MITOCHONDRIAL & METABOLIC TEST REQUISITION FORM

GeneDx an OPKO Health Company

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 CONSENT FOR GENETIC TESTING, FINANCIAL AGREEMENT AND GUARANTEE:

By signing this form, I acknowledge as the patient or relative being tested that I have read the GeneDx Informed Consent document available from my healthcare provider or at genedx.com/forms, and I authorize GeneDx to perform genetic testing as ordered. I understand that, for tests that evaluate data from multiple family members concurrently, 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 my signature below, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider. 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 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 and agree to be financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. I am aware that my insurance provider may send 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. 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 and I agree to pay any associated collection costs, including attorney fees.

More information, including the GeneDx Notice of Privacy Policies, is available on GeneDx's website: www.genedx.com

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

○ 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 to be used as a de-identified sample for test development and improvement, internal validation, quality assurance, and training purposes. Otherwise, New York law requires GeneDx to destroy my sample after 60 days and it cannot be used for the studies listed above.

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

○ Check this box if you do not want to receive ACMG secondary findings (Full Exome Sequencing and Genome Sequencing Tests ONLY; not for Xpanded® or Slice tests).

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

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 NECESSISTY

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

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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 (heteroplasmy detection range 25%-100%)
○ Known mtDNA Variant(s) Testing by NGS (heteroplasmy detection range 1.5%-100%) - Test Code 453
○ Known mtDNA Variant(s) Testing by NGS - URINE (heteroplasmy detection range 5-100%) - Test Code T822

Proband Name	Relationship to Proband	Proband GeneDx Accession #

Non-GeneDx Test:
○ Family member test report included (recommended if previous test was performed at another lab)
○ Positive control included/will be sent - **Positive control is recommended if previous test was performed at another lab.**
○ Positive control not available (caveat language will be included on a negative report)

Variant Information (please fill out the below information if family member report is not included) Number of Variants: _____

Gene	Coding DNA (c./m.)	Amino Acid (p.)	Transcript (NM#)
Gene	Coding DNA (c./m.)	Amino Acid (p.)	Transcript (NM#)

Copy Number Variants (CNV(s) require coordinates and genome build or transcript # and exon #) Number of Variants: _____

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

TESTING OPTIONS

CUSTOM DEL/DUP TESTING

○ 906 Deletion/Duplication Analysis of ONE Nuclear Gene ○ 703 Deletion/Duplication Analysis of 2-20 Nuclear Genes

Write-in Desired Gene(s) to be Tested: _____

FAMILY MEMBER TESTING (NO SEPARATE REPORT)

○ 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 GENETIC TESTING

MITOCHONDRIAL DISORDERS

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 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	1	
○ 557	*PUS1* Gene sequencing	1	
○ 582	*SDHA* Gene sequencing	1	

MITOCHONDRIAL & METABOLIC TEST REQUISITION FORM

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MITOCHONDRIAL GENETIC TESTING

METABOLIC GENETIC TESTING

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) Sequencing 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) Sequencing Panel	4	*GNS, HGSNAT, NAGLU, SGSH & del/dup of GNS*
○ 581	Niemann-Pick Disease, Type C Sequencing 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

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INBORN ERRORS OF METABOLISM SINGLE GENE TESTS

TEST CODE	TEST NAME	TEST CODE	TEST NAME
2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (HSD17B10)		Glutaric aciduria II / Multiple acyl-CoA dehydrogenase deficiency (MADD)	
○ 463	HSD17B10 sequencing	○ 280	*ETFDH*
○ 906	HSD17B10 del/dup testing, females	○ 279	*ETFB*
○ 508	3-Hydroxyacyl-CoA dehydrogenase deficiency *(HADH)*	○ 278	*ETFA*
3-Methylcrotonyl CoA carboxylase deficiency		Glycerol kinase Deficiency	
○ 2881	*MCCC2*	○ 438	*GK* sequencing
○ 2882	*MCCC1*	○ 906	*GK* del/dup testing, females
○ 501	3-Methylglutaconic aciduria type I *(AUH)*	○ TH08	Glycogen storage disease II (Pompe disease) (*GAA* sequencing and del/dup testing)
○ 380	6-pyruvoyl-tetrahydropterin synthase deficiency *(PTS)*	○ 649	Glycogen storage disease type V *(GSD V) (PYGM)*
○ 354	ß-ketothiolase deficiency *(ACAT1)*	○ 657	GM1-gangliosidosis *(GLB1)*
○ 2631	Acid sphingomyelinase deficiency *(SMPD1)*	○ 527	GTP cyclohydrolase I deficiency (*GCH1* sequencing and del/dup)
○ J975	Adrenoleukodystrophy, X-linked (*ABCD1* sequencing and del/dup testing)	○ 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* sequencing and del/dup testing)
○ 564	Canavan disease (*ASPA* sequencing and del/dup testing)	○ 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* sequencing and del/dup testing)
○ 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* sequencing and del/dup testing)
○ 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* sequencing and del/dup testing)	○ 4881	*BCKDHA*
○ 2321	Fabry disease (*GLA* sequencing and del/dup testing)	○ 4882	*BCKDHB*
○ 605	Free sialic storage disorders (*SLC17A5* sequencing and del/dup testing)	○ 4883	*DBT*
○ 661	Fucosidosis *(FUCA1)*	○ 488	*BCKDHA/BCKDHB/DBT* All NOW
○ 713	Fumarate hydratase deficiency (*FH* sequencing and del/dup testing)	○ 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* sequencing and del/dup testing)	○ 649	McArdle disease *(PYGM)*
○ TG94	Gaucher disease *(GBA)*	○ 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)*

