# RARE DISORDERS TEST REQUISITION FORM

GeneDx

## PATIENT INFORMATION

| | | |
|---|---|---|
| First Name | Last Name | |
| Genetic Sex ○ Male ○ Female<br>Gender Identification (optional): _____ | Date of Birth (mm/dd/yy) | |
| Ancestry ○ White/Caucasian ○ Hispanic ○ Black/African American<br>○ Native American ○ East Asian ○ South Asian<br>○ Middle Eastern ○ Ashkenazi Jewish ○ Other: _____ | | |
| Email | | |
| Address | | |
| City | State | Zip Code |
| Primary Phone | Is this patient deceased? ○ Yes ○ No<br>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: _____<br>(2-4 weeks of wait time is required for some testing) | |
| Patient has had an allogenic bone marrow transplant ○ Yes ○ No<br>Fibroblasts are recommended for patients who had an allogenic bone marrow transplant.<br>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<br>Fax #/Email: _____ | | |
| **Additional Reporting Provider's Name** | | |
| Send Report Via ○ Fax ○ Email ○ Portal<br>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<br>○ Self<br>○ Spouse ○ Child<br>○ Other: _____ | | Policy Holder's Name<br>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:<br>○ 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 |
|---|---|

# RARE DISORDERS TEST REQUISITION FORM

GeneDx

| GeneDx Account # | Account Name | |
|---|---|---|
| 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
○ Targeted Mosaic Variant Testing (Insurance Billing NOT Accepted; Patient Bill or Institutional Bill MUST be selected on page 1)

| Proband Name | Relationship to Proband | Proband GeneDx Accession # |
|---|---|---|
| | | |

Non-GeneDx Test:
○ 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: _____

### WRITE-IN TEST SELECTION

Write-in Test Selection: ○ Test Code: _____ ○ Test Name: _____

Write-in Test Selection: ○ Test Code: _____ ○ Test Name: _____

## HISTORY

FAMILY HISTORY: ○ No Known Family History ○ Pedigree Attached ○ Adopted Consanguinity: ○ Yes ○ No

| 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): _____

# RARE DISORDERS TEST REQUISITION FORM

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## RARE DISORDERS MULTI-GENE PANELS

| TEST CODE | TEST NAME | # GENES | GENE LIST |
|---|---|---|---|
| **DERMATOLOGIC DISORDERS** | | | |
| ○ 708 | Slice - Congenital Ichthyosis | 49 | *ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, ARSL (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 | Slice - Epidermolysis Bullosa (EB) | 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* |
| ○ 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* |
| ○ 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 | 36 | *ANOS1, CHD7, CYP19A1, DUSP6, ESR1, FEZF1, FGF17, FGF8, FGFR1, FLRT3^, FSHB, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R^, LEP, LEPR, LHB, LHCGR, NR0B1, NR5A1, NSMF, POLR3B, PROK2, PROKR2, PROP1, SEMA3A, SEMA3E, SOX10, SPRY4, TAC3, TACR3, WDR11, XRCC4^* |
| ○ 674 | Maturity-onset diabetes of the young (MODY) panel | 18 | *ABCC8, APPL1, BLK, CEL, GCK, GLUD1, HADH, HNF1B, HNF4A, HNF1A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1 (IPF1), RFX6, WFS1* |
| **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

