MITOCHONDRIAL & METABOLIC TEST REQUISITION FORM

GeneDx

PATIENT INFORMATION

First Name	Last Name	
Genetic 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: _____	

SAMPLE INFORMATION

Date Sample Collected (mm/dd/yy) (required):	Medical Record #
○ Blood ○ Buccal Swab ○ Other (specify 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: _____	

PATIENT CONSENTS

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.

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 test development studies.

○ Check this box if you wish to opt out of being contacted for research studies.

○ 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).

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

OPTIONAL AND FOR COMMERCIAL INSURANCE ONLY:

By entering my preferred contact information below, I give my permission to GeneDx to send me an email and/or text with a link to access my personalized Digital Patient Letter. Data rates may apply.

Mobile Number*	Email*

*Contact information provided must be for the individual authorizing the genetic testing.

ACCOUNT INFORMATION

GeneDx Account Number		**Account Name**
Phone		Fax
Address		City
State	Zip Code	Country
Ordering Provider Name		Role/Title
NPI		Phone Number
Send Report Via ○ Fax ○ Email ○ Portal Fax #/Email: _____		
Additional Reporting Provider's Name		
Send Report Via ○ Fax ○ Email ○ Portal Fax #/Email: _____		
SEND ADDITIONAL REPORT COPIES TO:		
Provider Name		GeneDx Acct#
Fax #/Email:		

ICD-10 CODES *(Required)*

ICD-10 Codes	
Clinical Diagnosis	Age of Onset

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

PAYMENT OPTIONS *(Select One)*

○ **INSURANCE BILL** *(select all that applies)*

○ Commercial
○ Medicaid
○ Medicare
○ Tricare

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

Patient Status
○ Hospital outpatient ○ Hospital inpatient; Date of Discharge _____
○ Not a hospital patient

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)	GeneDx Benefit Investigation #

Secondary Insurance Type:

Insurance Carrier	Insurance ID #	Subscriber Name	Date of Birth

Relationship to Insured:
○ Self ○ Spouse ○ Child ○ Other: _____

○ **PATIENT BILL**

Amount Due: _____

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

CLINICAL INFORMATION

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

CLINICAL INFORMATION

Relevant clinical records are required at the time of sample submission to ensure the information is included in data analysis.

Pre/Perinatal History
- ○ Cystic hygroma
- ○ Decreased body weight
- ○ Diaphragmatic hernia
- ○ Growth delay
- ○ Increased body weight
- ○ Intrauterine growth retardation
- ○ Neural tube defect
- ○ Nonimmune hydrops fetalis
- ○ Oligohydramnios
- ○ Polyhydramnios
- ○ Prematurity GA: _____
- ○ Prolonged neonatal jaundice

Structural Brain Abnormalities
- ○ 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
- ○ Brain atrophy
- ○ Cerebellar atrophy
- ○ Cerebellar hypoplasia (Pontocerebellar hypoplasia)
- ○ CNS hypomyelination
- ○ Cortical dysplasia
- ○ Holoprosencephaly
- ○ Hydrocephalus
- ○ Leukodystrophy
- ○ Lissencephaly
- ○ Pachygyria
- ○ Polymicrogyria
- ○ Pontocerebellar atrophy
- ○ Ventriculomegaly

Developmental/Behavioral Findings
- ○ Abnormal aggressive, impulsive or violent behavior
- ○ Abnormal social behavior
- ○ Absent speech
- ○ Attention deficit hyperactivity disorder
- ○ Autistic behavior
- ○ Clumsiness
- ○ Cognitive impairment
- ○ Delayed fine motor development
- ○ Delayed gross motor development
- ○ Delayed speech & language development
- ○ Developmental regression
- ○ Dysarthria
- ○ Frequent falls
- ○ Gait disturbance
- ○ Global developmental delay
- ○ Incoordination
- ○ Intellectual disability
- ○ Memory impairment
- ○ Sleep disturbance
- ○ Specific learning disability
- ○ Speech articulation difficulties
- ○ Stereotypy

Neurological Findings
- ○ Abnormality of nervous system
- ○ Ataxia
- ○ Cerebral palsy
- ○ Cortical visual impairment
- ○ Dysarthria
- ○ Dysphasia
- ○ Dystonia
- ○ Encephalopathy
- ○ Epileptic encephalopathy
- ○ Generalized seizures
- ○ Headaches
- ○ Hyperreflexia
- ○ Infantile spasms
- ○ Limb hypertonia
- ○ Myoclonus
- ○ Parkinsonism
- ○ Peripheral neuropathy
- ○ Seizures
- ○ Sensory neuropathy
- ○ Spasticity
- ○ Stroke-like episode
- ○ Syncope
- ○ Tremors
- ○ Vertigo

