STATEMENT OF MEDICAL NECESSITY

By submission of this test requisition and accompanying sample(s), 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

PATIENT CONSENT

By signing this form, I acknowledge as the patient or relative being tested that I have read the attached informed consent document and that I authorize GeneDx to perform genetic testing as described. For tests that evaluate data from multiple family members concurrently, such as a child and parents, results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their health care providers. I have been informed that GeneDx may contact me or my healthcare provider about research opportunities in the future. For the insurance bill, I understand and agree that GeneDx may use any information required for billing and to be my designated representative for purposes of appealing any denial of benefits.

Signature of Patient/Guardian (required) Date

Signature of Relative A (required) Date

Signature of Relative B (required) Date

PAYMENT OPTIONS

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 the studies listed above.

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

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### Pre/Perinatal History
- Growth delay
- Increased body weight
- Intrauterine growth retardation
- Prematurity GA: __________

### 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
- Brain atrophy
- Cerebellar atrophy
- Cerebellar hypoplasia (Pontocerebellar hypoplasia)
- CNS hypomyelination
- Cortical dysplasia
- Cortical tubers
- Frontotemporal cerebral atrophy
- Heterotopia (Periventricular nodular heterotopia)
- Holoprosencephaly
- Hydrocephalus
- Leukodystrophy
- Lissencephaly
- Molar tooth sign on MRI

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

### Neurological Findings
- Abnormality of nervous system
- Ataxia
- Cerebral palsy
- Chorea
- Cortical Visual Impairment
- Dementia
- Dysesthesia
- Dyskinesia
- Dysphasia
- Dysarthria
- Dystonia
- Encephalopathy
- Epileptic encephalopathy
- Familial Or Sporadic Hemiplegic Migraine
- Febrile Seizures
- Focal Seizures
- Frontotemporal dementia
- Generalized Seizures
- Headaches
- Hyperreflexia
- Infantile Spasms
- Myotonia
- Myoclonus
- Paresthesia
- Parkinsonism
- Peripheral neuropathy
- Reduced tendon reflexes
- Seizures
- Sensory neuropathy
- Spasticity
- Status epilepticus
- Stroke-like episode
- Tremors
- Upper motor neuron dysfunction
- Vocal cord paresis

### Craniofacial/Dysmorphism
- Abnormal facial shape (Dysmorphic features)
- Macrocephaly
- Microcephaly

### Eye Defects/ Vision
- Abnormality of Vision
- Cataracts
- Nyctalagus
- Optic Atrophy

### Hearing Impairment
- Abnormal Newborn Screen:
  - Sensorineural hearing impairment/bilateral

### Cardiac Findings
- Cardiac rhabdomyoma

### Respiratory Findings
- Apnea
- Hyperventilation
- Hypoventilation
- Respiratory distress
- Respiratory insufficiency

### Gastrointestinal Findings
- Failure to thrive
- Feeding difficulties

---

**Signature of provider (required)**

**Date**
NEUROLOGY TEST REQUISITION FORM

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 ○ Mosaic Carrier Testing

○ Known Familial Copy Number variant(s) ○ Known mtDNA Variant(s) Testing (heteroplasmy detection range 25%-100%)

○ Confirmation of Variant Identified in Research Lab ○ Known mtDNA Variant(s) Testing by NGS (heteroplasmy detection range 1.5%-100%) - Test Code 453

○ 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(s): __________________________ Coding DNA (c./m.): __________ Amino Acid (p.): __________ Transcript (NM#): __________

Gene(s): __________________________ Coding DNA (c./m.): __________ Amino Acid (p.): __________ Transcript (NM#): __________

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

Number of Variants: __________

Gene(s): __________________________ Exon #: __________ Coordinates: __________ Genome Build: __________

Gene(s): __________________________ Exon #: __________ Coordinates: __________ Genome Build: __________

TESTING OPTIONS

CUSTOM DEL/DUP TESTING

○ 906 Deletion/Duplication Analysis of ONE nuclear gene ○ 703 Deletion/Duplication Analysis of 2-20 nuclear genes

Write in desired gene(s) to be tested:

FAMILY MEMBER TESTING (NO SEPARATE REPORT, ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS)

○ J767 Ataxia Xpanded, Family Member Testing ○ 910 GenomeDx, Parental Testing

○ 954 Autism/ID Xpanded, Family Member Testing ○ J854 Leukodystrophy Xpanded, Family Member Testing

○ T997 Cerebral Palsy Xpanded, Family Member Testing ○ J513 Microcephaly Xpanded, Family Member Testing

