RARE DISORDERS 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.

Signature of Patient/Guardian (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 (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Additional clinical findings:
_____

_____

_____

_____
Signature of Provider (required) Date

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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	○ Targeted Mosaic Variant Testing **(Insurance Billing NOT Accepted; Patient Bill or Institutional Bill MUST be selected on page 1)**
○ Known Familial Copy Number variant(s)	○ Known mtDNA Variant(s) Testing
○ Confirmation of Variant Identified in Research Lab	

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.): _____ Amino Acid (p.): _____ Transcript (NM#): _____

Gene: _____ Coding DNA (c.): _____ 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:_____

WRITE-IN TEST SELECTION

○ Test Code:_____	Test Name: _____

HISTORY

FAMILY HISTORY

○ No Known Family History ○ Pedigree Attached ○ Adopted

RELATIONSHIP TO INDIVIDUAL TO BE TESTED	MATERNAL	PATERNAL	RELEVANT HISTORY	AGE AT DX
	○	○		
	○	○		
	○	○		

TESTING HISTORY

○ Test report included (recommended)

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

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COMMON VARIANTS			
Test Code	**Test Name**	**Gene**	**Variant List**
○ TF73	Alpha-1 Antitrypsin Deficiency	SERPINA1	c.1096G>A, p.E366K (Z allele) and c.863A>T, p.E288V (S allele)
○ TF72	Alzheimer Disease Risk	APOE	c.[388T>C;526=], p.[C130R;R176=] (ε4 allele)
○ TF74	Chronic Kidney Disease Risk	APOL1	c.1024A>G, p. S342G (G1 allele) and c.1164_1169delTTATAA, p.N388_Y389del (G2 allele)
○ TF76	Prothrombin (Factor II) Thrombophilia	F2	c.*97G>A
○ TF78	Factor V Leiden Thrombophilia	F5	c.1601G>A, p.R534Q
○ TF77	Hemochromatosis	HFE	c.187C>G, p.H63D and c.845G>A, p.C282Y
○ TF75	Hereditary Lung Cancer Risk	EGFR	c.2369C>T, p.T790M

RARE DISORDERS MULTI-GENE PANELS			
TEST CODE	**TEST NAME**	**# GENES**	**GENE LIST**
DERMATOLOGIC DISORDERS			
○ 708	Congenital ichthyosis XomeDxSlice	49	*ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, ARSE, CASP14, CDSN, CERS3, CHST8, CLDN1, CSTA, CYP4F22, EBP, ELOVL4, FLG, FLG2, GJB2, GJB3, GJB4, GJB6, KDSR, KRT1, KRT10, KRT2, KRT9, LIPN, LOR, MBTPS2, NIPAL4, NSDHL, PEX7, PHGDH, PHYH, PNPLA1, POMP, PSAT1, SDR9C7, SERPINB8, SLC27A4, SNAP29, SPINK5, ST14, STS, TGM1, TGM5, VPS33B, ZMPSTE24*
○ 707	Epidermolysis bullosa (EB) XomeDxSlice	28	*CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSG1, DSP, DST, EXPH5, FERMT1, FLG2, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, SERPINB8, TGM5*
○ B399	Melanoma panel	9	*BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN, RB1, TP53*
DYSMORPHOLOGY AND MULTIPLE CONGENITAL ANOMALIES			
○ TA46	Adams-Oliver syndrome panel	6	*ARHGAP, DLL4, DOCK6, EOGT, NOTCH1, RBPJ*
○ TA44	Baraitser-Winter syndrome panel	2	*ACTB, ACTG1*
○ T993	Coffin-Siris syndrome panel	8	*ARID1A, ARID1B, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SOX11*
○ 584	Cornelia de Lange syndrome panel	7	*ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC1A, SMC3*
○ TB04	Kabuki syndrome panel	2	*KMT2D, KDM6A*
○ 961	Neurofibromatosis type 1 and 2 panel	4	*NF1, NF2, SMARCB1, SPRED1*
○ 962	Neurofibromatosis type 1 panel	2	*NF1, SPRED1*
○ 963	Neurofibromatosis type 2 panel	3	*LZTR1, NF2, SMARCB1*
○ TA06	Noonan and comprehensive RASopathies panel	25	*A2ML1, ACTB, ACTG1, BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1*
○ TA39	Robinow syndrome panel	4	*DVL1, DVL3, ROR2, WNT5A*
○ TA38	Treacher Collins syndrome panel	6	*DHODH, EFTUD2, POLR1C, POLR1D, SF3B4, TCOF1*
ENDOCRINE DISORDERS			
○ 676	Hypogonadotropic hypogonadism panel	33	*ANOS1, CHD7, CYP19A1, DUSP6, ESR1, FEZF1, FGF17, FGF8, FGFR1, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, NR0B1, NR5A1, NSMF, POLR3B, PROK2, PROKR2, PROP1, SEMA3A, SEMA3E, SOX10, SPRY4, TAC3, TACR3, WDR11*
○ 674	Maturity-onset diabetes of the young (MODY) panel	16	*ABCC8, APPL1, BLK, CEL, GCK, GLUD1, HADH, HNF1B, HNF4A, HNF1A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1 (IPF1)*
HEMATOLOGIC DISORDERS			
○ 938	Congenital sideroblastic anemia panel (plus mitochondrial genome large deletion testing)	8	*ABCB7, ALAS2, GLRX5, PUS1, SLC19A2, SLC25A38, TRNT1, YARS2*
○ J450	Diamond-Blackfan anemia panel	13	*GATA1, RPL11, RPL15, RPL26, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7*
○ TB47	Dyskeratosis congenita panel	12	*ACD, CTC1, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, USB1, WRAP53*