Craniofacial/Dysmorphism
- ○ Abnormal facial shape (Dysmorphic features)
- ○ Abnormality of philtrum
- ○ Anteverted nares
- ○ Brachycephaly
- ○ Broad forehead
- ○ Bulbous nose
- ○ Cleft lip
- ○ Cleft palate
- ○ Coarse facial features
- ○ Craniosynostosis
- ○ Deeply set eye
- ○ Dental crowding
- ○ Depressed nasal bridge
- ○ Epicanthus
- ○ Facial asymmetry
- ○ Frontal bossing
- ○ High palate
- ○ Hypertelorism
- ○ Hypotelorism
- ○ Long face
- ○ Low set ears
- ○ Macrocephaly
- ○ Microcephaly
- ○ Micrognathia
- ○ Midface retrusion
- ○ Prominent nasal bridge
- ○ Retrognathia
- ○ Synophrys
- ○ Wide nasal bridge
- ○ Wide spaced teeth

Eye Defects/ Vision
- ○ Aniridia
- ○ Anophthalmia
- ○ Astigmatism
- ○ Cataracts
- ○ Coloboma

Eye Defects/ Vision *(continued)*
- ○ Corneal opacity
- ○ Ectopia lentis
- ○ Esotropia
- ○ Exotropia
- ○ External ophthalmoplegia
- ○ Microphthalmia
- ○ Myopia
- ○ Nystagmus
- ○ Optic atrophy
- ○ Optic neuropathy
- ○ Ptosis
- ○ Retinitis pigmentosa
- ○ Strabismus
- ○ Visual impairment

Hearing Impairment
- ○ Aminoglycoside-induced hearing loss
- ○ Conductive hearing impairment/bilateral
- ○ Hearing impairment
- ○ Sensorineural hearing impairment/bilateral

Cardiac Findings
- ○ Abnormal echocardiogram
- ○ Abnormal heart morphology
- ○ Abnormal heart valve morphology
- ○ Arrhythmia
- ○ Atrial septal defect
- ○ Cardiomegaly
- ○ Cardiomyopathy
- ○ Dilated cardiomyopathy
- ○ Hypertension
- ○ Hypertrophic cardiomyopathy
- ○ Palpitations
- ○ Tachycardia
- ○ Ventricular septal defect

Respiratory Findings
- ○ Apnea
- ○ Aspiration
- ○ Asthma
- ○ Hyperventilation
- ○ Hypoventilation
- ○ Recurrent upper respiratory infections
- ○ Respiratory distress
- ○ Respiratory insufficiency

Gastrointestinal Findings
- ○ Constipation
- ○ Diarrhea
- ○ Exocrine pancreatic insufficiency
- ○ Failure to thrive
- ○ Feeding difficulties
- ○ Gastroesophageal reflux
- ○ Gastrointestinal dysmotility
- ○ Gastroparesis
- ○ Hepatomegaly
- ○ Inflammatory bowel disease
- ○ Laryngomalacia
- ○ Nausea
- ○ Pancreatitis
- ○ Pyloric stenosis
- ○ Splenomegaly
- ○ Tracheoesophageal fistula
- ○ Vomiting

CLINICAL INFORMATION

GeneDx Account #	Account Name	
First Name	Last Name	Date of Birth

CLINICAL INFORMATION

Relevant clinical records are required at the time of sample submission to ensure the information is included in data analysis.

Musculoskeletal Findings

- ○ Abnormal connective tissue
- ○ Abnormal form of the vertebral bodies
- ○ Abnormality of joint mobility
- ○ Arthrogryposis
- ○ Bruising susceptibility
- ○ Craniosynostosis
- ○ Decreased muscle mass
- ○ Dolichocephaly
- ○ Dysostosis multiplex
- ○ Elevated serum creatine phosphokinase
- ○ Exercise intolerance
- ○ Fasciculations
- ○ Fatigue
- ○ Flexion contracture
- ○ Hemihypertrophy
- ○ Hypertonia
- ○ Hypotonia
- ○ Joint hypermobility
- ○ Muscle cramps
- ○ Muscle weakness
- ○ Myalgia
- ○ Myopathy
- ○ Pectus excavatum
- ○ Pes planus
- ○ Ptosis
- ○ Rhabdomyolysis
- ○ Scoliosis
- ○ Short stature
- ○ Skeletal dysplasia