○ TG86 Congenital Hypotonia Xpanded, Family Member Testing ○ J820 Mitoxpanded, Family Member Testing

○ 923 EpiXpanded, Family Member Testing

Mother: First Name: __________________ Last Name: __________________ DOB: __________

○ Asymptomatic ○ Symptomatic ○ Not Available ○ To be sent later**

Father: First Name: __________________ Last Name: __________________ DOB: __________

○ Asymptomatic ○ Symptomatic ○ Not Available ○ To be sent later**

Other: First Name: __________________ Last Name: __________________ DOB: __________

○ Asymptomatic ○ Symptomatic ○ Not Available ○ To be sent later**

Relationship to Proband: ___________________________

** ADDITIONAL SAMPLES MUST BE RECEIVED WITHIN 3 WEEKS

WRITE-IN TEST SELECTION

HISTORY

FAMILY HISTORY

○ No Known Family History ○ Pedigree Attached ○ Adopted

RELATIONSHIP 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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NEURODEVELOPMENTAL DISORDERS AND EPILEPSY

- 522 Fragile X syndrome (FMR1 repeat analysis)
- 910 Chromosomal Microarray (GenomeDx)
- T395 Autism/ID Panel (seq & del/dup of 103 genes)

Order of Reflex Testing:
- Concurrent analysis of 522 & 910, if non-diagnostic activate T395
- Start with 522, if non-diagnostic activate 910, if non-diagnostic activate T395

- 952 Autism/ID Xpanded Panel (2300+ genes, trios preferred)
- 195 PTEN-related disorders (PTEN seq & del/dup)
- 729 Rett/Angelman Related Disorders Panel (seq & del/dup of 20 genes)
- 549 Rett/Atypical Rett syndromes (MECP2 seq & del/dup)
- 566 Angelman syndrome methylaton-MLPA (UPD, deletion)
- 546 Angelman syndrome (UBE3A seq & del/dup)

- 595 Prader-Willi syndrome methylation-MLPA (UPD, deletion)
- 523 Comprehensive Epilepsy Panel (seq & del/dup of 127 genes)
- 814 STAT Epilepsy Panel (seq & del/dup of 26 genes)
- 541 Infantile Epilepsy Panel (seq & del/dup of 111 genes)
- 542 Childhood-Onset Epilepsy Panel (seq & del/dup of 84 genes)
- 544 Progressive Myoclonic Epilepsy Panel (seq & del/dup of 18 genes)
- 545 Rest of the Comprehensive Epilepsy Panel (if subpanel non-diagnostic)
- 921 Epilepsy Panel (1300+ genes, trios preferred)
- 953 Epilepsy Del/Dup Panel (128 genes) (not a trio based test)
- T400 Hemiplegic migraine panel (seq & del/dup of 4 genes)
- 730 Tuberous Sclerosis Panel (TSC1 & TSC2 seq & del/dup)

CNS MALFORMATIONS AND DISORDERS

- 691 Comprehensive Brain Malformations Panel (seq & del/dup of 103 genes)
- 698 Cortical Brain Malformations Panel (seq & del/dup of 61 genes)
- 700 Pontoocerebellar Hypoplasia Panel (seq & del/dup of 19 genes)
- 701 Joubert Syndrome and Related Disorders Panel (seq & del/dup of 29 genes)
- 946 Lissencephaly Panel (seq & del/dup of 26 genes)
- 722 Rest of the Brain Malformations Panel (if subpanel non-diagnostic)
- 689 Microcephaly Panel (seq & del/dup of 65 genes)
- J511 Microcephaly Xpanded Panel (800+ genes, trios preferred)
- 526 Syndromic Macrocephaly/Overgrowth Syndromes Panel (seq & del/dup of 29 genes)
- J853 Leukodystrophy Xpanded Panel (300+ genes, trios preferred)
- 552 X-linked hydrocephalus/X-linked spastic paraplegia/MAF/CRASH syndrome (LICAM seq & del/dup)
- T851 Comprehensive Holoprosencephaly Panel (seq & del/dup of 17 genes)
- 2371 Holoprosencephaly (SHH, ZIC2, SIX3, TGIF seq & del/dup)
- 526 Cerebral cavernous malformations (KRIT1, CCM2, DCCD10 seq & del/dup)
- T844 Dementia Panel (seq only of 11 genes, for patients 18 years and older)

