

Age of Onset

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

PATIENT INFORMATION						
irst Name	Last Name					
ex Assigned at Birth: Male Female atient Karyotype (if known):	Date of Birth (mm	n/dd/yy)				
ender Identification (optional): mail	_					
ddress						
Sity	State	Zip Code				
hone (mobile preferred)	Is this patient dec Deceased Date:	eased? O Yes O No				
	IFORMATION					
ate Sample Collected (mm/dd/yy)	Medical Record #	•				
Blood Buccal Swab Other (specify	y source):					
Treatment-related RUSH (optional) leason: Transplantation Pregnancy latient has had a blood transfusion Yes		r: t Transfusion:				
2-4 weeks of wait time is required for some						
atient has had an allogeneic bone marrov ibroblasts are required for patients who had ee www.genedx.com/specimen-requireme	d an allogeneic bone	~				
ratient has a personal history of a hemato	logic malignancy or	_				
Yes (specify diagnosis) yes, please call the lab to discuss with a gen	etic counselor the mo	. <b>()No</b> ost appropriate sample type.				
ORDERING PROV	IDER ATTESTA	TION				
By signing this form, the ordering provider of GeneDx to perform the testing indicated; (ii authorized by law to order the test(s) reque Requisition Form ("TRF") are reasonable and treatment of a disease, illness, impairment, results will determine the patient's medical potient's condition on this date of service; (authorized to make decisions for the patier any relatives', when applicable, has been sitesting, and has consented to undergo gendiagnosis codes are indicated to the highereimbursement from any third party, includ programs if testing is covered by GeneDx and GeneDx may share contact information for providers listed on the this order with third pand potential clinical trial or study opporture family member authorized to be contacted number provided for this and future testing	) he/she is the orderisted; (iii) any test(s) d medically necessal symptom, syndrome management and tr v) the patient or the it (collectively, the "pupplied with informat letic testing; (vi) the fst level of specificity; ling but not limited to not will inform the pat the ordering provide oarties regarding the nities; and (ix) the pat I via the email address	ing provider and is requested on this Test ry for the diagnosis or e or disorder; (iv) the test reatment decisions of this individual/family member atient"), in addition to tion regarding genetic 'ull and appropriate (vii) he/she will not seek is federal healthcare tient of the same; (viii) r and other healthcare requested genetic testing tient or the individual/				
Secondary Findings Opt-out. By checki not wish to receive ACMG secondary fin Sequencing Tests ONLY; not for Xpande	ndings. (Full Exome Se					
New York Retention Opt-In. By checking York State resident who gives permission longer than 60 days after testing has be	on for GeneDx to retai					
Patient Research Opt-Out. By checking opt out of being contacted for research		at the patient wishes to				
☐ Health Information Exchange Opt-in. C FL, MA, NV, NY, RI, and VT and wishes to a Health Information Exchange participat	opt-in to having their					
Health Information Exchange Opt-out. other US state or territory and wishes to						

GeneDx Account Number	Account Name	е
Phone	Fax	
Address		
City	State	Zip Code
Ordering Provider Name		Role/Title
NPI	Phone Number	r
Send Report Via: ☐ Fax ☐ Email ☐ Fax #/Email:	Portal	
Additional Ordering Provider Name (	optional)	Role/Title
NPI		
Send Report Via: ☐ Fax ☐ Email ☐ Fax #/Email:	]Portal	
SEND ADDITIONAL REPORT COPIES TO	(optional)	
Provider Name	GeneDx Acct#	
Fax #/Email:	ļ.	

