

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

PATIENT IN	FORMATION				
First Name	Last Name				
rirst name	Last Name				
Sex Assigned at Birth: OMale OFemale	Date of Birth (mm/dd/	/уу)			
Patient Karyotype (if known):					
Gender Identification (optional): Email					
Erridii					
Address					
City	State	Zip Code			
Primary Phone	Is this patient decease	d? OYes ONo			
	Deceased Date:				
	FORMATION				
Date Sample Collected (mm/dd/yy)	Medical Record #				
○ Blood ○ Buccal Swab ○ Other (specif	y source):				
Treatment-related RUSH (optional)	•				
Reason: O Transplantation O Pregnancy					
Patient has had a blood transfusion OYes (2-4 weeks of wait time is required for some t		ınsfusion:			
Patient has had an allogeneic bone marrow		)			
Fibroblasts are required for patients who had See www.genedx.com/specimen-requiremen	an allogeneic bone mari				
Patient has a personal history of a hematolo	gic malignancy or disec	ise			
O Yes (specify diagnosis)		ON₀			
If yes, please call the lab to discuss with a gene	etic counselor the most ap	opropriate sample type.			
PATIENT	CONSENT				
By signing this form, I acknowledge as the patient or relative being tested that I have read or have had read to me the GeneDx Informed Consent document at the end of this test requisition form, and understand the information regarding molecular genetics testing. I have had the opportunity to ask questions about the testing, the procedure, the risks, and the alternatives. By signing this form, I authorize GeneDx to perform genetic testing as ordered. 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.					
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.					
Check this box if you wish to opt out of b		arch studies			
Signature of Patient/Legal Guardian (requir	ed)	Date			
Signature of Relative A/Legal Guardian	Date				
Signature of Relative B/Legal Guardian		Date			
FOR COMMERCIAL INSURANCE ONLY: By entering my preferred contact information to be with a pertinget of the patients financial at					
me with an estimate of the patient's financial responsibility for testing. Data rates may appl  Email (required)*  Mobile Number					
Email (required)*	Mobile Number				
Email (required)*  *Contact information provided must be for the					

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:						

## 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 Ordering Provider

ICD-10-CM CODES				
ICD-10-CM Codes				
Clinical Diagnosis	Age of Onset			

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



First Name Last Name Date of Birth

CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)					
Is this person affected? O Yes O No Clinic	cal diagnosis:	Age of Onset:			
□Unilateral □Bilateral					
Intraocular Pressure:	ERG Results:				
Audiogram (dB): Left Right	ERG Results:				
• • • = = • = =					
Reason for testing: 🗆 Diagnosis 🗀 Presymptome	atic diagnosis 🛮 Carrier/Familial Variant Testing				
Please check all that apply. This is not a substitute	for submitting clinical records.				
Eye/Vision Abnormalities	Hearing Impairment				
☐ Abnormality of vision	☐ Abnormal newborn screen:				
□ Aniridia ´					
□ Anophthalmia	☐ Aminoglycoside-induced hearing loss				
□ Astigmatism	☐ Conductive hearing impairment				
□ Blue sclerae	□ Bilateral				
Cataracts	Unilateral				
Coloboma	☐ Enlarged vestibular aqueduct				
☐ Corneal arcus	Hearing impairment, mixed or unknown				
□ Ectopia lentis	□ Bilateral □ Unilateral				
□Esotropia □External ophthalmoplegia	☐ Morphological abnormality of the inner ear				
☐ External ophthalmobilegia ☐ Glaucoma	Sensorineural hearing impairment				
☐ Hyperopia	☐ Bilateral				
☐ Hypoplasia of the fovea	☐ Unilateral				
☐ Keratoconus/anterior lenticonus	☐ Tinnitus				
□ Microphthalmia					
□ Myopia	Immunologic Issues				
□ Night blindness	□ Recurrent infections				
□Nystagmus	Recurrent otitis media				
□ Optic atrophy					
□ Photophobia	Neurological Findings				
□ Ptosis □ Retinal detachment	□ Vocal cord paresis				
Retinitis pigmentosa	U vocal cola parcsis				
□ Strabismus	Ponal Findings				
☐ Visual impairment	Renal Findings				
papa	☐ Renal cysts ☐ Other renal:				
Craniofacial/Dysmorphism	Other rendi.				
	Chin / Univ Eindings				
□ Abnormal facial gestalt (dysmorphic features)	Skin/Hair Findings				
External ear malformation	☐ Allergic dermatitis ☐ Anhidrosis/hypohidrosis				
☐ Macrocephaly	Cutaneous photosensitivity				
☐ Microcephaly	□ Dermatitis				
	☐ Hypopigmentation of the skin				
Developmental/Bohavious	□ Ichthyosis				
Developmental/Behavioral	Skin fragility/blistering				
□ Absent speech □ Delayed fine motor development	☐ Sparse hair				
☐ Delayed fine Motor development					
Delayed gross motor development  Delayed speech & language development	Attach pedigree and/or include additional				
☐ Failure to thrive	clinical information:				
□ Incoordination					
☐ Intellectual disability					
,					
	·				



