RARE DISORDERS TEST REQUISITION FORM

GeneDx

All sections on this page are required unless otherwise specified. Incomplete information could result in a delay of testing.

PATIENT INFORMATION

First Name	**Last Name**	
Sex Assigned at Birth: ◯ **Male** ◯ **Female** Patient Karyotype (if known): _____ Gender Identification (optional): _____	**Date of Birth (mm/dd/yy)**	
Email		
Address		
City	State	Zip Code
Phone (mobile preferred)	Is this patient deceased? ◯ Yes ◯ No Deceased Date: _____	

SAMPLE INFORMATION

Date Sample Collected (mm/dd/yy)	**Medical Record #**

◯ **Blood** ◯ **Buccal Swab** ◯ **Other (specify source):** _____

☐ **Treatment-related RUSH** (optional)
Reason: ◯ Transplantation ◯ Pregnancy ◯ Surgery ◯ Other: _____

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 allogeneic bone marrow transplant ◯ **Yes** ◯ **No**
Fibroblasts are required for patients who had an allogeneic bone marrow transplant. See www.genedx.com/specimen-requirements for details.

Patient has a personal history of a hematologic malignancy or disease
◯ **Yes (specify diagnosis)** _____ ◯ **No**
If yes, please call the lab to discuss with a genetic counselor the most appropriate sample type.

ORDERING PROVIDER ATTESTATION

By signing this form, the ordering provider attests that (i) he/she authorizes and directs GeneDx to perform the testing indicated; (ii) he/she is the ordering provider and is authorized by law to order the test(s) requested; (iii) any test(s) requested on this Test Requisition Form ("TRF") are reasonable and medically necessary for the diagnosis or treatment of a disease, illness, impairment, symptom, syndrome or disorder; (iv) the test results will determine the patient's medical management and treatment decisions of this patient's condition on this date of service; (v) the patient or the individual/family member authorized to make decisions for the patient (collectively, the "patient"), in addition to any relatives', when applicable, has been supplied with information regarding genetic testing, and has consented to undergo genetic testing; (vi) the full and appropriate diagnosis codes are indicated to the highest level of specificity; (vii) he/she will not seek reimbursement from any third party, including but not limited to federal healthcare programs if testing is covered by GeneDx and will inform the patient of the same; (viii) GeneDx may share contact information for the ordering provider and other healthcare providers listed on the this order with third parties regarding the requested genetic testing and potential clinical trial or study opportunities; and (ix) the patient or the individual/family member authorized to be contacted via the email address or mobile phone number provided for this and future testing.

☐ **New York Retention Opt-In.** By checking this box, I confirm that the patient is a New York State resident who gives permission for GeneDx to retain any remaining sample longer than 60 days after testing has been completed.

☐ **Patient Research Opt-Out.** By checking this box, I confirm that the patient wishes to opt out of being contacted for research studies.

☐ **Health Information Exchange Opt-in.** Check this box if your patient resides in CA, FL, MA, NV, NY, RI, and VT and wishes to opt-in to having their information shared for Health Information Exchange participation.

☐ **Health Information Exchange Opt-out.** Check this box if your patient resides in any other US state or territory and wishes to opt-out of participation in Health Information Exchange.

Signature of Ordering Provider	**Date**

ACCOUNT INFORMATION

GeneDx Account Number	**Account Name**	
Phone	Fax	
Address		
City	State	Zip Code
Ordering Provider Name		**Role/Title**
NPI	Phone Number	

Send Report Via: ☐ Fax ☐ Email ☐ Portal
Fax #/Email: _____

Additional Ordering Provider Name (optional)	**Role/Title**
NPI	

Send Report Via: ☐ Fax ☐ Email ☐ Portal
Fax #/Email: _____

SEND ADDITIONAL REPORT COPIES TO (optional)

Provider Name	GeneDx Acct#
Fax #/Email:	

ICD-10-CM CODES

ICD-10-CM Codes to support all test(s) ordered

Clinical Diagnosis	Age of Onset

PAYMENT OPTIONS *(Select One)*

◯ **INSURANCE BILL**
Select all that apply
☐ Commercial
☐ Medicaid
☐ Medicare
☐ Tricare
☐ CHAMPVA