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)*	Pyruvate Dehydrogenase E1-Alpha Deficiency	
Mucolipidosis type IV		○ 461	*PDHA1* sequencing
○ 2432	*MCOLN1* sequencing	○ 906	*PDHA1* del/dup testing, females
○ 906	*MCOLN1* del/dup testing, including common 6.45kb deletion	○ 462	Pyruvate Dehydrogenase E1-Beta Deficiency *(PDHB)*
○ T386	Mucopolysaccharidosis type I (Hurler syndrome) (*IDUA* sequencing and del/dup testing)	○ 605	Salla disease (*SLC17A5* sequencing and del/dup testing)
○ T387	Mucopolysaccharidosis type II (Hunter syndrome) (*IDS* sequencing, del/dup recombination analysis)	○ 515	Sandhoff disease (*HEXB* sequencing and del/dup testing)
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* sequencing)	○ 591	Sanfilippo A (*SGSH* sequencing)
○ 592	MPSIII B (*NAGLU* sequencing)	○ 592	Sanfilippo B (*NAGLU* sequencing)
○ 593	MPSIII C (*HGSNAT* sequencing)	○ 593	Sanfilippo C (*HGSNAT* sequencing)
○ 609	MPSIII D (*GNS* sequencing and del/dup testing)	○ 609	Sanfilippo D (*GNS* sequencing and del/dup testing)
○ 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)*
Ornithine transcarbamylase deficiency (OTC)		○ 270	Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency *(ACADVL)*
○ 313	*OTC* sequencing, males	○ TG92	Wilson disease (*ATP7B* sequencing and del/dup testing)
○ 313E	*OTC* sequencing and del/dup testing, females	○ J975	X-linked adrenoleukodystrophy (*ABCD1* sequencing and del/dup testing)
○ 273	Phenylalanine hydroxylase *(PAH)*		
○ TH08	Pompe disease/glycogen storage disease type II (*GAA* sequencing and del/dup testing)		

INFORMED CONSENT

GeneDx Account #	Account Name	
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General Information About Genetic Testing

What is genetic testing?

DNA provides instructions for our body's growth and development. Genes are distinct sequences of DNA, and are arranged on chromosomes. The DNA in a gene contains instructions for making proteins, which determine things like growth and metabolism as well as traits like eye color and blood type. Genetic disorders are caused by certain changes in DNA affecting the structure or number of chromosomes. Genetic testing is a laboratory test that tries to identify these changes in chromosomes or the DNA. Genetic testing can be a diagnostic test, which is used to identify or rule out a specific genetic condition. Genetic screening tests are used to assess the chance for a person to develop or have a child with a genetic condition. Genetic screening tests are not typically diagnostic and results may require additional testing.

The purpose of this test is to see if I, or my child, may have a genetic variant or chromosome rearrangement causing a genetic disorder or to determine the chance that I, or my child, will develop or pass on a genetic disorder in the future. 'My child' can also mean my unborn child, for the purposes of this consent.

If I/my child already know the specific gene variant(s) or chromosome rearrangement that causes the genetic disorder in my family, I will inform the laboratory of this information.

What could I learn from this genetic test?

The following describes the possible results from the test:

1) Positive: A positive result indicates that a genetic variant has been identified that explains the cause of my/my child's genetic disorder or indicates that I/my child am at increased risk to develop the disorder in the future. It is possible to test positive for more than one genetic variant.

2) Negative: A negative result indicates that no disease-causing genetic variant was identified by the test performed. It does not guarantee that I/my child will be healthy or free from genetic disorders or medical conditions. If I/my child test negative for a variant known to cause the genetic disorder in other members of my/my child's family, this result rules out a diagnosis of the same genetic disorder in me/my child due to this specific change.