Skin/Hair Findings

- ○ Alopecia
- ○ Angiokeratoma
- ○ Brittle hair
- ○ Café-au-lait macules
- ○ Coarse hair
- ○ Dry skin
- ○ Eczema
- ○ Hemangiomas
- ○ Hyperextensible skin
- ○ Hyperpigmentation of the skin
- ○ Hypertrichosis
- ○ Hypopigmentation of the skin
- ○ Ichthyosis
- ○ Skin rash
- ○ Sparse hair
- ○ Velvety skin (Soft skin)
- ○ Xanthomatosis

Genitourinary Findings

- ○ Ambiguous genitalia
- ○ Cryptorchidism
- ○ Glomerulosclerosis
- ○ Hydronephrosis
- ○ Hypospadias
- ○ Inguinal hernia
- ○ Polycystic kidney disease
- ○ Renal agenesis
- ○ Renal insufficiency
- ○ Renal tubular acidosis
- ○ Renal tubular dysfunction
- ○ Urinary incontinence

Metabolic Issues/Mito (Attached relevant lab reports/values)

- ○ Abnl Plasma AA result: _____
- ○ Abnl Urine OA result: _____
- ○ Abnormal activity of mitochondrial respiratory chain
- ○ Abnormal mitochondria in muscle tissue
- ○ Abnormal Newborn Screen result: _____
- ○ Abnormality of mitochondrial metabolism
- ○ Cytochrome C oxidase-negative muscle fibers
- ○ Decreased activity of mitochondrial ATP synthase complex
- ○ Decreased activity of mitochondrial respiratory complexes
- ○ Decreased activity of the pyruvate dehydrogenase complex
- ○ Depletion of mitochondrial DNA in liver
- ○ Depletion of mitochondrial DNA in muscle tissue
- ○ Elevated CPK: _____
- ○ Elevated hepatic transaminases
- ○ Hyperammonemia
- ○ Hypoammonemia
- ○ Hypoglycemia
- ○ Increased serum pyruvate
- ○ Lactic acidosis
- ○ Multiple mitochondrial DNA deletions
- ○ Subsarcolemmal accumulations of abnormally shaped mitochondria
- ○ Vitamin D deficiency

Endocrine Findings

- ○ Diabetes Insipidus
- ○ Diabetes Mellitus
- ○ Hyperthyroidism
- ○ Hypothyroidism

Vascular System

- ○ Stroke
- ○ Thromboembolism

Draw/attach pedigree and/or include additional clinical information:

Signature of Provider (required)	Date

MITOCHONDRIAL & METABOLIC TEST REQUISITION FORM

GeneDx

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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
○ Known Familial Copy Number Variant(s)
○ Confirmation of Variant Identified in Research Lab
○ Known mtDNA Variant(s) Testing

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)

○ J820 Mito Xpanded®, Family Member Testing

Mother	First Name	Last Name	DOB	○ Asymptomatic ○ Symptomatic ○ At GeneDx ○ Not Available ○ To be Sent Within 3 Weeks*	
Father	First Name	Last Name	DOB	○ Asymptomatic ○ Symptomatic ○ At GeneDx ○ Not Available ○ To be Sent Within 3 Weeks*	
Other	Relationship to Proband				
	First Name	Last Name	DOB	○ Asymptomatic ○ Symptomatic ○ At GeneDx ○ Not Available ○ To be Sent Within 3 Weeks*	

>> 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
1	○	○		
2	○	○		
3	○	○		

TESTING HISTORY: ○ Test Report Included (recommended)

