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)