

### **Supplementary table 1 on line.**

#### **The mean amount of mtDNA (copy number) in peripheral blood for ten control subjects patients.**

Copy number measurements were determined by real-time PCR using iQ Sybr Green on the BioRad ICycler (mean of three measurements from the genomic sample). mtDNA: using a target template spanning from nt 3459 to nt 3569 of the mtDNA *ND1* gene. Nuclear DNA (nDNA) using a target template spanning from nt 804 to nt 903 of the single-copy nuclear gene *GAPDH*. The mean percentage mutated mtDNA  $\Delta Ct$  = difference between the threshold cycles for nDNA and mtDNA. Relative copy number was calculated from the  $\Delta Ct$  value,  $2(2^{-\Delta Ct})$ , to account for the two copies of *GAPDH* in each mononuclear peripheral blood cell. SD = standard deviation.

Subject	mtDNA	SD	nDNA	SD	$\Delta Ct$	Total copy number
1	20.07	0.058	27.67	0.014	-7.6	388.0
2	22.67	0.058	30.20	0.008	-7.5	370.5
3	19.77	0.153	27.17	0.012	-7.4	337.8
4	18.03	0.252	25.77	0.043	-7.7	425.6
5	19.83	0.153	27.10	0.006	-7.2	308.0
6	18.50	0.529	26.40	0.036	-7.9	477.7
7	19.77	0.115	27.37	0.015	-7.6	388.0
8	18.73	0.115	26.43	0.013	-7.7	415.9
9	20.70	0.173	27.80	0.007	-7.1	274.4
10	18.67	0.115	25.97	0.013	-7.3	315.2

### **Supplementary table 2 on line.**

#### **The percentage 3243A>G mutation and amount of mtDNA (copy number) relative to nuclear DNA (nDNA) in skeletal muscle for the study subjects.**

Numbering corresponds to the patient index in the text. The methods used were identical to those in the text. Muscle tissue was not available for subjects 6 and 8.

Subject	3243A>G (%)	mtDNA	SD	nDNA	SD	$\Delta Ct$	Total copy Number / diploid nucleus

<b>1</b>	64.6	20.1	0.115	30.6	0.208	-10.5	2896.3
<b>2</b>	59.9	20.9	0.153	30.5	0.058	-9.7	1625.5
<b>3</b>	85.5	20.1	0.173	29.3	0.300	-9.2	1176.3
<b>4</b>	87.6	19.7	0.058	31.2	0.173	-11.5	5928.0
<b>5</b>	28.6	23.6	0.058	33.8	0.346	-10.2	2298.8
<b>6</b>	-	-	-	-	-	-	-
<b>7</b>	70.8	18.4	0.252	33.0	0.058	-14.6	49667.0
<b>8</b>	-	-	-	-	-	-	-
<b>9</b>	58.3	22.6	0.200	31.8	0.321	-9.2	1203.8
<b>10</b>	38.3	24.3	0.265	35.2	0.300	-10.9	3821.7
<b>11</b>	48.0	22.1	0.231	33.3	0.351	-11.3	4927.6

**Supplementary table 3 on line.**

**The amount of mtDNA (copy number) relative to nuclear DNA (nDNA) in skeletal muscle from six healthy control subjects.** The methods used were identical to those in the text. These samples did not have the 3243A>G mutation using the last fluorescent cycle method described in the text.

Subject	mtDNA	SD	nDNA	SD	ΔCt	Total copy Number / diploid nucleus
1	20.9	0.100	28.8	0.321	-7.9	488.9
2	20.7	0.058	29.3	0.306	-8.7	812.7
3	21.6	0.058	29.8	0.153	-8.3	615.9
4	20.2	0.306	28.0	0.115	-7.8	445.7
5	19.8	0.231	28.2	0.058	-8.4	675.6
6	19.8	0.058	27.7	0.208	-7.9	477.7