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

MITOCHONDRIAL & METABOLIC TEST REQUISITION FORM

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MITOCHONDRIAL DISORDERS GENETIC TESTING

TEST CODE	TEST NAME	# OF GENES	GENE LIST/DESCRIPTION
○ J809 ○ 554	Mito Xpanded® Panel (~1800 genes)* Concurrent full sequence analysis & deletion testing of the mito genome (not a trio based test)		* "Xpanded® Family Member Testing" portion must be completed from Page 4. Relevant clinical records are required at the time of sample submission to ensure the information is included in data analysis. Gene list is updated every 6-12 months to include most newly discovered genes
○ 615	Combined Mito Genome Plus Mito Focused Nuclear Gene Panel	202	*AARS2, ABCB7, ACAD9, ACO2, AFG3L2, AGK, AIFM1, ALAS2, APOPT1, ATP5A1, ATP5E, ATP7B, ATPAF2, AUH, BCS1L, BOLA3, C12ORF65, C19orf12, CARS2, CLPB, COA5, COA6, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX6A1, COX6B1, COX8A, CYC1, DARS2, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX1L, FH, FLAD1, FOXRED1, GARS, GCDH, GFER, GFM1, GFM2, GLRX5, GTPBP3, GYG2, HARS2, HMGCL, HTRA2, IARS2, IBA57, ISCA2, ISCU, LAMP2, LARS, LARS2, LIAS, LIPT1, LRPPRC, LYRM4, LYRM7, MARS2, MFF, MFN2, MGME1, MICU1, MPC1, MPV17, MRPL12, MRPL3, MRPL44, MRPS16, MRPS22, MRPS7, MTFMT, MTO1, MTPAP, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NR2F1, NUBPL, OPA1, OPA3, OTC, PARS2, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PNPT1, POLG, POLG2, PRKAG2, PUS1, QARS, RARS, RARS2, RMND1, RNASEH1, RRM2B, SARS2, SCO1, SCO2, SDHA, SDHAF1, SERAC1, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SPAST, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TFAM, TIMM8A, TK2, TMEM126A, TMEM126B, TMEM70, TPK1, TRIT1, TRMT10C, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS2, WDR45, WFS1, YARS2* and Mitochondrial Genome
○ 554	Full sequence analysis and deletion testing of the mitochondrial genome	37	
○ 573	Mitochondrial Focused Nuclear Gene Panel	202	*AARS2, ABCB7, ACAD9, ACO2, AFG3L2, AGK, AIFM1, ALAS2, APOPT1, ATP5A1, ATP5E, ATP7B, ATPAF2, AUH, BCS1L, BOLA3, C12ORF65, C19orf12, CARS2, CLPB, COA5, COA6, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX6A1, COX6B1, COX8A, CYC1, DARS2, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX1L, FH, FLAD1, FOXRED1, GARS, GCDH, GFER, GFM1, GFM2, GLRX5, GTPBP3, GYG2, HARS2, HMGCL, HTRA2, IARS2, IBA57, ISCA2, ISCU, LAMP2, LARS, LARS2, LIAS, LIPT1, LRPPRC, LYRM4, LYRM7, MARS2, MFF, MFN2, MGME1, MICU1, MPC1, MPV17, MRPL12, MRPL3, MRPL44, MRPS16, MRPS22, MRPS7, MTFMT, MTO1, MTPAP, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NR2F1, NUBPL, OPA1, OPA3, OTC, PARS2, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PNPT1, POLG, POLG2, PRKAG2, PUS1, QARS, RARS, RARS2, RMND1, RNASEH1, RRM2B, SARS2, SCO1, SCO2, SDHA, SDHAF1, SERAC1, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SPAST, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TFAM, TIMM8A, TK2, TMEM126A, TMEM126B, TMEM70, TPK1, TRIT1, TRMT10C, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS2, WDR45, WFS1, YARS2*