All sequencing tests include del/dup analysis unless indicated by a ^ or otherwise noted

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RARE DISORDERS MULTI-GENE PANELS

TEST CODE	TEST NAME	# GENES	GENE LIST
IMMUNOLOGIC DISORDERS			
○ T990	Autoimmune lymphoproliferative syndrome (ALPS) panel	4	*FAS, CASP10, CASP8, FASL*
○ 603	B- SCID sub-panel	9	*ADA, AK2, DCLRE1C (ARTEMIS), LIG4, NHEJ1, PRKDC, RAC2, RAG1, RAG2*
○ 602	B+ SCID sub-panel	17	*TM, CD3D, CD3E, CD3Z, CORO1A, DOCK8, FOXN1, IL2RG, IL7R, JAK3, ORAI1, PNP, PTPRC, RMRP, STIM1, TBX1, ZAP70*
○ T989	Chronic granulomatous disease (CGD) panel	5	*CYBA, CYBB, NCF1, NCF2, NCF4*
○ 601	Comprehensive SCID panel	26	*ADA, AK2, ATM, CD3D, CD3E, CD3Z, CORO1A, DCLRE1C (ARTEMIS), DOCK8, FOXN1, IL2RG, IL7R, JAK3, LIG4, NHEJI, ORAI1, PNP, PRKDC, PTPRC, RAC2, RAG1, RAG2, RMRP, STIM1, TBX1, ZAP70*
○ 678	Hyper-IgE syndrome panel	4	*DOCK8, SPINK5, STAT3, TYK2*
○ T995	Hyper-IgM panel	4	*AICDA, CD40, CD40LG, UNG*
NEUROLOGIC DISORDERS			
○ 547	Aicardi-Goutieres syndrome panel^	4	*RNASEH2A, RNASEH2B, RNASEH2C, TREX1*
○ 526	Cerebral cavernous malformations panel	3	*CCM2, KRIT1, PDCD10*
○ TB51	Comprehensive holoprosencephaly panel	17	*CDON, DISP1, DLL1, FGF8, FGFR1, FOXH1, GLI2, GAS1, NODAL, PTCH1, SHH, SIX3, SMAD2, STIL, TDGF1, TGIF1, ZIC2*
○ 2371	Holoprosencephaly panel	4	*SHH, SIX3, TGIF, ZIC2*
PULMONARY DISORDERS			
○ TB48	Hermansky-Pudlak syndrome panel	10	*AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6*
○ TB46	Primary ciliary dyskinesia panel	30	*ARMC4, C21ORF59 (CFAP298), CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, DNAAF1, DNAAF2, DNAAF3, DNAAF5 (HEATR2), DNAH11, DNAH5, DNAI1, DNAI2, DNAJB13, DRC1, DYX1C1 (DNAAF4), GAS8, LRRC6, PIH1D3, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10*
○ TB49	Surfactant dysfunction panel	5	*ABCA3, CSF2RA, CSF2RB, SFTPB, SFTPC*
RENAL AND GASTROINTESTINAL DISORDERS			
○ TG20	Alagille Syndrome	3	*ATP8B1, JAG1, NOTCH2*
○ TG21	Alport Syndrome	6	*CD151^, COL4A3, COL4A4, COL4A5, COL4A6, MYH9*
○ TG96	Bartter Syndrome	12	*AP2S1, BSND, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, GNA11, HSD11B2, KCNJ1, MAGED2, SLC12A1*