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

Clinical Diagnosis

	PAYMENT O	PTIONS (Sele	ect One)				
O INSURANCE BILL Select all that apply  Commercial	Patient Status  OHospital outpatient OHospital inpatient; Date of Discharge: ONOt a hospital patient						
□ Medicaid □ Medicare	Name of Insurance	ce Carrier	Insurance ID#:				
☐ Tricare ☐ CHAMPVA	Relationship to Insured  OSelf OSpouse OChild OOther:						
FOR ALL INSURANCE PROVIDE FRONT	Policy Holder's Na	me	Policy Holder's Date	of Birth			
AND BACK COPY OF CARD(S)	Referral/Prior Auth (please attach)	norization #	Hold test for cost estimate and contact patient				
	Secondary Insurance Type:		if estimate is >\$250 (commercial insurance only)				
	Insurance Carrier	Insurance ID #	Subscriber Name	Date of Birth			
	Relationship to Insured OSelf OSpouse Ochild Oother:						
O 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.						
	ture						
O INSTITUTIONAL BILL	GeneDx Account #		- Place Sticker/Stamp Here				
	Hospital/Lab Name			•			

Signature of Ordering Provider

Date



First Name		Last Name			Date of Birth		
		[			l l		
		XOMEDX	(® TESTI	ING OPTI	ons		
TEST CODE	TEST NAME		TEST C	ODE TES	ST NAME		
□ 561a	XomeDx® - Trio*		□ 561a 561m	n   '•	meDx® Plus - Trio*, consists of two separate tests†: 561a XomeDx® - Trio; and 561m Mitochondrial Genome Sequencing & Deletion Testing		
□ 561e	XomeDx® - Duo*		□ 561e 561m	ո   •	eDx® Plus - Duo*, consists of two separate tests†: 561e <i>XomeDx®</i> - Duo; and 561m Mitochondrial Genome Sequencing & Deletion Testing		
□ 561b	XomeDx® - Proband		□ 561b 561m	n   '•	neDx® Plus - Proband, consists of two separate tests†: 561b XomeDx® - Proband; and 561m Mitochondrial Genome Sequencing & Deletion Testing		
		Family Member Samples to be Inclu- nome) will be billed and reported sep		sting section	below		
	The second secon	XOMEDX® RE	•	STING O	PTIONS		
TEST CODE	TEST NAME		TEST C	ODE TES	ST NAME		
□ 522	Reflex to FMR1 CGG Repeat	Analysis after exome	<b>910</b>	Ref	lex to Chromosomal Microarray (MicroarrayDx) after exome		
			•	'			
		FAMILY MEMBER SAM	PLES TO	D BE INCL	UDED IN TESTING		
codes may re	equire adjusting to appropria		nber sam		ITHIN 3 WEEKS FOR INCLUSION IN THE PROBAND'S TEST. Ordered test /ed. A change in the ordered test will impact billing, including prior		
	First Name	Last Name	DOB		O Asymptomatic O Symptomatic		
Biological Mother					O At GeneDx (Accession #:) O Not available O To be sent within 3 weeks		
	First Name	Last Name	DOB		O Asymptomatic O Symptomatic		
Biological Father					O At GeneDx (Accession #:) O Not available O To be sent within 3 weeks		
	Relationship to Proband				O Not available O to be sent within 3 weeks		
Otner	First Name Last Name DOB		DOB		O Asymptomatic O Symptomatic		
Biological Relative					O At GeneDx (Accession #:)		
					O Not available O To be sent within 3 weeks		
		CUSTOM SI	LICE TES	STING OF	TIONS		
TEST CODE	TEST NAME			TEST COD			
	Slice - Single Gene (1 gene)			Slice - Xpanded® (>150 genes, Proband or Trio*)			
☐ 706	Approved Slice ID:  Slice - Multi-Gene (2-150 genes)  Approved Slice ID:						
	Approved Slice ID:  test is ordered, please fill out the	Family Member Samples to be Inclu	uded in Tes	s <i>ting</i> section	above		
SKIN DISORDER SLICES							
□ 707 Slice - Epidermolysis Bullosa (EB) □ 708 Slice - Congenital Ichthyosis							
□ 707	Slice - Epidermolysis Bullos	a (EB)		_ , 00	Slice - Congenital Ichthyosis		
707	Slice - Epidermolysis Bullos	a (EB)  REANALYSIS OF 3			,		
	ons are only appropriate if the pat	REANALYSIS OF 2	XOMED	X® TESTII	,		
These test option	ons are only appropriate if the pat	REANALYSIS OF X	XOMED:	X® TESTII	NG OPTIONS  GeneDx. We recommend waiting at least one year from original/prior analysis		
These test optic before ordering	ons are only appropriate if the pat g a Reanalysis.	REANALYSIS OF ) ient previously had a XomeDx* test (i	XOMED:	X® TESTIII analysis) at Reason for	NG OPTIONS  GeneDx. We recommend waiting at least one year from original/prior analysis		