○ 575	Mitochondrial Encephalopathy/Leigh Syndrome Nuclear Gene Panel	134	*AARS2, ACAD9, ACO2, AFG3L2, AIFM1, APOPT1, ATP5A1, ATP5E, ATPAF2, AUH, BCS1L, BOLA3, C12ORF65, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX8A, CYC1, DARS2, DGUOK, DLAT, DLD, DNM1L, EARS2, ECHS1, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FH, FOXRED1, GCDH, GFER, GFM1, GFM2, GTPBP3, GYG2, HMGCL, HTRA2, IARS2, IBA57, ISCA2, LARS2, LIAS, LIPT1, LRPPRC, LYRM7, MARS2, MFF, MFN2, MPC1, MPV17, MRPL44, MRPS22, MTFMT, MTPAP, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB11, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PNPT1, POLG, RARS2, RMND1, RRM2B, SCO1, SCO2, SDHA, SDHAF1, SERAC1, SLC19A3, SLC22A5, SLC25A46, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TK2, TMEM70, TPK1, TRMU, TSFM, TTC19, TUFM, TWNK, TYMP, UQCC2, UQCC3, UQCRQ, VARS2*
○ 576	Lactic Acidosis/Pyruvate Metabolism Nuclear Gene Panel	130	*ACAD9, AGK, AIFM1, ATP5E, ATPAF2, BCS1L, BOLA3, C12ORF65, CARS2, COQ2, COQ4, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX6B1, CYC1, DARS2, DGUOK, DLAT, DLD, DNM1L, EARS2, ECHS1, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FBXL4, FDX1L, FH, FOXRED1, GFER, GFM1, GTPBP3, GYG2, HMGCL, HTRA2, IBA57, ISCU, LARS, LARS2, LIAS, LIPT1, LRPPRC, LYRM4, LYRM7, MFF, MPC1, MPV17, MRPL12, MRPL44, MRPS16, MRPS22, MRPS7, MTFMT, MTO1, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA9, NDUFAF1, NDUFAF3, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NFS1, NFU1, PARS2, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PNPT1, POLG, POLG2, PUS1, RARS2, RMND1, RNASEH1, RRM2B, SARS2, SCO2, SDHAF1, SERAC1, SFXN4, SLC25A26, SLC25A3, SLC25A4, SUCLA2, SUCLG1, SURF1, TARS2, TAZ, TK2, TMEM70, TPK1, TRMT10C, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, YARS2*
○ 577	Progressive External Ophthalmoplegia (PEO)/ Optic Atrophy Nuclear Gene Panel	44	*ACO2, AUH, C12ORF65, CLPB, DGUOK, DNA2, DNAJC19, DNM1L, EARS2, FH, GYG2, ISCA2, MFF, MFN2, MGME1, MTFMT, MTO1, MTPAP, NARS2, NDUFAF3, NR2F1, OPA1, OPA3, PDHX, PDSS1, POLG, POLG2, RNASEH1, RRM2B, SLC19A2, SLC19A3, SLC25A4, SLC25A46, SPG7, SUCLA2, TACO1, TIMM8A, TK2, TMEM126A, TSFM, TWNK, TYMP, VARS2, WFS1*
○ 578	Methylglutaconic Aciduria Nuclear Gene Panel	14	*AGK, ATP5E, ATPAF2, AUH, CLPB, DNAJC19, HMGCL, HTRA2, OPA3, POLG, SERAC1, SUCLA2, TAZ, TMEM70*
○ 938	Congenital Sideroblastic Anemia Panel	8	*ABCB7, ALAS2, GLRX5, PUS1, SLC19A2, SLC25A38, TRNT1, YARS2,* Mitochondrial genome large deletion testing
○ 704	mtDNA Point Variants Plus Large Deletions Panel		
○ TH12	Leber Hereditary Optic Neuropathy (LHON) Panel		
○ TB60	Deletion analysis of mito genome		
○ 394	*POLG* gene sequencing and del/dup testing	1	
○ 582	*SDHA* gene sequencing	1	