○ TG23	Cystic Kidney and Liver Diseases	49	*AHI1^, ALG8^, ALG9, ANKS6, BICC1, CC2D2A, CEP120^, CEP290, CEP83, COL4A1, CRB2, CSPP1^, GANAB, GLIS2, GLIS3, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MKKS^, MKS1, NEK8, NOTCH2, NPHP1, NPHP3, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, PRKCSH, RMND1^, RPGRIP1L, SEC61A1, SEC63, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR35^*
○ TG97	Distal Renal Tubular Acidosis	6	*ATP6V0A4, ATP6V1B1, CA2, HNF4A, SLC34A1, SLC4A1*
○ TG98	Hypokalemia and Related Disorders	38	*AP2S1, ATP6V0A4, ATP6V1B1, BSND, CA2, CACNA1D, CACNA1H, CACNA1S, CASR, CLCNKA^, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FAM111A, FXYD2, GNA11, HNF1B, HNF4A, HSD11B2, KCNA1, KCNJ1, KCNJ10, KCNJ5, MAGED2^, MAGT1, PCBD1, SARS2, SCN4A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC26A3, SLC34A1, SLC4A1, TRPM6*
○ TH01	Nephrolithiasis and Nephrocalcinosis	41	*ADCY10, AGXT, ALPL, AP2S1, APRT, ATP6V0A4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKA^, CLCNKB, CLDN16, CLDN19, CLPB^, CYP24A1, FAM20A, GNA11, GPHN, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, LRP2^, MAGED2^, MOCOS, OCRL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, VDR, XDH*
○ TG99	Nephrotic Syndrome/ Focal Segmental Glomerulosclerosis	55	*ACTN4, ADCK4 (COQ8B), ALG1, ANLN, APOL1*, ARHGAP24, ARHGDIA, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, CRB2, CUBN, DGKE, EMP2, FAN1, FAT1, FN1, GLA, INF2, ITGA3, ITGB4, KANK1, KANK2, KANK4, LAMB2, LMX1B, MAGI2, MYH9, MYO1E, NEIL1, NPHP1, NPHS1, NPHS2, NUP107, NUP205, NUP93, OCRL, PAX2, PDSS2, PLCE1, PMM2, PTPRO, SCARB2, SGPL1^, SMARCAL1, STS, TBC1D8B^, TRPC6, TTC21B, WDR73, WT1, XPO5* *G1 and G2 Risk Alleles Only
○ TG22 ○ TG25	Polycystic Kidney Disease Rest of Cystic Kidney and Liver Diseases after Polycystic Kidney Disease	7	*GANAB, HNF1B, PKD1, PKD2, PKHD1, PRKCSH, TSC2** *Test is designed to identify a contiguous gene deletion involving *PKD1* and *TSC2*, not to identify sequencing and exon-level copy number variants of *TSC2.*
○ TG27 ○ TG24	Polycystic Liver Disease Rest of Cystic Kidney and Liver diseases after Polycystic Liver Disease	8	*ALG8^, GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63*
○ TG90	Primary Hyperoxaluria	3	*AGXT, GRHPR, HOGA1*
○ TG26	Senior-Loken Syndrome	6	*CEP290, INVS, IQCB1, NPHP1, NPHP3, TRAF3IP1*