MOVEMENT DISORDERS

- 941 Comprehensive Hereditary Spastic Paraplegia Panel (seq & del/dup of 42 genes)
- 942 Uncomplicated Hereditary Spastic Paraplegia Panel (seq & del/dup of 14 genes)
- 943 Rest of Comprehensive Hereditary Spastic Paraplegia Panel (if subpanel non-diagnostic)
- 944 Hereditary Spastic Paraplegia Related Inborn Error of Metabolism Panel (seq & del/dup of 15 genes)
- T851 Cerebral Palsy Xpanded Panel (1100+ genes, trios preferred)
- J762 Ataxia Xpanded Panel (1300+ genes, trios preferred)
- 527 Dystonia and Parkinsonism Panel (seq & del/dup of 73 genes)
- T402 Dystonia Panel (seq & del/dup of 53 genes)
- T403 Parkinson Disease Panel (seq & del/dup of 29 genes)
- T919 Rest of Dystonia and Parkinsonism Panel (if subpanel non-diagnostic)
- 527 Dopa-responsive dystonia (GCH1 seq & del/dup)
- TA78 Dopa-responsive dystonia/Infantile Parkinsonism/TH deficiency (TH seq & del/dup)
- 218 Alexander disease (GFAP seq)
- 581 Niemann-Pick C disease (NPC1, NPC2 seq)
### NEUROMUSCULAR DISORDERS

<table>
<thead>
<tr>
<th>Code</th>
<th>Panel Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>737</td>
<td>Hereditary Neuropathy Panel (seq &amp; del/dup of 64 genes)</td>
</tr>
<tr>
<td>884</td>
<td>Core CMT Panel (seq &amp; del/dup of 6 genes)</td>
</tr>
<tr>
<td>885</td>
<td>Axonal CMT Panel (seq &amp; del/dup of 32 genes)</td>
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<tr>
<td>886</td>
<td>Demyelinating CMT Panel (seq &amp; del/dup of 23 genes)</td>
</tr>
<tr>
<td>J778</td>
<td>CMT Panel (seq &amp; del/dup of 43 genes)</td>
</tr>
<tr>
<td>T399</td>
<td>Hereditary Sensory and Autonomic Neuropathy Panel (seq del/dup of 14 genes)</td>
</tr>
<tr>
<td>887</td>
<td>Rest of the Hereditary Neuropathy Panel (if subpanel non-diagnostic)</td>
</tr>
<tr>
<td>742</td>
<td>CMT1A/HNPP (PMP22 del/dup)</td>
</tr>
<tr>
<td>888</td>
<td>HNPP/CMT1E (PMP22 seq)</td>
</tr>
<tr>
<td>T812</td>
<td>Erythermalgia/Paroxysmal Extreme Pain Disorder/Small Fiber Neuropathy/Congenital Insensitivity to Pain (SCN9A seq &amp; del/dup)</td>
</tr>
<tr>
<td>363</td>
<td>Familial Amyloid Polyneuropathy (TTR seq)</td>
</tr>
<tr>
<td>820</td>
<td>Spinal &amp; Bulbar Muscular Atrophy (AP repeat analysis)</td>
</tr>
<tr>
<td>889</td>
<td>Neuromuscular Disorders Panel (seq &amp; del/dup of 99 genes)</td>
</tr>
<tr>
<td>890</td>
<td>Limb-Girdle Muscular Dystrophy Panel (seq &amp; del/dup of 30 genes)</td>
</tr>
<tr>
<td>891</td>
<td>Syndromic Congenital Muscular Dystrophy Panel (seq &amp; del/dup of 19 genes)</td>
</tr>
<tr>
<td>892</td>
<td>Congenital Myopathy &amp; Muscular Dystrophy Panel (seq &amp; del/dup of 34 genes)</td>
</tr>
<tr>
<td>893</td>
<td>Myofibrillar Myopathy Panel (seq &amp; del/dup of 8 genes)</td>
</tr>
<tr>
<td>T818</td>
<td>Periodic Paralysis Panel (seq &amp; del/dup of 14 genes)</td>
</tr>
<tr>
<td>894</td>
<td>Rest of Neuromuscular Disorders Panel (if subpanel non-diagnostic)</td>
</tr>
<tr>
<td>787</td>
<td>Duchenne/Becker MD (DMO del/dup)</td>
</tr>
<tr>
<td>786</td>
<td>Duchenne/Becker MD (DMO seq)</td>
</tr>
</tbody>
</table>

