1. \*Family History of the trait: Maternal Side Paternal Side  Unknown

Neither  Both Maternal and Paternal Sides

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. 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*.*

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 classified as Supplemental – Highly Recommended