dup of 61 genes)	○ 699	Syndromic Macrocephaly/Overgrowth Syndromes Panel (seq & del/dup of 29 genes)
○ 700	Pontocerebellar Hypoplasia Panel (seq & del/dup of 19 genes)	○ J853	Leukodystrophy Xpanded Panel (300+ genes, trios preferred)
○ 701	Joubert Syndrome and Related Disorders Panel (seq & del/dup of 29 genes)	○ 552	X-linked hydrocephalus/X-linked spastic paraplegia/MASA/CRASH syndrome *(LICAM* seq & del/dup)
○ 946	Lissencephaly Panel (seq & del/dup of 26 genes)	○ TB51	Comprehensive Holoprosencephaly Panel (seq & del/dup of 17 genes)
○ 722	Rest of the Brain Malformations Panel (if subpanel negative)	○ 2371	Holoprosencephaly *(SHH, ZIC2, SIX3,TGIF* seq & del/dup)
○ 689	Microcephaly Panel (seq & del/dup of 65 genes)	○ 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)	○ T402	Dystonia and Parkinsonism Panel (seq & del/dup of 73 genes)
○ 942	Uncomplicated Hereditary Spastic Paraplegia Panel (seq & del/dup of 14 genes)	○ T403	Dystonia Panel (seq & del/dup of 53 genes)
○ 943	Rest of Comprehensive Hereditary Spastic Paraplegia Panel (if subpanel negative)	○ T401	Parkinson Disease Panel (seq & del/dup of 29 genes)
○ 944	Hereditary Spastic Paraplegia Related Inborn Error of Metabolism Panel (seq & del/dup of 15 genes)	○ T919	Rest of Dystonia and Parkinsonism Panel (if subpanel negative)
○ T851	Cerebral Palsy Xpanded Panel (1100+ genes, trios preferred)	○ 527	Dopa-responsive dystonia *(GCH1* seq & del/dup)
○ J762	Ataxia Xpanded Panel (1300+ genes, trios preferred)	○ 359	Dopa-responsive dystonia/Infantile Parkinsonism/TH deficiency (TH seq)
		○ 218	Alexander disease *(GFAP* seq)
		○ 581	Niemann-Pick C disease *(NPC1, NPC2* seq)

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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.

MITOCHONRDRIAL 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 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)
○ 704	65 mtDNA Point Variants Plus Large Deletions Panel
○ TB60	Deletion analysis of mito genome
○ 394	POLG gene sequencing

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)
○ 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 negative
○ 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

INFORMED CONSENT

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 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.