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)