# RARE DISORDERS TEST REQUISITION FORM

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

## 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* |
| ○ 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* |
| ○ 602 | B+ SCID sub-panel | 17 | *TM, CD3D, CD3E, CD3Z, CORO1A, DOCK8, FOXN1, IL2RG, IL7R, JAK3, ORAI1, PNP, PTPRC, RMRP, STIM1, TBX1, ZAP70* |
| ○ 603 | B- SCID sub-panel | 9 | *ADA, AK2, DCLRE1C (ARTEMIS), LIG4, NHEJ1, PRKDC, RAC2, RAG1, RAG2* |
| ○ 678 | Hyper-IgE syndromes panel | 5 | *DOCK8, PGM3^, SPINK5, STAT3, TYK2* |
| ○ T995 | Hyper IgM syndromes panel | 4 | *AICDA, CD40, CD40LG, UNG* |
| **NEUROLOGIC DISORDERS** | | | |
| ○ TJ37 | Aicardi-Goutieres syndrome panel | 7 | *ADAR^, IFIH1^, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1^, 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 and Progressive Familial Intrahepatic Cholestasis | 6 | *ABCB11, ABCB4, ATP8B1, JAG1, NOTCH2, TJP2* |
| ○ 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*<br>*G1 and G2 Risk Alleles Only |
| ○ TG22<br>○ TG25 | Polycystic Kidney Disease<br>Rest of Cystic Kidney and Liver Diseases after Polycystic Kidney Disease | 7 | *GANAB, HNF1B, PKD1, PKD2, PKHD1, PRKCSH, TSC2**<br>*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<br>○ TG24 | Polycystic Liver Disease<br>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

| 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 | 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^ | 7 | *ELANE (ELA2), LPIN2, MEFV, MVK, NLRP3 (CIAS1), PSTPIP1, TNFRSF1A* |
| **SKELETAL DISORDERS** | | | |
| ○ TA45 | Abnormal mineralization panel | 20 | *ALPL, ANKH, AP2S1, CASR, CLCN5, CTNS^, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, GALNT3, 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 | 11 | *AGPS, ARSL (ARSE), EBP, FAR1, GGCX^, GNPAT, LBR, MGP, NSDHL, PEX5, 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 | 10 | *BLHHA9, CDH3, DLX5, DYNC11 (del/dup only), FGFR1, TP63, WNT10B, and deletion/duplication coverage for 10q24 (FBXW4, LBX1, POLL)* |
| ○ TG50 | FGFR-related disorders panel | 3 | *FGFR1, FGFR2, FGFR3* |
| ○ T996 | Hereditary multiple exostoses panel | 3 | *EXT1, EXT2, PTPN11* |
| ○ TA42 | Limb abnormalities and reduction defects panel | 74 | *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 (FBXW4, LBX1, POLL)* |
| ○ 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 | Hypophosphatemic rickets panel | 12 | *CLCN5, CTNS^, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, PHEX, SLC34A3, VDR* |
| ○ TA43 | Skeletal dysplasia panel | 29 | *ALPL, ARSL (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 - CONNECTIVE TISSUE DISORDERS** | | | | | |
| ○ TJ38 | 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 | *ABCC6* |
| **DERMATOLOGIC DISORDERS - ECTODERMAL DYSPLASIA (ED)** | | | | | |
| ○ 1601E | An/hypohidrotic ectodermal dysplasia, X-linked | *EDA1* | ○ 306 | Focal dermal hypoplasia<br>Goltz syndrome | *PORCN* |
| ○ TB11 | An/hypohidrotic ectodermal dysplasia, autosomal dominant/recessive | *EDARADD* | ○ 553 | Incontinentia pigmenti<br>- full gene sequencing and common deletion concurrently | *IKBKG/NEMO* |
| ○ TA50 | An/hypohidrotic ectodermal dysplasia, autosomal dominant/recessive | *EDAR* | ○ 2861<br>○ 2862 | Incontinentia pigment<br>- Common deletion<br>- If negative reflex to: full gene sequencing | *IKBKG/NEMO* |
| ○ 157 | Clouston syndrome *GJB6 (Cx30)^* | *GJB6 (Cx30)^* | | | |
| ○ TA80 | Ectodermal dysplasia<br>Odonto-onycho-dermal dysplasia<br>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 |
|---|---|---|---|---|---|
| **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* | ○ TB15 | Haim-Munk syndrome<br>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<br>Bannayan-Riley-Ruvalcaba syndrome<br>Macrocephaly/ASD | *PTEN* | ○ 2071 | Peutz-Jeghers syndrome | *STK11* |
| **DYSMORPHOLOGY & MULTIPLE CONGENITAL ANOMALIES** | | | | | |
| ○ 491 | Aniridia WAGR | *PAX6* | ○ J660 | Neurofibromatosis type 1 | *NF1* |
| ○ TB27 | Oral-facial-digital syndrome type 1 | *OFD1 (CXORF5)* | ○ 315E | Branchiootorenal syndrome | *EYA1* |
| ○ TB21 | CHARGE syndrome | *CHD7* | ○ 2923 | Rubinstein-Taybi syndrome | *CREBBP* |
| ○ 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<br>DiGeorge syndrome | *TBX1^* |
| ○ TB20 | Hirschsprung disease | *RET* | | | |
| **ENDOCRINE DISORDERS** | | | | | |
| ○ TA56 | Allgrove (Triple-A) syndrome | *AAAS* | ○ 719 | Multiple endocrine neoplasia, type 1 | *MEN1* |
| ○ TA57 | Androgen insensitivity syndrome | *AR* | ○ 1771 | Multiple endocrine neoplasia, types 2A and 2B | *RET^* |
| ○ TB19 | Autoimmune polyendocrinopathy<br>APECED | *AIRE* | ○ TB03 | Pendred syndrome<br>DFNB4 Nonsyndromic hearing loss | *SLC26A4* |
| ○ 721 | Hyperparathyroidism-jaw tumor syndrome | *CDC73* | ○ TA94 | Septo-optic dysplasia | *HESX1* |
| ○ 332 | Von Hippel-Lindau syndrome | *VHL* | | | |
| **HEMATOLOGIC DISORDERS - DYSKERATOSIS CONGENITA (DKC)** | | | | | |
| ○ 107 | DKC, autosomal dominant | *TERC^* | ○ 682 | DKC, autosomal dominant/recessive | *TERT^* |