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First Name		Last Name Date of Birth					
			FAMILY I	HISTORY			
□ No Known Family History	□P€	edigree Att	ached	□ Adopted			
Relationship	Maternal	Paternal		Relevant F	listory		Age at Dx
1	0	0					
2	0	0					
3	0	0					
			PREVIOUS GEN	NETIC TESTING			
Personal or family history of g	enetic test	ing ON		ease complete all field	ds below)		
Relation to patient (self, sibling, etc	:.), Genetic T	est(s) and R	esult (e.g. positive, nego	ative, etc.). If relative was	tested at GeneDi	x, please also provide their a	ccession #:
	,	.,				,	
If patient or relative(s) were found Indicate any Variants of Interest‡ v			IS result on prior testing	, please provide details b	elow.		
Relation (self, sibling, etc.)	Gene	Transcrip	ot # c./p. (SN	V) or exon # (CNV)	Build, o	coordinates (CNV)	Variant of Interest‡?
1							
2							
3							
Required for sequence variants: gene, Required for CNVs: gene, transcript #, e							
Abnormal karyotype, FISH, or other	results:						
‡ For certain tests, GeneDx <b>may</b> 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: O Affected/Symptomatic O 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:							
VARIANT INFORMATION (please fill out the below information if family member report is not included)  Number of Variants:							
Gene	Codine	g DNA (c./m.)	A (c./m.) Amino Acid (p.) Transcript (NM#)		Transcript (NM#)		
Gene	Coding	g DNA (c./m.)		Amino Acid (p.)		Transcript (NM#)	
COPY NUMBER VARIANT						Number of Variants:	
Gene(s)	Exon #	ŧ		Coordinates		Genome Build	
Gene(s)	Exon #	ŧ		Coordinates		Genome Build	



First Name	Last Name	Date of Birth			
	TEST	MENU			
TEST CODE	T CODE TEST NAME TEST CODE TEST NAME				
HEARING LO	DSS TESTS				
☐ TA49			Hearing Loss Panel		
	common GJB6 deletions)	☐ TL50	Waardenburg Syndrome Panel		
OPHTHALM	OLOGY MULTI-GENE PANELS				
□ J958	Cataract Panel	☐ TA02	Stickler Syndrome Panel		
☐ TH12	Leber Hereditary Optic Neuropathy (LHON) Panel	☐ TG49	Stargardt Panel		
OPHTHALMOLOGY SINGLE GENE TESTS					
□ 188 Hermansky-Pudlak Syndrome: Puerto Rican Mutations ( <i>HPS1, HPS3</i> )					
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:					
DID YOU REMEMBER TO?					
☐ Label specimen tube appropriately with TWO identifiers					
☐ Get a signature for medical necessity and patient consent					

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.



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. 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. <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</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

## INTERNATIONAL SAMPLES

If I 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 residence.

## 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 deidentified 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.



First Name Last Name Date of Birth

## **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.

If I do not have health insurance, I agree to pay for the full cost of the genetic testing that was ordered by my healthcare provider and billed to me by GeneDx. I further understand and agree that, if I fail to make payment for genetic testing, in accordance with the payment policies of GeneDx, my account may be turned over to an external collection agency for non-payment. I agree to pay any associated collection costs, including attorney fees. By my signature on the GeneDx Test Requisition Form or at the bottom of this form, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider.

## MEDICARE

A completed Advance Beneficiary Notice (ABN) is required for Medicare patients, when applicable. Please visit our website, www.genedx.com/billing for more information.