MITOCHONDRIAL & METABOLIC TEST REQUISITION FORM

GeneDx

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METABOLIC DISORDERS GENETIC TESTING PANELS

TEST CODE	TEST NAME	# OF GENES	GENE LIST/DESCRIPTION
○ J976	Creatine Deficiency Syndromes Panel	3	*GAMT, GATM, SLC6A8*
○ J977	Congenital Disorders of Glycosylation Panel	108	*ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6AP1, ATP6V0A2, B3GALNT2, B3GALT6, B3GLCT, B3GAT3, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, CCDC115, CHST14, CHST3, CHST6, CHSY1, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EPG5, EXT1, EXT2, FKRP, FKTN, FUT8, G6PC3, GALNT3, GFAT1, GMPPA, GMPPB, GNE, GTDC2, ISPD, LARGE1, LFNG, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PAPSS2, PGAP1, PGAP2, PGAP3, PGM1, PGM3, PIGA, PIGL, PIGM, PIGN, PIGO, PIGT, PIGV, PIGW, PIGY, PMM2, POFUT1, POGLUT1, POMGNT1, POMK, POMT1, POMT2, RFT1, RPN2, RXYLT1, SEC23A, SEC23B, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A8, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, TMEM165, TMEM199, TRAPPC11, TRIP11, TUSC3, XYLT1*
○ J980	Disorders Associated with C4 Elevation	6	*ACAD8, ACADS, ETFA, ETFB, ETFDH, ETHE1*
○ J995	Disorders of Hyperphenylalaninemia and Biopterin Metabolism Panel	7	*DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR, SPR*
○ T382	Fatty Acid Oxidation Disorders Panel	15	*ACADM, ACADS, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, GLUD1, HADHA, HADHB, HMGCL, HMGCS2, SLC22A5, SLC25A20*
○ T010	Hyperammonemia, Urea Cycle and Transporter Defects Panel	48	*ACADM, ACADVL, ARG1, ASL, ASS1, BCKDHA, BCKDHB, CA5A, CPS1, CPT1A, CPT2, DBT, DLD, ETFA, ETFB, ETFDH, GLUD1, HADHA, HADHB, HCFC1, HLCS, HMGCL, HMGCS2, IVD, MCCC1, MCCC2, MMAA, MMAB, MMACHC, MMADHC, MUT, NAGS, OAT, OTC, PC, PCCA, PCCB, PDHA1, PIGA, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, SUCLA2, SUCLG1, TMEM70*
○ T013	Lysosomal Disorders Panel	57	*ABHD5, ADAMTSL2, AGA, ARSA, ARSB, ASAH1, ATP6AP1, CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSF, DNAJC5, FUCA1 , GAA, GALC, GALNS, GBA, GLA, GLB1, GNE, GNPTAB, GNPTG, GNS, GPC3, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, LAMP2, LIPA, LYST, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PNPLA2, PPT1, PSAP, SCARB2, SGSH, SLC17A5, SMPD1, SUMF1, TPP1, VPS33A*
○ J979	Lysosomal and Peroxisomal Disorders Panel, Combined	82	*ABCD1, ABHD5, ACOX1, ADAMTSL2, AGA, AGPS, AMACR, ARSA, ARSB, ASAH1, ATP6AP1, CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSF, DNAJC5, DNM1L, FAR1, FUCA1 , GAA, GALC, GALNS, GBA, GLA, GLB1, GNE, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GUSB, HEXA, HEXB, HGSNAT, HSD17B4, HYAL1, IDS, IDUA, LAMP2, LIPA, LYST, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PNPLA2, PPT1, PSAP, SCARB2, SCP2, SGSH, SLC17A5 , SMPD1, SUMF1, TPP1, TRIM37, VPS33A*
○ 488	Maple Syrup Urine Disease (MSUD) Panel	3	*BCKDHA, BCKDHB, DBT*
○ T012	Metabolic Myopathy Panel	30	*ACAD9, ACADM, ACADVL, AGL, ALDOA, CPT2, ETFA, ETFB, ETFDH, FKRP, GAA, GYG1, GYS1, HADHA, HADHB, ISCU, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PYGM, RYR1, SLC22A5, SLC25A20, SUCLA2, TANGO2, TK2*
○ T011	Methylmalonic Acidemia, Disorders of Cobalamin Metabolism and Related Disorders Panel	19	*ABCD4, ACSF3, AMN, CD320, CUBN, HCFC1, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC (C2ORF25) , MTR, MTRR, MUT, SUCLA2, SUCLG1, TCN2*
○ 610	Mucopolysaccharidosis III (MPSIII)/ Sanfilippo Syndrome Types A, B, C and D) Panel	4	*GNS, HGSNAT, NAGLU, SGSH*
○ 581	Niemann-Pick Disease, Type C Panel	2	*NPC1, NPC2*
○ J978	Peroxisomal Disorders Panel	25	*ABCD1, ACOX1, AGPS, AMACR, DNM1L, FAR1, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, SCP2, TRIM37*
○ TG90	Primary Hyperoxaluria Panel	3	*AGXT, GRHPR, HOGA1*
○ J981	Riboflavin Transporter Deficiency and Related Disorders	9	*ACAD9, ETFA, ETFB, ETFDH, FLAD1, SLC25A32, SLC52A1, SLC52A2, SLC52A3*
○ TG91	Tyrosinemia Panel	3	*FAH, HPD, TAT*

MITOCHONDRIAL & METABOLIC TEST REQUISITION FORM

GeneDx

GeneDx Account #	Account Name	
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INBORN ERRORS OF METABOLISM SINGLE GENE TESTS

(The following tests include sequencing and deletion/duplication testing unless otherwise noted)

