# 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%

<table>
<thead>
<tr>
<th>D1.0 - If you have the DNA Test Report, please scan and upload the complete report.</th>
</tr>
</thead>
</table>

| D1.1 - Was a genetic exam performed? | ○ Yes |
|               | ○ No |

(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) |
|                                                                                  | ○ MNG Laboratories (a.k.a. Medical Neurogenetics, LLC, Atlanta, GA) |
|                                                                                  | ○ Robert Guthrie Biochemical & Molecular Genetics Laboratory (Buffalo, NY) |
|                                                                                  | ○ University of Minnesota Medical Center (Minneapolis, MN) |
|                                                                                  | ○ other (specify) |

(Please select from above drop-down list. e.g. Prevention Genetics)

<table>
<thead>
<tr>
<th>D1.2.1 - If other, please specify</th>
<th>__________________________</th>
</tr>
</thead>
</table>

| D1.3 - Is the facility, which analyzed the DNA and generated the genetic testing results, a CLIA-approved laboratory? | ○ Yes |
|                                                                                           | ○ No |

<table>
<thead>
<tr>
<th>D1.4 - YEAR of the genetic testing results generated?</th>
<th>(type in year only (e.g. 2017))</th>
</tr>
</thead>
</table>

| D1.5 - Are the specimens stored in the lab? | ○ Yes |
|                                           | ○ 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)
### 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/variants, 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)

<table>
<thead>
<tr>
<th>70%</th>
</tr>
</thead>
<tbody>
<tr>
<td>D5.1 - Were any other GENE sequence variants?</td>
</tr>
</tbody>
</table>

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