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RARE DISORDERS TEST REQUISITION FORM

GeneDx Account #	Account Name	
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RARE DISORDERS MULTI-GENE PANELS

TEST CODE	TEST NAME	# GENES	GENE LIST
REPRODUCTIVE DISORDERS			
○ T991	Neonatal 46, XY disorders of sex development (DSD) panel	19	*AR, ARX, ATRX, CHD7, CYP11A1, CYP17A1, DHCR7, DHH, DYNC2H1, HSD17B3, HSD3B2, NEK1, NR5A1, POR, SOX9, SRD5A2, SRY, STAR, WT1*
○ 677	Premature ovarian failure panel	22	*BMP15, CYP17A1, CYP19A1, ESR1, FGFR1, FIGLA, FSHR, GDF9, KISS1, KISS1R, LHB, LHCGR, NOBOX, NR5A1, POR, PROK2, PROKR2, PSMC3IP, SEMA3A, TAC3, TACR3, WDR11*
RHEUMATOLOGIC DISORDERS			
○ 367	Comprehensive panel for periodic fever syndromes panel: Familial Hibernian fever/TRAPS; Familial Mediterranean fever; Hyper-IgD syndrome; Muckle Wells/familial cold urticaria, NOMID; Cyclic neutropenia; PAPA syndrome; Majeed syndrome^	6	*ELANE (ELA2), LPIN2, MEFV, MVK, NLRP3 (CIAS1), PSTPIP1, TNFRSF1A*
SKELETAL DISORDERS			
○ TA45	Abnormal mineralization panel	17	*ALPL, ANKH, AP2S1, CASR, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, GNA11, PHEX, SLC34A1, SLC34A3, SLC9A3R1, VDR*
○ J799	Achondrogenesis panel	3	*COL2A1, SLC26A2, TRIP11*
○ T992	Autosomal dominant osteogenesis imperfecta panel	3	*COL1A1, COL1A2, IFITM5*
○ J804	Chondrodysplasia punctata panel	5	*AGPS, ARSE, EBP, GNPAT, PEX7*
○ TA40	Craniosynostosis panel	30	*ALPL, ALX4, ASXL1, CDC45, CYP26B1, EFNB1, ERF, FGFR1, FGFR2, FGFR3, GLI3, IFT122, IFT43, IL11RA, MASP1, MEGF8, MSX2, P4HB, POR, RAB23, RECQL4, SEC24D, SKI, TCF12, TGFBR1, TGFBR2, TMCO1, TWIST1, WDR35, ZIC1*
○ TA41	Ectrodactyly/split hand-split foot malformation panel	13	*BLHHA9, CDH3, DLX5, DYNC11 (del/dup only), FGFR1, TP63, WNT10B, LBX1, BTRC, POLL, DPCD, FBXW4, 10q24 (chr10:102 962, 134-103, 476, 346)*
○ J800	FGFR-related disorders panel	2	*FGFR2, FGFR3*
○ T996	Hereditary multiple exostoses panel	3	*EXT1, EXT2, PTPN11*
○ TA42	Limb abnormalities and reduction defects panel	71	*ANKRD11, ARHGAP31, ARID1A, ARID1B, BHLHA9, BMP2, BMPR1B, CC2D2A, CDH3, CEP290, CHSY1,DLL4, DLX5, DOCK6, DVL1, DVL3, DYNC1I1, EOGT, ESCO2, FGF10, FGF16, FGFR1, FGFR2, FGFR3, GDF5, GLI3, GNAS, HDAC4, HDAC8, HOXD13, IHH, KIF7, KMT2A, LMBR1 (including ZRS regulatory region), LRP4, MGP, MKS1, MYCN, NIPBL, NOG, NOTCH1, NSDHL, PHF6, PIGV, PTHLH, RAD21, RBPJ, RECQL4, RBM8A, ROR2, RPGRIP1L, SALL1, SALL4, SHH, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SOX11, SOX9, TBX15, TBX3, TBX5, THPO, TP63, WNT10B, WNT3, WNT5A, WNT7A and deletion/duplication coverage for 10q24*
○ J797	Osteogenesis imperfecta panel	24	*ALPL, ANO5, B3GAT3, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, P3H1(LEPRE1), P4HB, PLOD2, PLS3, PP1B, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TAPT1, TMEM38B, WNT1*
○ T994	Hypophosphatasia and hypophosphatemic rickets panel	9	*CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FGF23, PHEX, SLC34A3, VDR*
○ TA43	Skeletal dysplasia panel	29	*ALPL, ARSE, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, DDR2, EBP, FGFR3, FLNB, HSPG2, INPPL1, LBR, LIFR, MMP9, MMP13, NKX3-2, NSDHL, PEX7, PTH1R, RMRP, SBDS, SLC26A2, SLC35D1, SOX9, TRIP11, TRPV4*

