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