ACMG secondary findings, as discussed in the Informed Consent and Authorization Form, are only returned for the patient if an XomeDx\* test (full exome analysis) is completed.

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 and list of genes 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.

☐ Test Code:



First Name		llast	Name				Date of Birth	
		2400						
			FAN	MILY HISTORY*				
	*T	his section i	s not intended fo	r ordering a targeted	variant te	sting report.		
□ No Known Family History	□P€	edigree Att	ached	☐ Adopted				
Relationship	Maternal	Paternal		I	Relevant H	listory		Age at Dx
1	0	0						
2	0	0						
3	0	0						
		•						
	*T	his section i		GENETIC TESTING ordering a targeted		sting report.		
Personal or family history of genetic testing ONO OYes (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 four Indicate any Variants of Interest	nd to have a p via the check	ositive or VL dox below.	S result on prior	testing, please provide	e details b	elow.		
Relation (self, sibling, etc.)	Gene	Transcrip	t# c./	p. (SNV) or exon # (C	NV)	Build, coord	dinates (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.

(Continue to the next page)



First Name Last Name Date of Birth

CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)  Relevant clinical records are required at the time of sample submission to ensure the information is included in data analysis.						
Genes of interest (limit to 10):						
Differential diagnosis:						
Due / Devise other Utintons	Neural aging Findings	Hansing languing out				
Pre/Perinatal History	Neurological Findings	Hearing Impairment  ☐ Abnormal newborn screen:				
□ Cystic hygroma □ Diaphragmatic hernia	<ul><li>□ Abnormality of nervous system</li><li>□ Ataxia</li></ul>	☐ Conductive hearing impairment				
☐ Encephalocele	☐ Cerebral palsy	☐ Sensorineural hearing impairment				
☐ Growth delay	□ Chorea	_ consenied at nearing impairment				
☐ Increased nuchal translucency	☐ Cortical visual impairment	Endocrino Findingo				
☐ Intrauterine growth retardation	□ Dementia '	Endocrine Findings				
□ Nonimmune hydrops fetalis	□ Dysarthria	□ Delayed puberty				
☐ Oligohydramnios	□ Dyskinesia	□ Diabetes insipidus □ Diabetes mellitus				
□ Omphalocele	□ Dysphasia	☐ Hyperthyroidism				
Polyhydramnios	□ Dystonia	☐ Hypophosphatemia				
Prematurity GA:	☐ Encephalopathy	☐ Hypothyroidism				
□ Prolonged neonatal jaundice	□ Headaches □ Hemiplegia	☐ Maturity-onset diabetes of the young				