RARE DISORDERS SINGLE GENE TESTS

TEST CODE	TEST NAME	GENE	TEST CODE	TEST NAME	GENE
DERMATOLOGIC DISORDERS - CONGENITAL ICHTHYOSIS					
○ 1181	Epidermolytic ichthyosis (epidermolytic hyperkeratosis)	*KRT1, KRT10 hotspots only*	○ 119	Erythrokeratodermia variabilis	*GJB3^, GJB4^*
○ 122	Epidermolytic ichthyosis (epidermolytic hyperkeratosis)	*KRT2 hotspots only*	○ TB14	Ichthyosis follicularis with atrichia and photophobia/keratosis follicularis spinulosa decalvans	*MBTPS2*
○ 208	Epidermolytic PPK of Vörner	*KRT9 hotspots only*	○ 130	Syndromic palmoplantar keratoderma Vohwinkel syndrome KID syndrome^	*GJB2 (Cx26)^*
DERMATOLOGIC DISORDERS - CONNECTIVE TISSUE DISORDERS					
○ TB16	Prolidase deficiency	*PEPD^*	○ 2641	Pseudoxanthoma elasticum common mutations	*ABCC6*
○ TA86	Supravalvular aortic stenosis/autosomal dominant cutis laxa	*ELN*	○ 2642	If negative, reflex to: full gene sequencing	
DERMATOLOGIC DISORDERS - ECTODERMAL DYSPLASIA (ED)					
○ 1601E	An/hypohidrotic, X-linked	*EDA1*	○ 306	Focal dermal hypoplasia Goltz syndrome	*PORCN*
○ TB11	An/hypohidrotic ED, autosomal dominant	*EDARADD*	○ 553	Incontinentia pigmenti common deletion and full gene sequencing	*IKBKG/NEMO*
○ TA50	Autosomal recessive/dominant hypohidrotic ED	*EDAR*	○ 2861	Incontinentia pigmenti common deletion-females	*IKBKG/NEMO*
○ 157	Clouston syndrome *GJB6 (Cx30)^*	*GJB6 (Cx30)^*	○ 2862	Ifnegative, reflex to: full gene sequencing	
○ TA80	Ectodermal dysplasia Odonto-onycho-dermal dysplasia Schöpf-Schulz-Passarge syndrome	*WNT10A*			

RARE DISORDERS TEST REQUISITION FORM

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RARE DISORDERS SINGLE GENE TESTS