TEST CODE	TEST NAME	TEST CODE	TEST NAME
○ 463	2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency *(HSD17B10)*	Glutaric aciduria II / Multiple acyl-CoA dehydrogenase deficiency (MADD)	
○ 508	3-Hydroxyacyl-CoA dehydrogenase deficiency *(HADH)*	○ 280	*ETFDH*
3-Methylcrotonyl CoA carboxylase deficiency		○ 279	*ETFB*
○ 2881	*MCCC2*	○ 278	*ETFA*
○ 2882	*MCCC1*	○ 438	Glycerol kinase deficiency *(GK)*
○ 501	3-Methylglutaconic aciduria type I *(AUH)*	○ TH08	Glycogen storage disease II (Pompe disease) *(GAA)*
○ 380	6-pyruvoyl-tetrahydropterin synthase deficiency *(PTS)*	○ 649	Glycogen storage disease type V *(GSD V) (PYGM)*
○ 354	ß-ketothiolase deficiency *(ACAT1* sequencing only)	○ 657	GM1-gangliosidosis *(GLB1)*
○ 2631	Acid sphingomyelinase deficiency *(SMPD1)*	○ 527	GTP cyclohydrolase I deficiency *(GCH1)*
○ J975	Adrenoleukodystrophy, X-linked *(ABCD1)*	○ 3211	HMG CoA lyase deficiency *(HMGCL)*
○ 465	Arginase deficiency *(ARG1)*	○ 320	Holocarboxylase synthetase deficiency *(HLCS)*
○ 426	Argininosuccinic aciduria *(ASL)*	○ 331	Homocystinuria *(CBS)*
○ 658	Aspartylglucosaminuria *(AGA)*	○ T387	Hunter syndrome (mucopolysaccharidosis type II) *(IDS* sequencing, del/dup, recombination analysis)
○ 294	Biotinidase deficiency *(BTD)*	○ T386	Hurler syndrome/ mucopolysaccharidosis type I *(IDUA)*
○ 564	Canavan disease *(ASPA)*	○ 351	Isobutyryl CoA dehydrogenase deficiency *(ACAD8)*
○ 429	Carnitine-acylcarnitine translocase deficiency *(SLC25A20)*	○ 3191	Isovaleric acidemia *(IVD)*
○ 425	Carnitine palmitoyltransferase IA deficiency *(CPT1A)*	○ 507	Krabbe disease *(GALC)*
○ 334	Carnitine palmitoyltransferase deficiency type II *(CPT2)*	LCHAD/trifunctional protein deficiency *(HADHA/HADHA* and *HADHB)*	
○ 500	Citrin deficiency *(SLC25A13)*	○ 2712	*HADHA*
○ 382	Classic citrullinemia *(ASS1)*	○ 272	*HADHB*
○ 274	Cobalamin C deficiency *(MMACHC)*	○ TA73	Lowe syndrome *(OCRL)*
○ 659	Combined malonic and methylmalonic aciduria *(ACSF3)*	○ 655	Lysosomal acid lipase deficiency *(LIPA)*
○ 490	Dihydrolipoamide dehydrogenase deficiency *(DLD)*	○ 404	Malonyl-CoA decarboxylase deficiency *(MLYCD)*
○ 381	Dihydropteridine reductase *(DHPR)* deficiency *(QDPR)*	Maple Syrup Urine Disease (MSUD)	
○ 558	Ethylmalonic encephalopathy *(ETHE1)*	○ 4881	*BCKDHA*
○ 2321	Fabry disease *(GLA)*	○ 4882	*BCKDHB*
○ 605	Free sialic storage disorders *(SLC17A5)*	○ 4883	*DBT*
○ 661	Fucosidosis *(FUCA1)*	○ 488	*BCKDHA/BCKDHB/DBT* All NOW
○ 713	Fumarate hydratase deficiency *(FH)*	○ 565	Maroteaux-Lamy syndrome/mucopolysaccharidosis VI *(ARSB)*
○ 499	Galactokinase deficiency *(GALK1)*	○ 2682	Medium chain acyl-CoA dehydrogenase (MCAD) deficiency *(ACADM)*
○ 349E	Galactosemia / Galactosyltransferase deficiency *(GALT)*	○ 649	McArdle disease *(PYGM)*
○ TG94	Gaucher disease (*GBA* sequencing only)	○ 563	Metachromatic leukodystrophy *(ARSA)*
○ 399	Glutaric aciduria type I *(GCDH)*	○ 473	Methionine adenosyltransferase I/III deficiency ***(MAT1A)***

MITOCHONDRIAL & METABOLIC TEST REQUISITION FORM

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INBORN ERRORS OF METABOLISM SINGLE GENE TESTS (CONTINUED)

(The following tests include sequencing and deletion/duplication testing unless otherwise noted)

