# DNA Test Results (form D)

Please complete the survey below.

Thank you!

**DNA Test facility:**

**You have completed ... (of Form D - DNA Test Results)**
**10%**

D1.0 - If you have the DNA Test Report, please scan and upload the complete report.

| | |
|---|---|
| D1.1 - Was a genetic exam performed? | ◯ Yes<br>◯ No<br>(if No, please skip DNA Test Results survey.) |
| D1.2 - Please select which facility in which the specimens being tested and analyzed. | ◯ Prevention Genetics (Marshfield, WI)<br>◯ MNG Laboratories (a.k.a. Medical Neurogenetics, LLC, Atlanta, GA)<br>◯ Robert Guthrie Biochemical & Molecular Genetics Laboratory (Buffalo, NY)<br>◯ University of Minnesota Medical Center (Minneapolis, MN)<br>◯ other (specify)<br>(Please select from above drop-down list. e.g. Prevention Genetics) |
| D1.2.1 - If other, please specify | _____ |
| D1.3 - Is the facility, which analyzed the DNA and generated the genetic testing results, a CLIA-approved laboratory? | ◯ Yes<br>◯ No |
| D1.4 - YEAR of the genetic testing results generated? | _____<br>(type in year only (e.g. 2017)) |
| D1.5 - Are the specimens stored in the lab? | ◯ Yes<br>◯ No |

## Gene Searching and Mutations/Variations

## Report 1

**You have completed ... (of Form D - DNA Test Results)**

**20%**

D2.1 - According to the most current interpretation of gene mutation-disease association, this mutation is associated with

◯ Malignant Hyperthermia (MH)
◯ Central Core Disease (CCD)
◯ likely pathologic & associated with Malignant Hyperthermia Susceptible (MHS) or with CCD
◯ Variant of Uncertain Significance (VUS)
◯ Polymorphism (Benign)

D2.2 - Gene Name

◯ RYR1
◯ CACNA1S
◯ STAC3
◯ other (specify)

D2.2.1 - If other, please specify:

_____

D2.3 - Gene symbol

_____
(type in gene symbol, ref. OMIM (e.g. RYR1, ARG2458HIS))

D2.4 - the Exon #:

_____

D2.5 - Aminoacid change

_____
(e.g. p.2458Arg>His)

D2.6 - Nucleotide change

_____
(e.g. c.488G>T)

## Gene Searching and Mutations/Variations

## Report 2

**You have completed ... (of Form D - DNA Test Results)**

**50%**

D3.1 - According to the most current interpretation of gene mutation-disease association, this mutation is associated with

◯ Malignant Hyperthermia (MH)
◯ Central Core Disease (CCD)
◯ likely pathologic & associated with Malignant Hyperthermia Susceptible (MHS) or with CCD
◯ Variant of Uncertain Significance (VUS)
◯ Polymorphism (Benign)

D3.2 - Gene Name

◯ RYR1
◯ CACNA1S
◯ STAC3
◯ other (specify)

D3.2.1 - If other, please specify:

_____

D3.3 - Gene symbol

_____
(type in gene symbol, ref. OMIM (e.g. RYR1, ARG2458HIS))

D3.4 - the Exon #:

_____

D3.5 - Aminoacid change

_____
(e.g. p.2458Arg>His)

D3.6 - Nucleotide change

_____
(e.g. c.488G>T)

## Gene Searching and Mutations/Variations

**Report 3**

**You have completed ... (of Form D - DNA Test Results)**

**60%**

D4.1 - According to the most current interpretation of gene mutation-disease association, this mutation is associated with

◯ Malignant Hyperthermia (MH)
◯ Central Core Disease (CCD)
◯ likely pathologic & associated with Malignant Hyperthermia Susceptible (MHS) or with CCD
◯ Variant of Uncertain Significance (VUS)
◯ Polymorphism (Benign)

D4.2 - Gene Name

◯ RYR1
◯ CACNA1S
◯ STAC3
◯ other (specify)

D4.2.1 - If other, please specify:

_____

D4.3 - Gene symbol

_____
(type in gene symbol, ref. OMIM (e.g. RYR1, ARG2458HIS))

D4.4 - the Exon #:

_____

D4.5 - Aminoacid change

_____
(e.g. p.2458Arg>His)

D4.6 - Nucleotide change

_____
(e.g. c.488G>T)

## Gene Searching and Mutations/Variations

**Report 4**

**You have completed ... (of Form D - DNA Test Results)**

**60%**

D4.1 - According to the most current interpretation of gene mutation-disease association, this mutation is associated with

◯ Malignant Hyperthermia (MH)
◯ Central Core Disease (CCD)
◯ likely pathologic & associated with Malignant Hyperthermia Susceptible (MHS) or with CCD
◯ Variant of Uncertain Significance (VUS)
◯ Polymorphism (Benign)

D4.2 - Gene Name

◯ RYR1
◯ CACNA1S
◯ STAC3
◯ other (specify)

D4.2.1 - If other, please specify:

_____

D4.3 - Gene symbol

_____
(type in gene symbol, ref. OMIM (e.g. RYR1, ARG2458HIS))

D4.4 - the Exon #:

_____

D4.5 - Aminoacid change

_____
(e.g. p.2458Arg>His)

D4.6 - Nucleotide change

_____
(e.g. c.488G>T)

**Gene Searching and Mutations/Variations**

**Report 5**

**You have completed ... (of Form D - DNA Test Results)**

**60%**

D4.1 - According to the most current interpretation of gene mutation-disease association, this mutation is associated with

◯ Malignant Hyperthermia (MH)
◯ Central Core Disease (CCD)
◯ likely pathologic & associated with Malignant Hyperthermia Susceptible (MHS) or with CCD
◯ Variant of Uncertain Significance (VUS)
◯ Polymorphism (Benign)

D4.2 - Gene Name

◯ RYR1
◯ CACNA1S
◯ STAC3
◯ other (specify)

D4.2.1 - If other, please specify:

_____

D4.3 - Gene symbol

_____
(type in gene symbol, ref. OMIM (e.g. RYR1, ARG2458HIS))

D4.4 - the Exon #:

_____

D4.5 - Aminoacid change

_____
(e.g. p.2458Arg>His)

D4.6 - Nucleotide change

_____
(e.g. c.488G>T)

**Other Gene Mutations/Variations:**

**(If you need more space to report additional gene mutations/variations, please contact NAMHR administrator. We will send you the instruction and format how you can submit your complete genetic report.)**

**You have completed ... (of Form D - DNA Test Results)**
**70%**

D5.1 - Were any other GENE sequence variants?

◯ Yes
◯ No

D5.1.1 - If yes, please report with the following order:

(ASSOCIATE_DISEASE, GENE_NAME, GENE_SYMBOL, EXON#, AMINOACID_CHANGE, NUCLEOTIDE_CHANGE)

_____

D5.1.2 - please list all other BENIGN variants not yet included above, with the following order:

(GENE_NAME, GENE_SYMBOL, EXON#, AMINOACID_CHANGE, NUCLEOTIDE_CHANGE)

_____

!This field is for official use only!

Congratulations. You have completed ... (of Form D - DNA Test Results)
100%

## NEXT SURVEY

Would you like to fill out the Clinical Information about this participant?

◯ Yes
◯ No