### MITOCHONDRIAL DISORDERS

<table>
<thead>
<tr>
<th>Code</th>
<th>Panel Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>J809</td>
<td>MitoXpanded Panel (1800+ genes, trios preferred)</td>
</tr>
<tr>
<td>554</td>
<td>Concurrent full sequence analysis &amp; deletion testing of the mito genome (not a trio based test)</td>
</tr>
<tr>
<td>554</td>
<td>Full sequence analysis and deletion testing of the mitochondrial genome</td>
</tr>
<tr>
<td>704</td>
<td>mtDNA Point Variants Plus Large Deletions Panel</td>
</tr>
<tr>
<td>TH12</td>
<td>Leber Hereditary Optic Neuropathy (LHON) Panel</td>
</tr>
<tr>
<td>T860</td>
<td>Deletion analysis of mito genome</td>
</tr>
<tr>
<td>394</td>
<td>POLG gene sequencing</td>
</tr>
<tr>
<td>615</td>
<td>Combined Mito Genome Plus Mito Focused Nuclear Gene Panel (seq &amp; del/dup of mito genome and 202 nuclear genes)</td>
</tr>
<tr>
<td>573</td>
<td>Mitochondrial Focused Nuclear Gene Panel (seq &amp; del/dup of 202 genes)</td>
</tr>
<tr>
<td>575</td>
<td>Mitochondrial Encephalopathy/Leigh Syndrome Nuclear Gene Panel (seq &amp; del/dup of 134 genes)</td>
</tr>
<tr>
<td>576</td>
<td>Lactic Acidosis/Pyruvate Metabolism Nuclear Gene Panel (seq &amp; del/dup of 130 genes)</td>
</tr>
<tr>
<td>577</td>
<td>Progressive External Ophthalmoplegia (PEO)/Optic Atrophy Nuclear Gene Panel (seq &amp; del/dup of 44 genes)</td>
</tr>
<tr>
<td>578</td>
<td>Methylglutaconic Acidura Nuclear Panel (seq &amp; del/dup of 14 genes)</td>
</tr>
<tr>
<td>Account #</td>
<td>Account Name</td>
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</tbody>
</table>

**NEUROMETABOLIC DISORDERS**

- **J979** Combined Lysosomal and Peroxisomal Disorders Panel (seq & del/dup of 82 genes)
- **T013** Lysosomal Disorders Panel (seq & del/dup of 57 genes)
- **J978** Peroxisomal Disorders Panel (seq & del/dup of 25 genes)
- **J977** Congenital Disorders of Glycosylation Panel (seq & del/dup of 108 genes)
- **J976** Creatine Deficiency Syndromes Panel (seq & del/dup of 3 genes)
- **J985** Disorders of Hyperphenylalaninemia and Bioprotein Metabolism Panel (seq & del/dup of 7 genes)
- **T382** Fatty Acid Oxidation Disorders Panel (seq & del/dup of 15 genes)
- **T010** Hyperammonemia, Urea Cycle and Transporter Defects Panel (seq & del/dup of 48 genes)
- **T012** Metabolic Myopathy Panel (seq & del/dup of 30 genes)
- **T011** Methylmalonic Acidemia, Disorders of Cobalamin Metabolism and Related Disorders Panel (seq & del/dup of 19 genes)
- **J981** Riboflavin Transporter Deficiency and Related Disorders (seq & del/dup of 9 genes)
- **334** Carnitine Palmitoyltransferase II Deficiency (CPT2 seq)
- **2321** Fabry Disease (GLA seq)
- **TG94** Gaucher disease (GBA seq)
- **507** Krabbe Disease (GALC seq & del/dup)
- **TH08** Pompe disease/glycogen storage disease type II (GAA seq and del/dup)
- **TG92** Wilson disease (ATP7B seq & del/dup)
- **J975** X-linked adrenoleukodystrophy (ABCD1 seq & del/dup)

**NEUROFIBROMATOSIS**

- **961** Comprehensive NF panel: NF1, SPRED1, NF2 and SMARCB1 sequencing and deletion/duplication testing
- **962** NF1 panel: NF1 and SPRED1 sequencing and deletion/duplication testing
- **TA65** Reflex to Noonan syndrome and RASopathies panel (sequencing of 25 genes) if 962 is non-diagnostic
- **963** NF2 panel: LZTR1, NF2 and SMARCB1 sequencing and deletion/duplication testing
INFORMED CONSENT

First Name
Last Name
Account Name
Account #
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 change that occurred in my family to develop the disorder in the future. It is possible to test positive for more than one genetic variant.

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 my child is 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 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 your 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.