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

Patient Status
Is this individual currently a Hospital Inpatient? ◯ Yes ◯ No

Name of Insurance Carrier		**Insurance ID#:**	
Relationship to Insured ◯ Self ◯ Spouse ◯ Child ◯ Other: _____			
Policy Holder's Name		Policy Holder's Date of Birth	
Referral/Prior Authorization # (please attach)		☐ Hold test for cost estimate and contact patient if estimate is >$250 (for in-network/ contracted commercial insurance only)	
Secondary Insurance Type:			
Insurance Carrier	Insurance ID #	Subscriber Name	Date of Birth
Relationship to Insured ◯ Self ◯ Spouse ◯ Child ◯ Other: _____			

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

◯ **INSTITUTIONAL BILL**
GeneDx Account #
Hospital/Lab Name
Place Sticker/Stamp Here

RARE DISORDERS TEST REQUISITION FORM

GeneDx

First Name	Last Name	Date of Birth

CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

Is this person affected: ○ Yes ○ No Clinical diagnosis: _____

Reason for testing: ☐ Diagnosis ☐ Presymptomatic diagnosis ☐ Carrier/Familial variant testing

Please check all that apply. This is not a substitute for submitting clinical records.

Pre/Perinatal History
- ☐ Cystic hygroma
- ☐ Growth delay
- ☐ Increased nuchal translucency
- ☐ Intrauterine growth retardation
- ☐ Nonimmune hydrops fetalis
- ☐ Multiple prenatal fractures
- ☐ Oligohydramnios
- ☐ Polyhydramnios

Structural Brain Abnormalities
- ☐ Abnormal myelination
- ☐ Abnormality of basal ganglia
- ☐ Abnormality of the corpus callosum
- ☐ Aplasia/hypoplasia of cerebellum
- ☐ Arnold Chiari malformation
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Lissencephaly
- ☐ Molar tooth sign on MRI
- ☐ Ventriculomegaly

Developmental/Behavioral
- ☐ Absent speech
- ☐ Attention deficit hyperactivity disorder
- ☐ Autistic behavior
- ☐ Behavioral abnormality
- ☐ Delayed fine motor development
- ☐ Delayed gross motor development
- ☐ Delayed speech & language development
- ☐ Developmental regression
- ☐ Global developmental delay
- ☐ Hyperactivity
- ☐ Intellectual disability
- ☐ Obsessive compulsive disorder
- ☐ Specific learning disability
- ☐ Stereotypy

Neurological
- ☐ Abnormality of nervous system
- ☐ Anosmia, congenital
- ☐ Ataxia
- ☐ Cerebral palsy
- ☐ Dystonia
- ☐ Encephalopathy
- ☐ Epileptic encephalopathy
- ☐ Familial or sporadic hemiplegic migraine
- ☐ Focal seizures
- ☐ Headaches
- ☐ Hyperreflexia
- ☐ Infantile spasms
- ☐ Peripheral neuropathy
- ☐ Reduced tendon reflexes
- ☐ Seizures
- ☐ Sensory neuropathy
- ☐ Spasticity
- ☐ Stroke-like episode(s)
- ☐ Tremors

Craniofacial/Dysmorphism
- ☐ Abnormal facial shape
- ☐ Cleft lip
- ☐ Cleft palate
- ☐ Craniosynostosis
- ☐ Downslanted palpebral fissures
- ☐ Epicanthus
- ☐ External ear malformation
- ☐ Facial asymmetry
- ☐ Frontal bossing
- ☐ High palate
- ☐ Hypertelorism
- ☐ Low set ears
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Micrognathia
- ☐ Retrognathia
- ☐ Short neck
- ☐ Synophrys
- ☐ Wide nasal bridge

Eye/Vision Abnormalities
- ☐ Abnormality of vision
- ☐ Anophthalmia
- ☐ Blue sclerae
- ☐ Cataracts
- ☐ Coloboma
- ☐ Ectopia lentis
- ☐ External ophthalmoplegia
- ☐ Microphthalmia
- ☐ Myopia
- ☐ Nystagmus
- ☐ Photophobia
- ☐ Ptosis
- ☐ Strabismus

Hearing Impairment
- ☐ Conductive hearing impairment ☐ bilateral ☐ unilateral
- ☐ Sensorineural hearing impairment ☐ bilateral ☐ unilateral
- ☐ Hearing impairment, mixed or unknown ☐ bilateral ☐ unilateral