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

# RARE DISORDERS TEST REQUISITION FORM

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## RARE DISORDERS SINGLE GENE TESTS

| TEST CODE | TEST NAME | GENE | TEST CODE | TEST NAME | GENE |
|---|---|---|---|---|---|
| **HEMATOLOGIC DISORDERS - BONE MARROW FAILURE** | | | | | |
| ○ 109 | Shwachman-Diamond syndrome | *SBDS^* | ○ TA97 | X-linked thrombocytopenia –or– X-linked neutropenia | *WAS* |
| **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* | ○ TA60 | Congenital insensitivity to pain and anhidrosis | *NTRK1* |
| ○ TB12 | Erythermalgia Paroxysmal extreme pain disorder Small fiber neuropathy Congenital insensitivity to pain | *SCN9A* | ○ 552 | X-linked hydrocephalus, X-linked spastic paraplegia MASA CRASH syndrome | *L1CAM* |
| **PULMONOLOGY DISORDERS** | | | | | |
| ○ T829 | Cystic fibrosis/congenital bilateral absence of the vas deferens | *CFTR* | | | |
| **RENAL DISORDERS** | | | | | |
| ○ TA64 | Alport syndrome, X-linked | *COL4A5* | ○ TA59 | Dent disease X-linked recessive nephrolithiasis | *CLCN5* |
| ○ TA71 | Branchiootic syndrome 3 | *SIX1* | ○ TB29 | Renal-Coloboma syndrome Papillorenal syndrome | *PAX2* |
| ○ TA73 | Dent disease 2 Lowe syndrome | *OCRL* | | | |
| **REPRODUCTIVE DISORDERS - DISORDERS OF SEXUAL DIFFERENTIATION** | | | | | |
| ○ 259 | XY gonadal dysgenesis | *SRY^* | | | |
| **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 | *NLRP3 (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 | *ANKRD11* |
| ○ TA61 | Pseudoachondroplasia Multiple epiphyseal dysplasia | *COMP* | ○ TB31 | Familial hypocalciuric hypercalcemia | *CASR* |
| ○ 1861E | X-linked dominant hypophosphatemia | *PHEX* | | | |

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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 | |
|---|---|---|
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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

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**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).