



Supp. Figure S1. Frozen muscle - 400 X. **A.** H&E stain Variation in fiber size with isolated small atrophic fibers and hypereosinophilic fibers. **B.** COX Positive coarse granular pattern. **C.** PAS stain: Variation in fiber size with scattered atrophic small fibers. **D.** SDH Positive coarse granular pattern.

Supp. Table S1. Mitochondrial Respiratory Chain Enzymes analysis [RCA] in Muscle

Enzyme	Patient [% of mean]	Controls [+/- SD]	Interpretation
NADH: ferricyanide dehydrogenase [CoI]	421 [150]	280+/-91	Normal
NADH: cytochrome c reductase [Co I+III]	70.1 [137]	51.2+/-8.6	Normal
NADH: cytochrome c reductase, rotenone sensitive [Co I+III]	22.8 [96]	23.6+/-4.6	Normal
Succinate dehydrogenase [CoII]	10.93 [135]	8.1+/-2.5	Normal
Succinate cytochrome C reductase [CoII +III]	3.85 [91]	4.2+/-1.9	Normal
Cytochrome C oxydase [Co IV]	64 [219]	29.2+/-9.1	Normal
Citrate Synthetase	360 [129]	280+/-95	Normal

As per clinical request on frozen portion of muscle, RCA was performed by BaylorCollege of Medicine Genetics Laboratory in Houston, TX.

Interpretation: Deficiencies in mitochondrial electron chains enzymes were not detected using spectrophotometer. The interpretation of the results is done on the assumption that the specimen has been handled properly. This result cannot confirm or exclude the diagnosis of a mitochondrial respiratory chain disorder.

Supp. Table S2. Peptides corresponding to the 110kDa band identified by LC-MS/MS*heat shock 70 protein 4 (Hsp 70)*

Coverage 39.5%

Peptide	Position	P value	XCorr	DeltaCn
AGGIETIANEYSDR	20 - 33	4.68E-09	3.723	0.3
AFSDPFVEAEK	74 - 84	4.55E-06	3.580	0.6
SNLAYDIVQLPTGLTGIK	85 - 102	2.31E-11	4.231	0.5
NFTTEQVTAMLLSK	111 - 124	3.41E-08	3.972	0.4
LMNETTAVALAYGIYK	170 - 185	7.23E-10	4.536	0.6
VLATAFDTTLGGR	222 - 234	1.19E-09	4.047	0.6
LKKEDIYAVEIVGGATR	330 - 346	7.00E-04	2.339	0.4
KEDIYAVEIVGGATR	332 - 346	5.62E-09	3.398	0.5
EDIYAVEIVGGATR	333 - 346	1.04E-07	2.530	0.5
ELSTTLNADEAVTR	361 - 374	7.81E-05	2.674	0.5
EFSITDVVPYPISLR	391 - 405	8.68E-07	2.670	0.6
EPFTLEAYYSSPQDLPPYDPAIAQFSVQK	438 - 466	4.16E-14	3.993	0.4
VNVHGIFSVSSASLVEVHK	484 - 502	3.15E-03	2.315	0.2
TSTVDLPIENQLLWQIDR	574 - 591	1.18E-08	4.504	0.5
EMLNLYIENEGK	592 - 603	9.18E-05	3.678	0.3
NAVVEYVYEMR	619 - 629	9.48E-06	2.953	0.3
FVSEDDRNSFTLK	639 - 651	3.91E-03	2.341	0.2
LEDTENWLYEDGEDQPK	652 - 668	3.94E-11	3.938	0.4
STNEAMEWMNNK	737 - 748	2.34E-02	2.115	0.4
IKELTSTCSPISKPK	772 - 787	1.61E-03	4.221	0.5
NAEQNGPVDGQGDNPGPQAAEQGTDTAVPSDSK	799 - 832	1.63E-04	3.861	0.2
KLPEMDID	833 - 840	3.10E-02	1.768	0.2