Cardiac
- ☐ Abnormal heart morphology
- ☐ Aortic root dilation
- ☐ Arrhythmia
- ☐ Atrial septal defect
- ☐ Cardiomyopathy ☐ DCM ☐ HCM
- ☐ Coarctation of aorta
- ☐ Heart murmur
- ☐ Heterotaxy
- ☐ Hypertension
- ☐ Patent ductus arteriosus
- ☐ Tetralogy of Fallot
- ☐ Ventricular septal defect

Respiratory
- ☐ Asthma
- ☐ Bronchiectasis
- ☐ Pneumothorax
- ☐ Pulmonary fibrosis
- ☐ Recurrent upper respiratory infections
- ☐ Respiratory distress
- ☐ Respiratory insufficiency

Gastrointestinal
- ☐ Abnormality of the liver
- ☐ Aganglionic megacolon
- ☐ Cholestasis
- ☐ Congenital diaphragmatic hernia
- ☐ Constipation
- ☐ Diarrhea
- ☐ Duodenal stenosis/atresia
- ☐ Exocrine pancreatic insufficiency
- ☐ Failure to thrive
- ☐ Feeding difficulties
- ☐ Gastroesophageal reflux
- ☐ Gastrointestinal dysmotility
- ☐ Gastroschisis
- ☐ Hepatomegaly/Splenomegaly
- ☐ Hepatic fibrosis
- ☐ Inflammatory bowel disease
- ☐ Intestinal perforation
- ☐ Intrahepatic biliary atresia
- ☐ Laryngomalacia
- ☐ Nausea
- ☐ Pancreatitis
- ☐ Pyloric stenosis
- ☐ Tracheoesophageal fistula
- ☐ Vomiting

Musculoskeletal
- ☐ Abnormal connective tissue
- ☐ Abnormality of bone mineral density
- ☐ Abnormality of the ribs
- ☐ Abnormality of the upper limb
- ☐ Bowing of the long bones
- ☐ Bruising susceptibility
- ☐ Clinodactyly
- ☐ Ectrodactyly
- ☐ Fractures of the long bones
- ☐ Hyperostosis
- ☐ Hypertonia
- ☐ Hypotonia
- ☐ Limb joint contracture
- ☐ Overgrowth %ile: _____
- ☐ Pectus carinatum
- ☐ Pectus excavatum
- ☐ Polydactyly
- ☐ Short stature
- ☐ Skeletal dysplasia
- ☐ Small chest circumference
- ☐ Syndactyly
- ☐ TC ratio: _____
- ☐ Thoracic hypoplasia
- ☐ Vertebral abnormalities

Skin/Hair
- ☐ Abnormal blistering of the skin
- ☐ Abnormality of hair _____
- ☐ Abnormality of nail
- ☐ Alopecia
- ☐ Angiokeratoma
- ☐ Café-au-lait macules
- ☐ Dry skin
- ☐ Eczema
- ☐ Hyperextensible skin
- ☐ Hyperpigmentation of the skin
- ☐ Hypertrichosis
- ☐ Hypopigmentation of the skin
- ☐ Ichthyosis
- ☐ Recurrent skin infections
- ☐ Velvety skin (soft skin)
- ☐ Xanthomatosis

Genitourinary
- ☐ Abnormal renal biopsy: _____
- ☐ Abnormal urine analysis: _____
- ☐ Ambiguous genitalia
- ☐ Chronic kidney disease
- ☐ Cryptorchidism
- ☐ Cystic renal dysplasia
- ☐ Hydronephrosis
- ☐ Hypospadias
- ☐ Micropenis
- ☐ Nephrocalcinosis
- ☐ Nephrotic syndrome
- ☐ Nephrolithiasis
- ☐ Polycystic kidney disease
- ☐ Renal agenesis
- ☐ Renal insufficiency
- ☐ Renal tubular dysfunction/acidosis

Metabolic/Mitochondrial
(Attach relevant lab reports/values)
- ☐ Abnormal LDL-Cholesterol levels
- ☐ Abnormal newborn screen result: _____
- ☐ Abnormal plasma AA result: _____
- ☐ Abnormal urine OA result: _____
- ☐ Elevated CPK: _____
- ☐ Elevated hepatic transaminases
- ☐ Hyperglycemia
- ☐ Hypoglycemia
- ☐ Hypokalemia
- ☐ Increased serum pyruvate
- ☐ Lactic acidosis
- ☐ Vitamin D deficiency

