

RAPID SEQUENCING TEST REQUISITION FORM

PATIENT INFORMATION		
First Name	Last Name	
Genetic Sex <input type="radio"/> Male <input type="radio"/> Female Gender Identification (optional): _____	Date of Birth (mm/dd/yy)	
Ancestry <input type="radio"/> White/Caucasian <input type="radio"/> Hispanic <input type="radio"/> Black/African American <input type="radio"/> Native American <input type="radio"/> East Asian <input type="radio"/> South Asian <input type="radio"/> Middle Eastern <input type="radio"/> Ashkenazi Jewish <input type="radio"/> Other: _____		
Email		
Address		
City	State	Zip Code
Primary Phone	Is this patient deceased? <input type="radio"/> Yes <input type="radio"/> No Deceased Date: _____	
Individual to be Tested: <input type="radio"/> Affected/Symptomatic <input type="radio"/> Unaffected/Asymptomatic		

SAMPLE INFORMATION	
Date Sample Collected (mm/dd/yy) (required)	Medical Record #
<input type="radio"/> Blood <input type="radio"/> Other (call lab) _____	
Patient has had a blood transfusion <input type="radio"/> Yes <input type="radio"/> No Date of last transfusion _____ (2-4 weeks of wait time is required for some testing)	
Specimens are not accepted for patients who have had allogeneic bone marrow transplants. See www.genedx.com/specimen-requirements for details.	

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 wish to receive ACMG secondary findings.

Signature of Patient/Guardian (required)	Date
Signature of Relative A	Date
Signature of Relative B	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 <input type="radio"/> Fax <input type="radio"/> Email <input type="radio"/> Portal Fax #/Email: _____	
Additional Reporting Provider's Name	
Send Report Via <input type="radio"/> Fax <input type="radio"/> Email <input type="radio"/> Portal Fax #/Email: _____	
SEND ADDITIONAL REPORT COPIES TO:	
Provider Name	GeneDx Acct#
Fax #/Email: _____	

ICD-10 CODES (Required)	
ICD-10 Codes	
Clinical Diagnosis	Age of Onset

STATEMENT OF MEDICAL NECESSITY

By submission of this test requisition and accompanying sample(s), I: (i) authorize and direct GeneDx to perform the testing indicated; (ii) certify that the person listed as the ordering provider is authorized by law to order the test(s) requested; (iii) certify that any custom panel and/or ordered test(s) requested on this test requisition form are reasonable and medically necessary for the diagnosis and/or treatment of a disease, illness, impairment, symptom, syndrome or disorder; (iv) the test results will determine my patient's medical management and treatment decisions of this patient's condition on this date of service; (v) have obtained this patient's and relatives', when applicable, written informed consent to undergo any genetic testing requested; and (vi) that the full and appropriate diagnosis code(s) are indicated to the highest level of specificity.

Signature of Provider (required)	Date
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PAYMENT OPTIONS (Select One)

<input type="radio"/> PATIENT BILL	If Patient Bill is selected, I am electing to be treated as a self-pay patient for this testing. I agree that neither GeneDx nor I will submit a claim to my insurance for this testing, if I have insurance. GeneDx will send an invoice to the patient listed above.	
Amount Due: _____	Authorized Patient/Guardian Signature	
<input type="radio"/> INSTITUTIONAL BILL	GeneDx Account #	Place Sticker/Stamp Here
	Hospital/Lab Name	

GeneDx Account #	Account Name	
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CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

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

REQUESTED GENES AND DIFFERENTIAL DIAGNOSIS

Genes of Interest: _____

Differential Diagnosis: _____

Pre/Perinatal History

- Cystic hygroma
- Diaphragmatic hernia
- Encephalocele
- Growth delay
- Increased nuchal translucency
- Intrauterine Growth Retardation
- Nonimmune hydrops fetalis
- Oligohydramnios
- Omphalocele
- Polyhydramnios
- Prematurity GA: _____
- Prolonged neonatal jaundice

Structural Brain Abnormalities

- Abnormal myelination
- 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
- Arnold Chiari malformation
- Cerebellar atrophy
- Heterotopia (Periventricular nodular heterotopia)
- Holoprosencephaly
- Hydrocephalus
- Leukodystrophy
- Lissencephaly
- Pachygyria
- Polymicrogyria
- Ventriculomegaly

Developmental/Behavioral Findings

- Absent speech
- Aggressive behavior
- Anxiety
- Autistic Behavior
- Cognitive impairment
- Delayed speech & language development
- Developmental regression
- Dysarthria
- Gait disturbance
- Global developmental delay
- Hyperactivity
- Incoordination
- Intellectual disability
- Learning disability
- Memory impairment
- Sleep disturbance
- Stereotypy