TEST CODE	TEST NAME	GENE	TEST CODE	TEST NAME	GENE
○ TA53	Epidermolysis bullosa, dystrophic	*COL7A1*	○ 168	Epidermolysis bullosa, simplex	*KRT5, KRT14 hotspots only*
○ 1631	Epidermolysis bullosa, junctional type	*LAM5 hotspots only*			
DERMATOLOGIC DISORDERS - OTHER SKIN/NAIL/HAIR/MUCOSAL DISORDERS					
○ TA79	Bloom syndrome	*BLM*	○ 2091	Pachyonychia congenita	*KRT16, KRT6a hotspots only*
○ TA54	Darier disease	*ATP2A2*	○ 2092	Pachyonychia congenita	*KRT17, KRT6b hotspots only*
○ TA55	Hailey-Hailey disease	*ATP2C1*	○ 2131	White sponge nevus	*KRT4, KRT13 hotspots only*
○ TB15	Haim-Munk syndrome Papillon-Lefevre syndrome	*CTSC*			
DERMATOLOGIC DISORDERS - PIGMENTARY DISORDERS					
○ 189	Hermansky-Pudlak syndrome: Ashkenazi splice mutation	*HPS3*	○ 188	Hermansky-Pudlak syndrome: Puerto Rican mutations	*HPS1^, HPS3*
DERMATOLOGIC DISORDERS - SKIN CANCERS					
○ 714	Birt-Hogg-Dube syndrome	*FLCN*	○ 205	Gorlin syndrome	*PTCH1*
○ 715	Carney complex	*PRKAR1A*	○ 713	Hereditary leiomyomatosis and renal cell cancer	*FH*
○ 195	Cowden syndrome Bannayan-Riley-Ruvalcaba syndrome Macrocephaly/ASD	*PTEN*	○ 2071	Peutz-Jeghers syndrome	*STK11*
DYSMORPHOLOGY & MULTIPLE CONGENITAL ANOMALIES					
○ 491	Aniridia WAGR	*PAX6*	○ J660	Neurofibromatosis type 1	*NF1*
○ 1004	Alagille syndrome	*JAG1*	○ TB27	Oral-facial-digital syndrome type 1	*OFD1 (CXORF5)*
○ 315E	Branchiootorenal syndrome	*EYA1*	○ 2923	Rubinstein-Taybi syndrome	*CREBBP*
○ TB21	CHARGE syndrome	*CHD7*	○ 415E	Simpson-Golabi-Behmel syndrome	*GPC3*
○ 550	Coffin-Lowry syndrome	*RPS6KA3 (RSK2)*	○ 2511	Smith-Magenis syndrome	*RAI1*
○ TA58	Cohen syndrome	*VPS13B*	○ 406	Sotos syndrome	*NSD1*
○ TB26	Craniofrontonasal dysplasia	*EFNB1*	○ TA62	Van der Woude syndrome	*IRF6*
○ TA63	Feingold syndrome	*MYCN*	○ 358	Velocardiofacial syndrome DiGeorge syndrome	*TBX1^*
○ TB20	Hirschsprung disease	*RET*			
ENDOCRINE DISORDERS					
○ 402	17-alpha hydroxylase/17,20-lyase deficiency	*CYP17A1^*	○ 719	Multiple endocrine neoplasia, type 1	*MEN1*
○ TA56	Allgrove (Triple-A) syndrome	*AAAS*	○ 1771	Multiple endocrine neoplasia, types 2A and 2B	*RET^*
○ TA57	Androgen insensitivity syndrome	*AR*	○ TB03	Pendred syndrome DFNB4 Nonsyndromic hearing loss	*SLC26A4*
○ TB19	Autoimmune polyendocrinopathy APECED	*AIRE*	○ TA94	Septo-optic dysplasia	*HESX1*
○ 721	Hyperparathyroidism-jaw tumor syndrome	*CDC73*	○ 332	Von Hippel-Lindau syndrome	*VHL*
HEMATOLOGIC DISORDERS - DYSKERATOSIS CONGENITA (DKC)					
○ 107	DKC, autosomal dominant	*TERC^*	○ 414	DKC, autosomal dominant (exon 6 sequencing only)	*TINF2^*
○ 682	DKC, autosomal dominant/recessive	*TERT^*	○ 108	DKC, X-linked	*DKC1^*

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RARE DISORDERS TEST REQUISITION FORM

GeneDx an OPKO Health Company

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RARE DISORDERS SINGLE GENE TESTS