Endocrine
- ☐ Amenorrhea
- ☐ BMI: _____
- ☐ Delayed puberty
- ☐ Diabetes insipidus
- ☐ Diabetes mellitus
- ☐ Ectopic calcification
- ☐ Elevated hemoglobin A1c
- ☐ Goiter
- ☐ Hypercalcemia
- ☐ Hyperthyroidism
- ☐ Hypophosphatemia
- ☐ Hypothyroidism
- ☐ Low alkaline phosphatase
- ☐ MODY: age of onset _____
- ☐ Pancreatic islet autoantibody negativity
- ☐ Rickets

Hematological or Immunological
- ☐ Anemia
- ☐ Bone marrow hypocellularity
- ☐ Immunodeficiency
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Recurrent infections
- ☐ Recurrent otitis media
- ☐ Thrombocytopenia
- ☐ Thromboembolism

Vascular System
- ☐ Aneurysm
- ☐ Arterial calcification
- ☐ Arterial dissection
- ☐ Arteriovenous malformation
- ☐ Lymphedema
- ☐ Stroke

Additional Clinical Findings:

RARE DISORDERS TEST REQUISITION FORM

GeneDx

First Name	Last Name	Date of Birth

FAMILY HISTORY

☐ No Known Family History ☐ Pedigree Attached ☐ Adopted

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

PREVIOUS GENETIC TESTING

Personal or family history of genetic testing ○ No ○ Yes (If yes, please complete all fields below)

Relation to patient (self, sibling, etc.), Genetic Test(s) and Result (e.g. positive, negative, etc.). If relative was tested at GeneDx, please also provide their accession #:

If patient or relative(s) were found to have a positive or VUS result on prior testing, please provide details below.
Indicate any Variants of Interest‡ via the checkbox below.

Relation (self, sibling, etc.)	Gene	Transcript #	c./p. (SNV) or exon # (CNV)	Build, coordinates (CNV)	Variant of Interest‡?
1					☐
2					☐
3					☐

Required for sequence variants: gene, c./p., transcript #
Required for CNVs: gene, transcript #, exon # OR build, coordinates

Abnormal karyotype, FISH, or other results:

‡ For certain tests, GeneDx **may** be able to specifically comment upon the presence or absence of previously identified variant(s) of interest in the report. Complete variant information must be provided in the table above at the time the test order is placed. If you do not complete the table above and check off that a previously identified variant is a variant of interest, it will not be possible to comment upon the presence or absence of the variant in the report retrospectively. This service is not applicable to targeted variant testing.

TARGETED VARIANT TESTING

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

☐ Known Familial Variant(s) in a Nuclear Gene ☐ Confirmation of Variant Identified in Research Lab ☐ Targeted Mosaic Variant Testing*
☐ Known Familial Copy Number Variant(s) ☐ Known mtDNA Variant(s) Testing