	☐ Infantile spasms	□Rickets				
Structural Brain Abnormalies	☐ Migraines					
☐ Abnormal myelination	□ Myoclonus	Respiratory Findings				
□ Abnormality of basal ganglia	, □ Parkinsonism	Asthma □ Asthma				
☐ Abnormality of brainstem	□ Peripheral neuropathy	☐ Bronchiectasis				
Abnormality of periventricular white matter	☐ Seizures	☐ Hyperventilation				
☐ Abnormality of the corpus callosum	☐ Sensory neuropathy	☐ Hypoventilation				
□ Aplasia/hypoplasia of cerebellar vermis □ aplasia/hypoplasia of cerebellum	□ Spasticity	□ Pneumothorax				
☐ Arnold chiari malformation	Syncope	☐ Pulmonary fibrosis				
☐ Cerebellar atrophy	□ Tremors □ Vertigo	□ Respiratory insufficiency				
☐ Heterotopia (periventricular nodular	□ vertigo					
heterotopia)		Hematologic or Immunologic Findings				
□ Holoprosencephaly	Craniofacial/Dysmorphism	☐ Allergic rhinitis				
☐ Hydrocephalus	□ Abnormal facial shape (dysmorphic	□ Anemia				
Leukodystrophy	features) specify:	☐ Immunodeficiency				
Lissencephaly	Brachycephaly	☐ Neutropenia				
□ Pachygyria	☐ Cleft lip and/or palate	□ Pancytopenia				
□ Polymicrogyria	<ul><li>□ Coarse facial features</li><li>□ Craniosynostosis</li></ul>	☐ Recurrent infections				
□ Ventriculomegaly	☐ Macrocephaly	□ Thrombocytopenia				
	☐ Microcephaly					
Developmental/Behavioral Findings	☐ Short neck	Skin/Hair Findings				
☐ Absent speech	Synophrys	☐ Abnormal blistering of the skin				
□ Aggressive behavior	, , ,	☐ Abnormality of nail				
□ Anxiety	Eva Defeate Mision	□ Alopecia ´				
Autistic behavior	Eye Defects/Vision	☐ Anhidrosis				
Cognitive impairment	☐ Abnormality of vision	□ Café-au-lait macules				
☐ Delayed speech & language development☐ Developmental regression	□ Anophthalmia □ Cataracts	Coarse hair				
□ Dysarthria	□ Coloboma	□ Cutis laxa				
Gait disturbance	☐ Corneal opacity	□Eczema □Hemangiomas				
☐ Global developmental delay	□ Ectopia lentis	☐ Hyperextensible skin				
Hyperactivity	□ External ophthalmoplegia	☐ Hyperpigmentation of the skin				
□Incoordination	□Microphthalmia	☐ Hypohidrosis				
☐ Intellectual disability	□Myopia	☐ Hypopigmentation of the skin				
Learning disability	□Nystagmus	☐ Ichthyosis				
☐ Memory impairment	Optic atrophy	☐ Skin rash				
☐ Sleep disturbance	□ Optic neuropathy	□ Sparse hair				
□ Stereotypy	□ Ptosis □ Retinal detachment	☐ Telangiectasia				
	☐ Retinitis pigmentosa	□ Vascular skin abnormality				
	☐ Strabismus	□ Velvety skin				