Neurological Findings

- Abnormality of nervous system
- Ataxia
- Cerebral palsy
- Chorea
- Cortical Visual Impairment
- Dementia
- Dysarthria
- Dyskinesia
- Dysphasia
- Dystonia
- Encephalopathy
- Headaches
- Hemiplegia
- Infantile Spasms
- Migraines
- Myoclonus
- Parkinsonism
- Peripheral neuropathy
- Seizures
- Sensory neuropathy
- Spasticity
- Syncope
- Tremors
- Vertigo

Craniofacial/Dysmorphism

- Abnormal facial shape (Dysmorphic features)
Specify: _____
- Brachycephaly
- Cleft lip and/or palate
- Coarse facial features
- Craniosynostosis
- Macrocephaly
- Microcephaly
- Short neck
- Synophrys

Eye Defects/ Vision

- Abnormality of Vision
- Anophthalmia
- Cataracts
- Coloboma
- Corneal opacity
- Ectopia lentis
- External ophthalmoplegia
- Microphthalmia
- Myopia
- Nystagmus
- Optic atrophy
- Optic neuropathy
- Ptosis
- Retinal detachment
- Retinitis pigmentosa
- Strabismus

Hearing Impairment

- Abnormal Newborn Screen: _____
- Conductive hearing impairment
- Sensorineural hearing impairment

Endocrine Findings

- Delayed puberty
- Diabetes Insipidus
- Diabetes Mellitus
- Hyperthyroidism
- Hypophosphatemia
- Hypothyroidism
- Maturity-onset diabetes of the young
- Rickets

Respiratory Findings

- Asthma
- Bronchiectasis
- Hyperventilation
- Hypoventilation
- Pneumothorax
- Pulmonary fibrosis
- Respiratory insufficiency

Hematologic or Immunologic Findings

- Allergic rhinitis
- Anemia
- Immunodeficiency
- Neutropenia
- Pancytopenia
- Recurrent infections
- Thrombocytopenia

Skin/Hair Findings

- Abnormal blistering of the skin
- Abnormality of nail
- Alopecia
- Anhidrosis
- Café-Au-Lait Macules
- Coarse hair
- Cutis Laxa
- Eczema
- Hemangiomas
- Hyperextensible skin
- Hyperpigmentation of the skin
- Hypohidrosis
- Hypopigmentation of the skin
- Ichthyosis
- Skin rash
- Sparse hair
- Telangiectasia
- Vascular skin abnormality
- Velvety skin

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CLINICAL INFORMATION (DETAILED MEDICAL RECORDS MUST BE ATTACHED)

Cardiac Findings

- Abnormal heart morphology
- Amyloidosis
- Aortic root dilation
- Arrhythmia
- Atrial septal defect
- Bicuspid aortic valve
- Bradycardia
- Coarctation of aorta
- Dilated cardiomyopathy
- Heterotaxy
- Hypertension
- Hypertrophic cardiomyopathy
- Mitral valve prolapse
- Noncompaction cardiomyopathy
- Patent ductus arteriosus
- Patent foramen ovale
- Prolonged QTc interval
- Sudden death
- Tetralogy of Fallot
- Ventricular septal defect
- Ventricular tachycardia

Gastrointestinal Findings

- Constipation
- Diarrhea
- Duodenal stenosis/atresia
- Exocrine pancreatic insufficiency
- Failure to thrive
- Feeding difficulties
- Gastroesophageal reflux
- Hepatomegaly
- Inflammatory bowel disease
- Intrahepatic biliary atresia
- Laryngomalacia
- Nausea
- Pancreatitis
- Pyloric stenosis
- Splenomegaly
- Tracheoesophageal fistula
- Vomiting

Genitourinary Findings

- Ambiguous genitalia
- Cryptorchidism
- Cystic renal dysplasia
- Horseshoe kidney
- Hydronephrosis
- Hypospadias
- Inguinal hernia
- Micropenis
- Nephrolithiasis
- Polycystic kidney disease
- Renal agenesis
- Umbilical hernia

Musculoskeletal Findings

- Abnormal connective tissue
- Abnormal form of the vertebral bodies
- Abnormality of the ribs
- Arachnodactyly
- Arthralgia
- Arthrogryposis
- Bruising susceptibility
- Clinodactyly
- Decreased muscle mass
- Ectrodactyly
- Exercise intolerance
- Fatigue
- Hemihypertrophy
- Hypertonia
- Hypotonia
- Joint hypermobility
- Muscle weakness
- Myalgia
- Myopathic facies
- Myopathy
- Osteoarthritis
- Osteopenia
- Pain
- Pectus carinatum
- Pectus excavatum
- Polydactyly
- Recurrent fractures
- Rhabdomyolysis
- Scoliosis
- Short stature
- Skeletal dysplasia
- Syndactyly
- Tall stature

Cancer

- Type: _____
- Location: _____
- Age of onset: _____

Additional Clinical Findings:

Metabolic Findings (Attached relevant lab reports/values)