**Insurance Billing NOT Accepted; Patient Bill or Institutional Bill MUST be selected on page 1*

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 VARIANT

Number of Variants: _____

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

RARE DISORDERS TEST REQUISITION FORM

GeneDx

First Name	Last Name	Date of Birth

TEST MENU

TEST CODE	TEST NAME	TEST CODE	TEST NAME
DERMATOLOGIC DISORDERS			
☐ 714	Birt-Hogg-Dube syndrome (*FLCN*)	☐ 708	Slice — Congenital Ichthyosis
☐ 553	Incontinentia Pigmenti — Full Gene Sequencing and Common Deletion (*IKBKG/NEMO*)	☐ 707	Slice — Epidermolysis Bullosa (EB)
☐ 713	Hereditary Leiomyomatosis and Renal Cell Cancer (*FH* sequencing and del/dup analysis)	☐ 188	Hermansky-Pudlak Syndrome: Puerto Rican Mutations (*HPS1, HPS3* sequencing and del/dup analysis)
DYSMORPHOLOGY AND MULTIPLE CONGENITAL ANOMALIES			
☐ 962	Neurofibromatosis Type 1 Panel	☐ 963	Neurofibromatosis Type 2 Panel
☐ J660	Neurofibromatosis Type 1 (*NF1* single gene sequencing and del/dup analysis)	☐ TA06	Noonan and Comprehensive RASopathies Panel
ENDOCRINE DISORDERS			
☐ 676	Hypogonadotropic Hypogonadism Panel	☐ 674	Maturity-Onset Diabetes of the Young (MODY) Panel
HEMATOLOGIC DISORDERS			
☐ TB47	Dyskeratosis Congenita Panel	☐ 109	Shwachman-Diamond Syndrome (*SBDS* gene sequencing)
☐ 107	Dyskeratosis Congenita, Autosomal Dominant (*TERC* gene sequencing)		
IMMUNOLOGIC DISORDERS			
☐ T990	Autoimmune Lymphoproliferative Syndrome (ALPS) Panel	☐ TA48	Severe Congenital Neutropenia, Autosomal Dominant (*ELANE/ELA2*)
☐ T989	Chronic Granulomatous Disease (CGD) Panel		
NEUROLOGIC DISORDERS			
☐ 526	Cerebral Cavernous Malformations Panel	☐ 552	X-linked Hydrocephalus, X-linked Spastic Paraplegia, Masa, Crash Syndrome (*L1CAM*)
☐ TB51	Comprehensive Holoprosencephaly Panel		
PULMONARY DISORDERS			
☐ T829	Cystic Fibrosis/Congenital Bilateral Absence of The Vas Deferens (*CFTR* sequencing and del/dup analysis)	☐ TB46	Primary Ciliary Dyskinesia Panel
RENAL AND GASTROINTESTINAL DISORDERS			
☐ TG21	Alport Syndrome Panel	☐ TH01	Nephrolithiasis and Nephrocalcinosis Panel
☐ TG23	Cystic Kidney and Liver Diseases Panel	☐ TG22	Polycystic Kidney Disease Panel
☐ TG98	Hypokalemia and Related Disorders Panel	☐ TG90	Primary Hyperoxaluria Panel
☐ TG99	Nephrotic Syndrome/Focal Segmental Glomerulosclerosis Panel		
REPRODUCTIVE DISORDERS			
☐ 522	FMR1-associated Premature Ovarian Failure, CGG Repeat Analysis Only (*FMR1*)	☐ 677	Premature Ovarian Failure Panel
RHEUMATOLOGIC DISORDERS			
☐ 367	Periodic Fever Syndromes Panel: Familial Hibernian fever/TRAPS; Familial Mediterranean fever; Hyper-IgD syndrome; Muckle Wells/familial cold urticaria, NOMID; Cyclic neutropenia; PAPA syndrome; Majeed syndrome		

(Continue on the next page)

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

RARE DISORDERS TEST REQUISITION FORM

First Name	Last Name	Date of Birth

TEST MENU *(continued)*

TEST CODE	TEST NAME	TEST CODE	TEST NAME
SKELETAL DISORDERS			
☐ T992	Autosomal Dominant Osteogenesis Imperfecta Panel	☐ TA42	Limb Abnormalities and Reduction Defects Panel
☐ TA40	Craniosynostosis Panel	☐ J797	Osteogenesis Imperfecta Panel
☐ TA41	Ectrodactyly/Split Hand-Split Foot Malformation Panel	☐ TA43	Skeletal Dysplasia Panel
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: _____			
WRITE-IN TEST SELECTION			
☐ Test Code: _____	Test Name: _____		

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

INFORMED CONSENT

GeneDx

First Name	Last Name	Date of Birth

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

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

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

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

WHAT IS TRIO/DUO-BASED GENETIC TESTING?
For some genetic tests, including samples from the biological parents and/or other biological relatives along with the patient's sample can help with the interpretation of the test results. These tests are often referred to as "trio tests" since they typically include samples from the patient and both parents.

Samples from relatives should be submitted with the patient's sample. Clinical information must be provided for the patient and any relative who submits a sample.

I understand that GeneDx will use the relative sample(s) when needed for the interpretation of my test results and that my test report may include clinical and genetic information about a relative when it is relevant to the interpretation of the test results. I further understand that relatives will not receive an independent analysis of data nor a separate report.

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

PATIENT CONFIDENTIALITY AND GENETIC COUNSELING
It is recommended that I receive genetic counseling before and after having this genetic test. I can find a genetic counselor in my area at www.nsgc.org. Further testing or additional consultations with a healthcare provider may be necessary.

