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

J Mol Diagn. 2013 Jan;15(1):110-5.

Novel heat pulse extension-PCR-based method for detection of large CTG-repeat expansions in myotonic dystrophy type 1.

Orpana AK1, Ho TH, Alagrund K, Ridanpää M, Aittomäki K, Stenman J

Genet Test Mol Biomarkers. 2012 Dec;16(12):1428-31. .

Triplet-primed PCR is more sensitive than southern blotting-long PCR for the diagnosis of myotonic dystrophy type1.

Addis M1, Serrenti M, Meloni C, Cau M, Melis MA.

Diagn Mol Pathol. 2011 Mar;20(1):48-51.

Effect of unexpected sequence interruptions to conventional PCR and repeat primed PCR in myotonic dystrophy type 1 testing.

Radvansky J1, Ficek A, Minarik G, Palffy R, Kadasi L.

J Mol Diagn. 2013 Jul;15(4):518-25.

Development of a genomic DNA reference material panel for myotonic dystrophy type 1 (DM1) genetic testing.

Kalman L, Tarleton J, Hitch M, Hegde M, Hjelm N, Berry-Kravis E, Zhou L, Hilbert JE, Luebbe EA, Moxley RT 3rd, Toji L.

Genet Med. 2009 Jul;11(7):552-5

Technical standards and guidelines for myotonic dystrophy type 1 testing.

Prior TW; American College of Medical Genetics (ACMG) Laboratory Quality Assurance Committee.

Acta Myol. 2006 Jun;25(1):23-33.

Italian guidelines for molecular analysis in myotonic dystrophies.

Botta A, Bonifazi E, Vallo L, Gennarelli M, Garrè C, Salehi L, Iraci R, Sansone V, Meola G, Novelli G.

Clin Chem Lab Med. 2001 Dec;39(12):1259-62.

A simple and rapid analysis of triplet repeat diseases by expand long PCR.

Hećimović S, Vlasić J, Barisić L, Marković D, Culić V, Pavelić K.

Am J Hum Genet. 1995 Jan;56(1):123-30.

Normal variation at the myotonic dystrophy locus in global human populations.

Zerylnick C, Torroni A, Sherman SL, Warren ST.

Hum Mol Genet. 2010 Apr 15;19(8):1399-412.

Variant CCG and GGC repeats within the CTG expansion dramatically modify mutational dynamics and likely contribute toward unusual symptoms in some myotonic dystrophy type 1 patients.

Braida C, Stefanatos RK, Adam B, Mahajan N, Smeets HJ, Niel F, Goizet C, Arveiler B, Koenig M, Lagier-Tourenne C, Mandel JL, Faber CG, de Die-Smulders CE, Spaans F, Monckton DG.

Clin Chem. 1995 Jan;41(1):69-72.

A molecular protocol for diagnosing myotonic dystrophy.

Guida M, Marger RS, Papp AC, Snyder PJ, Sedra MS, Kissel JT, Mendell JR, Prior TW.

J Biol Chem. 2005 Jan 14;280(2):941-52

The myotonic dystrophy type 1 triplet repeat sequence induces gross deletions and inversions.

Wojciechowska M, Bacolla A, Larson JE, Wells RD.