Whether the Genetic Testing was performed on a CLINICAL DIAGNOSTIC basis or on a RESEARCH basis should be mentioned with each test

1. Was genetic testing performed?

[ ]  Yes

[ ]  No

If yes, please answer questions below:

1. What year was the genetic testing performed?[[1]](#footnote-1)
2. Test name and or code
3. What type of testing was performed?

[ ]  Single gene(s) sequence analysis

1. What tissue?

[ ]  Blood

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  Sanger

[ ]  Next Generation Sequencing (NGS)

[ ]  Other, please specify

[ ]  Unknown

[ ]  Single gene(s) deletion analysis

1. What tissue?1

[ ]  Blood

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  Multiplex ligation-dependent probe amplification (MLPA)

[ ]  Fluorescent in situ hybridization (FISH)

[ ]  Other, please specify

[ ]  Unknown

[ ]  Whole exome sequence (WES) analysis

1. What tissue?1

[ ]  Blood

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  Proband

[ ]  Trio

[ ]  Next Generation Sequencing (NGS)

[ ]  Other, please specify

[ ]  Unknown

[ ]  Whole genome sequence analysis

1. What tissue?1

[ ]  Blood

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  Next Genome Sequence (NGS)

[ ]  Other, please specify

[ ]  Unknown

[ ]  mtDNA panel testing (common mutations)

1. What tissue?1

[ ]  Blood

[ ]  Muscle

[ ]  Liver

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  Sanger

[ ]  Next Genome Sequencing (NGS)

[ ]  Restriction PCR

[ ]  Wave-surveyor

[ ]  Other, please specify

[ ]  Unknown

[ ]  Whole mtDNA genome sequence analysis

1. What tissue?1

[ ]  Blood

[ ]  Muscle

[ ]  Liver

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  Sanger

[ ]  Next Generation Sequencing (NGS)

[ ]  Affy MitoChip version (fill in version number)

[ ]  Other, please specify

[ ]  Unknown

1. Comments

[ ]  mtDNA genome deletion/duplication analysis

1. What tissue?1

[ ]  Blood

[ ]  Muscle

[ ]  Liver

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  Southern blot

[ ]  Probe capture-NGS

[ ]  Long-range PCR

[ ]  Other, please specify

[ ]  Unknown

[ ]  mtDNA content analysis

1. What tissue?1

[ ]  Blood

[ ]  Muscle

[ ]  Liver

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  qPCRn

[ ]  Southern blot

[ ]  Other, please specify

[ ]  Unknown

[ ]  Microarray

1. What tissue?1

[ ]  Blood

[ ]  Amnio

[ ]  Skin

[ ]  Other, please specify

1. What genes?
2. Test Methodology:1

[ ]  Affymetrix

[ ]  Agilent

[ ]  Illumnia

[ ]  Nimblegen

[ ]  Other, please specify

[ ]  Unknown

[ ]  Karyotype

1. What tissue?1

[ ]  Blood

[ ]  Amnio

[ ]  Skin

[ ]  Other, please specify

1. What genes?

[ ]  Methylation analysis

1. What tissue?1

[ ]  Blood

[ ]  Other, please specify

1. What genes?

[ ]  Trinucleotide repeat analysis

1. What tissue?1

[ ]  Blood

[ ]  Other, please specify

1. What genes?
1. Supplemental – Highly Recommended [↑](#footnote-ref-1)