TEST CODE	TEST NAME	TEST CODE	TEST NAME
Methylmalonic acidemia		Propionic acidemia	
○ 2752	*MUT*	○ 2901	*PCCB*
○ 276	*MMAA*	○ 2902	*PCCA*
○ 277	*MMAB*	○ 365	Primary/systemic carnitine deficiency *(SLC22A5)*
○ 657	Morquio B disease *(GLB1)*	○ 528	PSAP-related disorders *(PSAP)*
○ 608	Morquio syndrome A/ Mucopolysaccharidosis IVA *(GALNS)*	○ 540	Pyruvate carboxylase deficiency *(PC)*
○ 648	Mucolipidosis I *(NEU1)*	○ 461	Pyruvate dehydrogenase E1-alpha deficiency *(PDHA1)*
○ 2432	Mucolipidosis type IV *(MCOLN1)*	○ 462	Pyruvate dehydrogenase E1-beta deficiency *(PDHB)*
○ T386	Mucopolysaccharidosis type I (Hurler syndrome) *(IDUA)*	○ 605	Salla disease *(SLC17A5)*
○ T387	Mucopolysaccharidosis type II (Hunter syndrome) (*IDS* sequencing, del/dup recombination analysis)	○ 515	Sandhoff disease *(HEXB)*
Mucopolysaccharidosis III (MPSIII)/Sanfilippo syndrome (Types A, B, C and D)		Sanfilippo syndrome/ Mucopolysaccharidosis III (MPS IIIA, IIIB, IIIC, and IIID)	
○ 591	MPSIII A *(SGSH)*	○ 591	Sanfilippo A *(SGSH)*
○ 592	MPSIII B *(NAGLU)*	○ 592	Sanfilippo B *(NAGLU)*
○ 593	MPSIII C *(HGSNAT)*	○ 593	Sanfilippo C *(HGSNAT)*
○ 609	MPSIII D *(GNS)*	○ 609	Sanfilippo D *(GNS)*
○ 610	*SGSH/NAGLU/HGSNAT/GNS* All NOW	○ 610	*SGSH/ NAGLU/ HGSNAT/ GNS* All NOW
○ 608	Mucopolysaccharidosis IVA/Morquio syndrome A *(GALNS)*	○ 528	Saposin deficiency (combined, SapA, SapB, and SapC) *(PSAP)*
○ 565	Mucopolysaccharidosis VI (Maroteaux-Lamy syndrome) *(ARSB)*	○ 383	Short/branched chain acyl-CoA dehydrogenase deficiency *(ACADSB)*
○ 657	Mucopolysaccharidosis type IVB *(GLB1)*	○ 269	Short-chain acyl-CoA dehydrogenase *(SCAD)* deficiency *(ACADS)*
○ 611	Multiple sulfatase deficiency *(SUMF1)*	○ 648	Sialidosis *(NEU1)*
○ 478	N-acetylglutamate synthase deficiency *(NAGS)*	○ 2502	Smith-Lemli-Opitz syndrome *(DHCR7)*
○ 607	Neuronal ceroid-lipofuscinosis 2 *(TPP1)*	○ 519	Tay-Sachs disease *(HEXA)*
Niemann-Pick disease *(NPD)*, type C		Tyrosinemia	
○ 246	NPD type C1 *(NPC1)*	○ 3661	Type I *(FAH)*
○ 247	NPD type C2 *(NPC2)*	○ 494	Type II *(TAT)*
○ 581	*NPC1* and *NPC2* both NOW	○ 495	Type III *(HPD)*
○ 313	Ornithine transcarbamylase (OTC) deficiency *(OTC)*	○ 270	Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency *(ACADVL)*
○ 273	Phenylalanine hydroxylase *(PAH)*	○ TG92	Wilson disease *(ATP7B)*
○ TH08	Pompe disease/glycogen storage disease type II *(GAA)*	○ J975	X-linked adrenoleukodystrophy *(ABCD1)*

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.

INFORMED CONSENT

GeneDx Account #	Account Name	
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: 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 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.

INFORMED CONSENT

GeneDx Account #	Account Name	
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, or 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. Please visit our website, *www.genedx.com/billing* for more information.

DIGITAL PATIENT LETTER CONSENT

- Applicable Only for Commercial Insurance
- Estimate is provided by your health insurance company and therefore NO estimate will be sent for any orders placed with federal or state-funded insurance plans (e.g. Medicare, Medicaid, Tricare, etc.), institutional bill, or patient bill (self-pay).

To provide you with the estimated out-of-pocket expenses related to your test, GeneDx will send you an email and/or text with the link to access your personalized Digital Patient Letter. In order to send this information, we need your consent and agreement to the following items:

1. GeneDx can use your email address or mobile phone number solely for the purpose of GeneDx sending your estimated financial obligation. Text message data rates may apply. GeneDx is not responsible for undelivered messages due to incorrect or illegible contact information.
2. GeneDx will send you an email and/or text message containing a link to view your personalized Patient Letter that includes the test out-of-pocket estimate. The link is time-sensitive and will only be available for 72 hours from the time the message is sent. In order to view the estimate, you must click the link in the message.
3. If you take no action, GeneDx will assume that you agree to move ahead with testing and will bill your health insurance. You can approve testing with insurance, switch to self-pay, or cancel the test via the link within the given 72-hour window. In turn, if GeneDx receives your sample(s) and the billing method hasn't been changed, or the test hasn't been cancelled, we will move ahead with testing as ordered, and you will be responsible for any out-of-pocket costs for the completion of the test(s).