To maintain confidentiality, test results will only be released to the referring healthcare provider, the ordering laboratory, to me, to other healthcare providers involved in my care, diagnosis and treatment, or to others with my consent or as permitted or required by law. Federal laws prohibit unauthorized disclosure of this information. More information can be found at: www.genome.gov/10002077

SAMPLE RETENTION
After testing is complete, my sample may be de-identified and be used for test development and improvement, internal validation, quality assurance, and training purposes. GeneDx will not return DNA samples to you or to referring healthcare providers, unless specific prior arrangements have been made.

I understand that samples from residents of New York State will not be included in the de-identified research studies described in this authorization and GeneDx will not retain them for more than 60 days after test completion, unless specifically authorized by my selection. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language. GeneDx will not perform any tests on the biological sample other than those specifically authorized.

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

EPILEPSY PARTNERSHIP PROGRAM PARTICIPATION
I understand that GeneDx will send de-identified test results data, excluding ACMG secondary findings, to third parties for research or commercial purposes and that GeneDx is compensated for the provision of testing services and for data sharing with third parties that is compliant with applicable law. At no time will GeneDx share any patient personally identifiable information. GeneDx may share contact information for providers listed on the Test Requisition Form with third parties.

INFORMED CONSENT

First Name	Last Name	Date of Birth

PATIENT RECONTACT FOR RESEARCH PARTICIPATION
GeneDx may collaborate with other 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, GeneDx may contact my healthcare provider for research purposes, such as the development of new testing, drug development, or other treatment modalities. In some situations, such as if my healthcare provider is not available, I may be contacted directly. I can opt out of being contacted directly regarding any of the above activities by having my healthcare provider check the box for Patient Research Opt-Out. 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 to (my/my child's) heirs.

EXOME/GENOME SEQUENCING SECONDARY FINDINGS

- Applicable only for full exome sequencing and genome sequencing tests
- Does not pertain to *Xpanded®* or Slice tests

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

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

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

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

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

LIMITATIONS
Pathogenic and/or likely pathogenic variants may be present in a portion of the gene not covered by this test and therefore are not reported. The absence of reportable secondary findings for any particular gene does not mean there are no pathogenic and/or likely pathogenic variants in that gene. Pathogenic variants and/or likely pathogenic variants that may be present in a relative, but are not present in the proband, will not be identified nor reported. Only changes at the sequence level will be reported in the secondary findings report. Larger deletions/duplications, abnormal methylation, triplet repeat or other expansion variants, or other variants not routinely identified by clinical exome and genome sequencing will not be reported.

FINANCIAL AGREEMENT AND GUARANTEE
For insurance billing, I understand and authorize GeneDx to bill my health insurance plan on my behalf, to release any information required for billing, and to be my designated representative for purposes of appealing any denial of benefits. I irrevocably assign to and direct that payment be made directly to GeneDx.

I understand that my out-of-pocket costs may be different than the estimated amount indicated to me by GeneDx as part of a benefit investigation. I agree to be financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. If my insurance provider sends a payment directly to me for services performed by GeneDx on my behalf, I agree to endorse the insurance check and forward it to GeneDx within 30 days of receipt as payment towards GeneDx's claim for services rendered.

By signing this form: (i) I acknowledge that I have read or have had read to me the GeneDx Informed Consent document, and understand the information regarding genetic testing; (ii) I have had the opportunity to ask questions about the testing, the procedure, the risks, and the alternatives; (iii) I authorize GeneDx to perform genetic testing as ordered; (iv) I understand that, for tests that evaluate data from multiple family members concurrently, test results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their healthcare providers; (v) if at any time I or my provider provide an email address or mobile phone number at which I may be contacted, I consent to receiving email or text messages from GeneDx; and (vi) I understand that this consent applies to all future communications unless I request a change in writing.

- ☐ **Secondary Findings Opt-out.** Check this box if you do not wish to receive ACMG secondary findings (Full Exome Sequencing and Genome Sequencing Tests ONLY; not for *Xpanded®* or Slice tests).
- ☐ **New York Retention Opt-in.** 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 within 60 days, and it cannot be used for test development studies.
- ☐ **Patient Research Opt-out.** Check this box if you wish to opt out of being contacted for research studies.
- ☐ **Health Information Exchange Opt-in.** Check this box if you reside in CA, FL, MA, NV, NY, RI, and VT and wish to opt-in to my health information to be shared for Health Information Exchange participation.
- ☐ **Health Information Exchange Opt-out.** Check this box if you reside in any other US state or territory and wish to opt-out of participation in Health Information Exchange.

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