- Abnormal activity of mitochondrial respiratory chain
- Abnormal Newborn Screen: _____
- Abnormality of mitochondrial metabolism
- Elevated CPK
- Elevated hepatic transaminase
- Hyperammonemia
- Hyperglycemia
- Hypoammonemia
- Hypoglycemia
- Increased serum pyruvate
- Lactic acidosis
- Plasma AA: _____
- Urine OA: _____

Vascular System

- Aneurysm
- Arterial calcification
- Arterial dissection
- Arterial tortuosity
- Arteriovenous malformation
- Epistaxis
- Lymphedema
- Pulmonary hypertension
- Stroke

Other Testing/Imaging (Please provide copy or report if possible)

- Echo: _____
- EEG: _____
- EMG: _____
- Gene Panel: _____
Results: _____
Performed at: _____
- Gene Sequencing*: _____
Results: _____
Performed at: _____
*If you would like us to comment on the presence/absence of previously identified variants, including parental status (if included), provide complete variant information or a copy of the original report.
- Microarray: _____
- MRI: _____
- Muscle Biopsy: _____
- Ultrasound: _____
- X-rays: _____

Signature of provider (required)

Date

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TEST MENU

Directions to Order Rapid Testing

- Client must email GeneDx at Xpress@genedx.com for all rapid testing cases prior to samples arriving
- Trios (proband and both biological parents) are strongly recommended for Xpress tests to increase diagnostic yield and to reduce the number of variants of uncertain significance (VUS); non-trios must be approved by GeneDx prior to samples arriving
- Parental samples must be sent with the proband sample
- For Custom Slice Xpanded®Xpress and Custom Slice Xpanded®Priority, select a gene list below or submit your custom list through the Slice Tool at www.genedx.com for approval before submitting samples
- Fresh blood samples are the preferred specimen type
- Institutional or Self-Pay only

896 XomeDxXpress® (Trios Only)

Related Testing: _____

TH78 GenomeXpress (Trios Only)

TL27 NICUXpress Panel (Trios Preferred)

TG71 Slice Xpanded®Xpress (Trios Preferred)

Custom Slice ID: _____ (gene list must be submitted through the online Slice Tool prior to sample submission)

- Epi Xpanded® gene list
- Congenital Hypotonia Xpanded® gene list
- Microcephaly Xpanded® gene list
- Leukodystrophy Xpanded® gene list

- Verbal results will be called within 7 calendar days* after the proband and biological parental samples are received.
- Please be sure to provide contact information for the verbal results:

Phone: _____ Email: _____

- The written results report will be sent to the ordering provider within approximately 14 days.

* International clients will receive an email status update at 7 days and receive the written report within 2 weeks.

TF37 XomeDx®Priority (Trios Only)

Related Testing: _____

TG75 Slice Xpanded®Priority (Trios Preferred)

Slice ID: _____ (gene list must be submitted through the online Slice Tool prior to sample submission)

- Epi Xpanded® gene list
- Congenital Hypotonia Xpanded® gene list
- Microcephaly Xpanded® gene list
- Leukodystrophy Xpanded® gene list

- The written results report will be sent to the ordering provider within approximately 4 weeks.

BIOLOGICAL RELATIVE SAMPLE INFORMATION

All samples must be received before testing is activated

Mother:	Date of Birth	<input type="radio"/> At GeneDx	<input type="radio"/> Not Available
First Name	Last Name	<input type="radio"/> Asymptomatic	<input type="radio"/> Symptomatic
Father:	Date of Birth	<input type="radio"/> At GeneDx	<input type="radio"/> Not Available
First Name	Last Name	<input type="radio"/> Asymptomatic	<input type="radio"/> Symptomatic
Other (Relationship):	Date of Birth	<input type="radio"/> At GeneDx	<input type="radio"/> Not Available
First Name	Last Name	<input type="radio"/> Asymptomatic	<input type="radio"/> Symptomatic

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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 health 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 health 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.

INFORMED CONSENT

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

APPLICABLE ONLY FOR FULL EXOME SEQUENCING AND GENOME SEQUENCING TESTS

Does not pertain to Xpanded®, Slice, or Panel tests.

Exome/Genome Sequencing Secondary Findings & Opt-Out

As many different genes and conditions are analyzed in an Exome Sequencing or Genome Sequencing test, these tests may reveal some findings not directly related to the reason for ordering ES/GS. Such findings are called "incidental" or "secondary" and can provide information that was not anticipated.

Secondary findings are variants, identified by an Exome Sequencing 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 genome or exome 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.

What will be reported for the proband?

- All known and/or expected pathogenic variants identified in the genes (for which a minimum of 10X coverage was achieved by an Exome Sequencing or a minimum of 15X coverage was achieved by a Genome Sequencing test), as recommended by the ACMG.

What will be reported for relatives?

- The presence or absence for any secondary findings reported for the proband will be provided for all relatives analyzed by an Exome Sequencing or Genome Sequencing test.

Limitations

- 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 variants in that gene.
- 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.