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:
5. \*Mutational analysis results:
6. Mutation(s) detected:

Homozygous

Hemizygous

Heterozygous – two alleles identified

Heterozygous – only one allele identified

Digenic (mutations in more than one gene)

No mutation detected

1. Are there additional variants in other genes of unknown significance?  Yes  No

If yes, indicate reason:

1. Are there additional genes sequenced with no mutations detected?  Yes  No

If yes, indicate reason:

1. Allele specific Information
2. Allele #1
3. Gene Name:
4. Mutation Class:  Deletion  Duplication  Missense  Nonsense

Splice  Pseudoexon

Potential (variant of unknown significance)

Subexonic Insertion/Deletion  Other, specify:

1. For Exonic Deletions/ Duplications:
2. Was the copy number directly tested for all exons? Yes  No  Unknown
3. Are the limits of deletions and duplications completely defined?

Yes  No  Unknown

1. First Deleted/Duplicated Exon:
2. Last Deleted/Duplicated Exon:
3. Whole gene deletion?  Yes  No  Unknown
4. Predicted reading frame:  In  Out  Unknown
5. Are known gene promoters deleted: Yes  No Unknown
6. For Point Mutations or Pseudoexons:
7. Was the entire coding region sequenced:  Yes  No
8. Targeted mutational analysis only:  Yes  No
9. If Yes, type of analysis:  Hot-spot  Known familial mutation

Other, specify:

1. Point mutation location (choose one):
2. Exon (Point Mutation):
3. Intron:
4. Other:
5. Point mutation subclass information:
6. Insertion Deletion:  Insertion  Deletion  Insertion/Deletion
7. Nonsense Type:  UAA  UAG  UGA

Not applicable

1. mRNA analysis
2. mRNA analysis performed:  Yes  No Unknown
3. If Yes, were implications confirmed:  Yes  No
4. Mutation Information (HUGO Mutation Nomenclature)
5. cDNA: (if relevant, data to be entered by site)
6. mRNA: (if relevant, data to be entered by site)
7. Protein: (if relevant, data to be entered by site)
8. Allele Specific Information
9. Allele #2
10. Was a second disease allele identified?  Yes  No (Stop completing form)
11. Is allele #2 identical to allele #1 (Homozygous only):

Yes (Stop completing form)  No

1. Gene Name: (if relevant, data to be entered by site)
2. Mutation Class:  Deletion  Duplication  Missense  Nonsense

Splice  Pseudoexon

Potential (variant of unknown significance)

Subexonic Insertion/Deletion  Other, specify:

1. For Exonic Deletions/ Duplications:
2. Was the copy number directly tested for all exons?

Yes  No  Unknown

1. Are the limits of deletions and duplications completely defined?

Yes  No  Unknown

1. First Deleted/Duplicated Exon: (if relevant, data to be entered by site)
2. Last Deleted/Duplicated Exon: (if relevant, data to be entered by site)
3. Whole gene deletion?  Yes  No  Unknown
4. Predicted reading frame:  In  Out  Unknown
5. Are known gene promoters deleted:  Yes  No  Unknown
6. For Point Mutations or Pseudoexons:
7. Was the entire coding region sequenced:  Yes  No
8. Targeted mutational analysis only:  Yes  No
9. If Yes, type of analysis:  Hot-spot  Known familial mutation

Other, specify:

1. Point mutation location (choose one):
2. Exon (Point Mutation): (if relevant, data to be entered by site)
3. Intron: (if relevant, data to be entered by site)
4. Other: (if relevant, data to be entered by site)
5. Point mutation subclass information:
6. Insertion Deletion:  Insertion  Deletion  Insertion/Deletion
7. Nonsense Type:  UAA  UAG  UGA  Not applicable
8. mRNA analysis
9. mRNA analysis performed:  Yes  No  Unknown
10. If Yes, were implications confirmed:  Yes  No
11. Mutation Information (HUGO Mutation Nomenclature)
12. cDNA: (if relevant, data to be entered by site)
13. mRNA: (if relevant, data to be entered by site)
14. Protein: (if relevant, data to be entered by site)

## General Instructions

This CRF includes data typically recorded for mutation analysis.

## Specific Instructions

Please see the Data Dictionary for definitions for each of the data elements included in this CRF Module*.*

Important note: Some of the data elements included on this CRF are considered Core (\*) (i.e., strongly recommended for all studies to collect). Other data elements are supplemental and supplemental – highly recommended (\*) and should be collected on clinical trials and only if the research team considers them appropriate for their study.

Mutation Information (HUGO Mutation Nomenclature): Please visit the HUGO Mutation Nomenclature website at [Human Genome Variation Society](http://www.hgvs.org/rec.html)

\*Element is Core for NMD, MG and SMA and Supplemental – Highly Recommended for Cerebral Palsy.