Diagnostic Lab name:

Date report issued[[1]](#footnote-1):

Table : Genetic Testing Results

| Gene1 | References | Result | DNA change\* | Test Methodology1 | Tissue1 | Region Tested: Coverage1 | Clinical Category1 |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Data to be entered by site | Data to be entered by site | Mutation Variant  Variant of Unknown Signifi-cance  Unknown | Data to be entered by site | Data to be entered by site | Blood  Amniocytes  Skin  Other: | Data to be entered by site | Definite pathogenic  Likely pathogenic  Variant of uncertain significance  Likely benign  Definitely benign |

**\*** Note : The mtDNA genome is rather small, completely sequenced and numbered. According to current recommendations variants in the mitochondrial DNA should be described in relation to the full mitochondrial DNA sequence, i.e. for human the Homo sapiens mitochondrion, complete genome(GenBank [NC\_012920.1](http://www.ncbi.nlm.nih.gov/nuccore/NC_012920)). Descriptions should be preceded by "m.", like m.8993T>C ([*see Recommendations*](http://www.hgvs.org/mutnomen/recs.html#prefix)). The mtDNA encodes a range of different proteins. To prevent confusion, changes at protein level should be described including a reference to the protein changed, like ATP6:p.Leu156Pro (GenBank [YP\_003024031.1](http://www.ncbi.nlm.nih.gov/protein/251831112), ATP synthase 6). (*HGVS recommendations)*

1. Basis for the evaluation of the clinical category1:

Previously reported[[2]](#footnote-2)

Familial segregation

Prevision software (name of the software)

Experimental validation

Functional complementation in cultured cells

Other

1. For Mitochondrial DNA mutation:
2. Quantitative analyses (Heteroplasmy assessment)1

Evaluation method

Restriction PCR

Deep sequencing

Allele specific PCR

qPCR (deletions, depletion)

Southern blot

Other

Heteroplasmy level1

Blood

Muscle

Urinary sediment

Buccal cells

Other

1. Basis for the clinical evaluation of the clinical category1:

Previously reported

Familial segregation

Tissular segregation

Prevision software (name of the software)

Experimental validation

Transfer into cybrid cells

Single muscle fiber

1. Supplemental – Highly Recommended [↑](#footnote-ref-1)
2. Mito Version 1.0 Page of [↑](#footnote-ref-2)