3) Inconclusive/Variant of Uncertain Significance (VUS): A finding of a variant of uncertain significance indicates that a genetic change was detected, but it is currently unknown whether that change is associated with a genetic disorder either now or in the future. A variant of uncertain significance is not the same as a positive result and does not clarify whether I/my child is at increased risk to develop a genetic disorder. The change could be a normal genetic variant or it could be disease-causing. Further analysis may be recommended, including testing parents and other family members. Detailed medical records or information from other family members also may be needed to help clarify results.

4) Unexpected results: In rare instances, this test may reveal an important genetic change that is not directly related to the reason for ordering this test. For example, this test may tell me about the risk for another genetic condition I/my child is not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. This information may be disclosed to the ordering health care provider if it likely impacts medical care.

Result interpretation is based on currently available information in the medical literature, research and scientific databases. Because the literature, medical and scientific knowledge are constantly changing, new information that becomes available in the future may replace or add to the information GeneDx used to interpret my/my child's results. Providers can contact GeneDx at any time to discuss the classification of an identified variant. In addition, I or my/my child's health care providers may monitor publicly available resources used by the medical community, such as ClinVar (www.clinvar.com), to find current information about the clinical interpretation of my/my child's variant(s).

For tests that evaluate data from multiple family members, my spouse, or partner concurrently, results may be included in a single comprehensive report.

What 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 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/my child's test results. The patient report may include clinical and genetic information about a relative when it is relevant to the interpretation of the results. Relatives do not receive an independent analysis of data nor a separate report.

What are the risks and limitations of this genetic test?

- Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer. In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology.
- Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in my/my child's family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results. In some cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. This includes non-paternity (the stated father of an individual is not the biological father) and consanguinity (the parents of an individual are related by blood). It may be necessary to report these findings to the health care provider who ordered the test.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to: mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, or the presence of change(s) in such a small percentage of cells that the change(s) may not be detectable by the test (mosaicism).
- This test does not have the ability to detect all of the long-term medical risks that I/my child might experience. The result of this test does not guarantee my health or the health of my child/fetus. Other diagnostic tests may still need to be done, especially when only a genetic screening test has been performed previously.
- Occasionally, an additional sample may be needed if the initial specimen is not adequate.

Patient Confidentiality and Genetic Counseling

It is recommended that I receive genetic counseling before and after having this genetic test. I can find a genetic counselor in my area here: www.nsgc.org. Further testing or additional consultations with a health care provider may be necessary.

To maintain confidentiality, the test results will only be released to the referring health care provider, to the ordering laboratory, to me, to other health care providers involved in my/my child's diagnosis and treatment, or to others as entitled by law. The United States Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, I understand that I can visit www.genome.gov/10002077.

International Specimens

If I/my child reside outside the United States, I attest that by providing a sample for testing, I am not knowingly violating any export ban or other legal restriction in the country of my/my child's residence.

Additional information about the specific test being ordered is available from my health care provider or I can go to the GeneDx website, www.genedx.com.This information includes the complete gene lists, the specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, the limitations of genetic testing, as well as information about how specimens and information are stored and used.

Specimen Retention

After testing is complete, the de-identified submitted specimen may be used for test development and improvement, internal validation, quality assurance, and training purposes. DNA specimens are not returned to individuals or to referring heath care providers unless specific prior arrangements have been made.

I understand that samples from residents of New York State will not be included in the de-identified research studies described in this authorization and will not be retained for more than 60 days after test completion, unless specifically authorized by my selection. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language. No tests other than those authorized shall be performed on the biological sample.

Database Participation

De-identified heath history and genetic information can help health care providers and scientists understand how genes affect human health. Though I/my child may not personally benefit, sharing this information helps health care providers to provide better care for their patients and researchers to make discoveries. GeneDx shares this type of information with health care providers, scientists and health care databases. No personal identifying information will be shared, as it will be replaced with a unique code.

Even though only a code is used for the reporting to the database, there is a risk that I/my child could be identified based on the genetic and health information that is shared. GeneDx believes that this is unlikely, though the risk is greater if I have already shared my/my child's genetic or health information with public resources, such as genealogy websites.

Recontact for Research Participation

Separate from the above, GeneDx may collaborate with scientists, researchers and drug developers to advance knowledge of genetic diseases and to develop new treatments. If there are opportunities to participate in research relevant to the disorder in my/my child's family, and if I have consented for recontact, GeneDx may allow my healthcare provider to be recontacted for research purposes, such as the development of new testing, drug development, or other treatment modalities. In some situations, such as if my health care provider is not available, I may be contacted directly.

Any research that results in medical advances, including new products, tests or discoveries, may have potential commercial value and may be developed and owned by GeneDx or the collaborating researchers. If any individuals or corporations benefit financially from these studies, no compensation will be provided to my/my child or my/my child's heirs.