First Name Last Name Date of Birth

CLINICAL INFORMA	TION (DETAILED MEDICAL RECORDS MUST E	BE ATTACHED)
Cardiac Findings	Musculoskeletal Findings	Vascular System
☐ Abnormal heart morphology	☐ Abnormal connective tissue	☐ Aneurysm
□ Amyloidosis	☐ Abnormal form of the vertebral bodies	☐ Arterial calcification
☐ Aortic root dilation	☐ Abnormality of the ribs	☐ Arterial dissection
☐ Arrhythmia	☐ Arachnodactyly	☐ Arterial tortuosity
☐ Atrial septal defect	□ Arthralgia	☐ Arteriovenous malformation
☐ Bicuspid aortic valve	□ Arthrogryposis	□ Epistaxis
□ Bradycardia	☐ Bruising susceptibility	□ Lymphedema
☐ Coarctation of aorta	Clinodactyly	☐ Pulmonary hypertension
□ Dilated cardiomyopathy	Decreased muscle mass	□ Stroke
□ Heterotaxy	☐ Ectrodactyly	
☐ Hypertension	□ Exercise intolerance	
☐ Hypertrophic cardiomyopathy	□Fatigue	Cancer
☐ Mitral valve prolapse	□Hemihypertrophy	□Type:
□ Noncompaction cardiomyopathy	□Hypertonia	Location:
□ Patent ductus arteriosis	□Hypotonia	Location:
□ Patent foramen ovale	☐ Joint hypermobility	Age of onset:
□ Prolonged QTc interval	☐ Muscle weakness	
□ Sudden death	□ Myalgia	
☐ Tetralogy of Fallot	☐ Myopathic facies	
☐ Ventricular septal defect	☐ Myopathy	Other Testing/Imaging
□ Ventricular tachycardia	☐ Osteoarthritis	(Please provide copy or report if possible)
	□ Osteopenia	☐ Echo:
	Pain	
Gastrointestinal Findings	Pectus carinatum	□ EEG:
Constipation	Pectus excavatum	□ EMG:
☐ Diarrhea	Polydactyly	☐ MRI:
☐ Duodenal stenosis/atresia	Recurrent fractures	☐ Muscle Biopsy:
Exocrine pancreatic insufficiency	□ Rhabdomyolysis □ Scoliosis	Ultrasound:
☐ Failure to thrive	☐ Short stature	□ V-raye:
☐ Feeding difficulties	☐ Skeletal dysplasia	□ X-rays:
☐ Gastroesophageal reflux	☐ Syndactyly	
□Hepatomegaly	☐ Tall stature	
☐ Inflammatory bowel disease	_ ran otataro	
□ Intrahepatic biliary atresia		Additional Clinical Findings:
□ Laryngomalacia	Makabalia Biratia an	
□Nausea	Metabolic Findings (Attached relevant lab reports/values)	
□ Pancreatitis		
☐ Pyloric stenosis	☐ Abnormal activity of mitochondrial	
□ Splenomegaly	respiratory chain	
☐ Tracheoesohageal fistula	Abnormal newborn screen:	
□ Vomiting	☐ Abnormality of mitochondrial metabolism☐ Elevated CPK	
	<del>_</del>	
	☐ Elevated hepatic transaminase	
Genitourinary Findings	☐ Hyperammonemia	
□ Ambiguous genitalia	□ Hyperglycemia □ Hypoammonemia	
☐ Cryptorchidism	☐ Hypoglycemia	
Cystic renal dysplasia	☐ Increased serum pyruvate	
☐ Horseshoe kidney	□ Lactic acidosis	
☐ Hydronephrosis	☐ Plasma AA:	
☐ Hypospadias	Urine OA:	
□ Inguinal hernia		
□ Micropenis		
□Nephrolithiasis		
□ Polycystic kidney disease		
□ Renal agenesis		
□ Umbilical hernia		



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. <u>Positive</u>: 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. <u>Negative</u>: 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. <u>Variant of Uncertain Significance (VUS)</u>: 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. <u>Unexpected Results (ACMG Secondary Findings)</u>: 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



irst 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.

#### IMITATIONS

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.

		•				
Sign	ature of Relative B/Legal Guardian	Relative B Relationship to Patient	Date			
	ature of Relative A/Legal Guardian	Relative A Relationship to Patient	Date			
Sign	ature of Patient/Legal Guardian (required)		Date			
	<b>Health Information Exchange Opt-out.</b> Check this box if you reside in any o Exchange.	ther US state or territory and wish to opt-out of participa	tion in Health Information			
	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.					
	Patient Research Opt-out. Check this box if you wish to opt out of being co	ntacted for research studies.				
	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.					
	<b>Secondary Findings Opt-out.</b> Check this box if you do not wish to receive Ar ONLY; not for <i>Xpanded®</i> or Slice tests).	CMG secondary findings (Full Exome Sequencing and Ge	nome Sequencing Tests			
and and fan	signing this form, I acknowledge as the patient or relative being tested that I d understand the information regarding molecular genetics testing. I have ho d the alternatives. By signing this form, I authorize GeneDx to perform genetic nily members concurrently, test results from these family members may be il lividuals and their healthcare providers.	ad the opportunity to ask questions about the testing, the testing as ordered. I understand that, for tests that evalu	e procedure, the risks, uate data from multiple			