TEST CODE	TEST NAME	GENE	TEST CODE	TEST NAME	GENE
HEMATOLOGIC DISORDERS - BONE MARROW FAILURE					
○ TA47	Congenital amegakaryocytic thrombocytopenia	*MPL*	○ TA97	X-linked thrombocytopenia –or– X-linked neutropenia	*WAS*
○ 109	Shwachman-Diamond syndrome	*SBDS^*			
HEMATOLOGIC DISORDERS - OTHER					
○ 2341	Hereditary angioedema (HAE) type I/II	*SERPING1 (C1NH)*	○ 388	Hereditary angioedema type III exon 9/Thr328 mutation only	*F12^*
IMMUNOLOGIC DISORDERS					
○ 2862	Ectodermal dysplasia with immunodeficiency Incontinentia pigmenti	*IKBKG/NEMO^*	○ TA70	Severe congenital neutropenia, autosomal recessive	*HAX1*
○ TA69	IRAK4 deficiency	*IRAK4*	○ 154	X-linked Agammaglobulinemia	*BTK*
○ TA48	Severe congenital neutropenia, autosomal dominant	*ELANE (ELA2)*			
NEUROLOGICAL DISORDERS					
○ TA81	Angelman Angelman-like syndrome	*SLC9A6*	○ TA78	Tyrosine hydroxylase deficiency	*TH*
○ TA60	Congenital insensitivity to pain and anhidrosis	*NTRK1*	○ 548	X-linked early infantile epileptic encephalopathy Atypical Rett syndrome West syndrome	*CDKL5*
○ TB12	Erythromelalgia Paroxysmal extreme pain disorder Small fiber neuropathy Congenital insensitivity to pain	*SCN9A*	○ 552	X-linked hydrocephalus, X-linked spastic paraplegia MASA CRASH syndrome	*L1CAM*
○ 549	Rett/atypical Rett syndromes	*MECP2*			
PULMONOLOGY DISORDERS					
○ T829	Cystic fibrosis/congenital bilateral absence of the vas deferens	*CFTR*			
RENAL DISORDERS					
○ TA64	Alport syndrome	*COL4A5*	○ TA59	Dent disease X-linked recessive nephrolithiasis	*CLCN5*
○ TA71	Branchiootic syndrome 3	*SIX1*	○ T422	Polycystic kidney disease, deletion/duplication only	*PKD1/PKD2/TSC2*
○ TA73	Dent disease 2 Lowe syndrome	*OCRL*	○ TB29	Renal-Coloboma syndrome Papillorenal syndrome	*PAX2*
REPRODUCTIVE DISORDERS - DISORDERS OF SEXUAL DIFFERENTIATION					
○ 339	Adrenal hyperplasia POR deficiency	*POR^*	○ 259	XY gonadal dysgenesis	*SRY^*
○ TA89	X-linked adrenal hypoplasia congenita	*NR0B1 (DAX1)*			
REPRODUCTIVE DISORDERS - INFERTILITY					
○ 522	FMR1-associated premature ovarian failure, CGG repeat analysis only	*FMR1*			
RHEUMATOLOGIC DISORDERS					
○ 215	Familial Hibernian fever TRAPS exons 2-5 sequencing only	*TNFRSF1A*	○ 216	Hyper-IgD syndrome (MVK) exons 8 and 10 sequencing only	*MVK*
○ 214	Familial Mediterranean fever exons 2, 3 and 10 sequencing only	*MEFV*	○ 217	Muckle-Wells Familial cold urticaria NOMID exon 3 sequencing only	*CIAS1*
SKELETAL DISORDERS					
○ TA74	Campomelic dysplasia	*SOX9*	○ 472	Grieg cephalopolysyndactyly syndrome	*GLI3*
○ 225	Cartilage-hair hypoplasia and associated disorders	*RMRP^*	○ TB22	Holt-Oram syndrome	*TBX5*
○ 285	Cherubism	*SH3BP2^*	○ TB13	KBG syndrome	*ANKRDI1*
○ 282E	Chondrodysplasia punctata, X-linked	*ARSE*	○ TA61	Pseudoachondroplasia Multiple epiphyseal dysplasia	*COMP*
○ TB31	Familial hypocalciuric hypercalcemia	*CASR*	○ 1861E	X-linked dominant hypophosphatemia	*PHEX*

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INFORMED CONSENT

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

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.