1. \*Mutational analysis results available on this participant/ subject:[ ]  Yes [ ]  No (Stop completing form)
2. \*Mutational analysis performed on the participant/ subject:[ ]  Yes [ ]  No
3. \*If no, was mutational analysis performed on a family member? [ ]  Yes [ ]  No
4. If no, provide explanation:

\*Mutational analysis results:

c. Tissue used for analysis

[ ]  Full blood

[ ]  Muscle tissue

[ ]  Buccal smear

[ ]  Urine

[ ]  Others

1. Mutation(s) detected:

[ ]  Repeat allele

Repeat allele length analyzed? [ ]  Yes [ ]  No

 If yes,

Result copy numbers: (number)

Repeat length: kb

[ ]  Homozygous

[ ]  Digenic (mutations in more than one gene)

[ ]  No mutation detected

Were repeat interruptions found [ ]  Yes [ ]  No

If so, which type of interruptions?

Methods used:

[ ]  PCR

[ ]  Triplet-primed PCR

[ ]  Southern blot

1. Are there additional variants/mutations in DMPK gene or ZNF9 gene [ ]  Yes [ ]  No
2. mRNA analysis
3. mRNA analysis performed: [ ]  Yes [ ]  No [ ] Unknown
4. If Yes, were implications confirmed: [ ]  Yes [ ]  No
5. Are target RNA species for alternative splicing investigated : [ ]  Yes [ ]  No
6. Which target RNA species were investigated in which tissue?
7. Mutation Information (HUGO Mutation Nomenclature)
8. cDNA:
9. mRNA: ( e.g. was RNA foci quantification in muscle performed)
10. Protein: (e.g. was immunoassay for RAN translation products performed?))

## General Instructions

This CRF includes data typically recorded for mutation analysis in myotonic dystrophy.

## Specific Instructions

Please see the Data Dictionary for definitions for each of the data elements included in this CRF Module*.* Mutation Information (HUGO Mutation Nomenclature): Please visit the HUGO Mutation Nomenclature website at [Human Genome Variation Society](http://www.hgvs.org/rec.html)

## Optional references

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