

Figure 1: Brain MRI of Patients 2, 3 and 4 with the m.1644G>A mutation

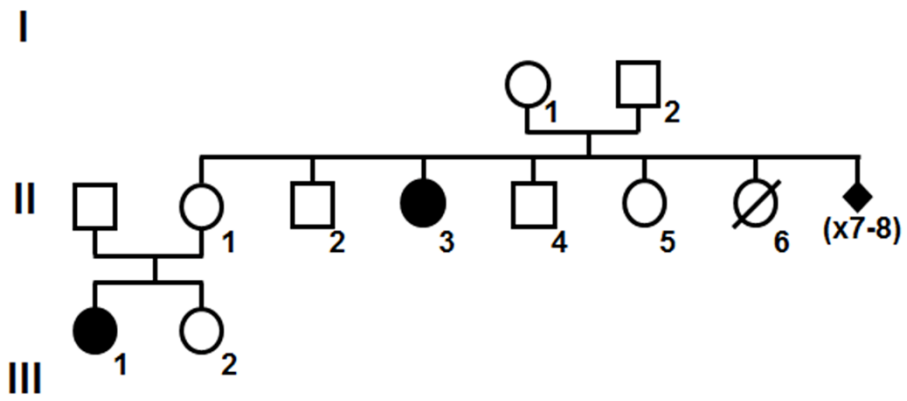


Figure 2: Pedigree of Family 3

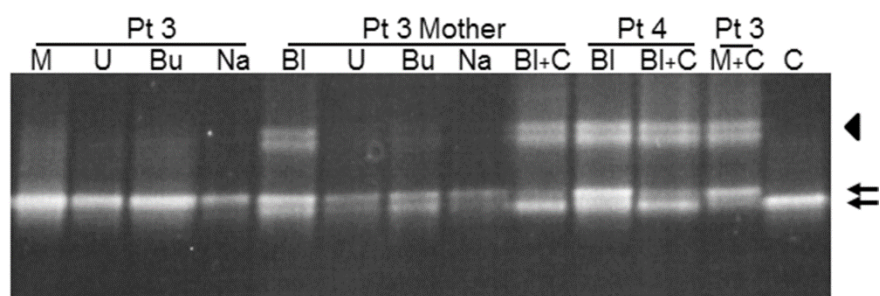
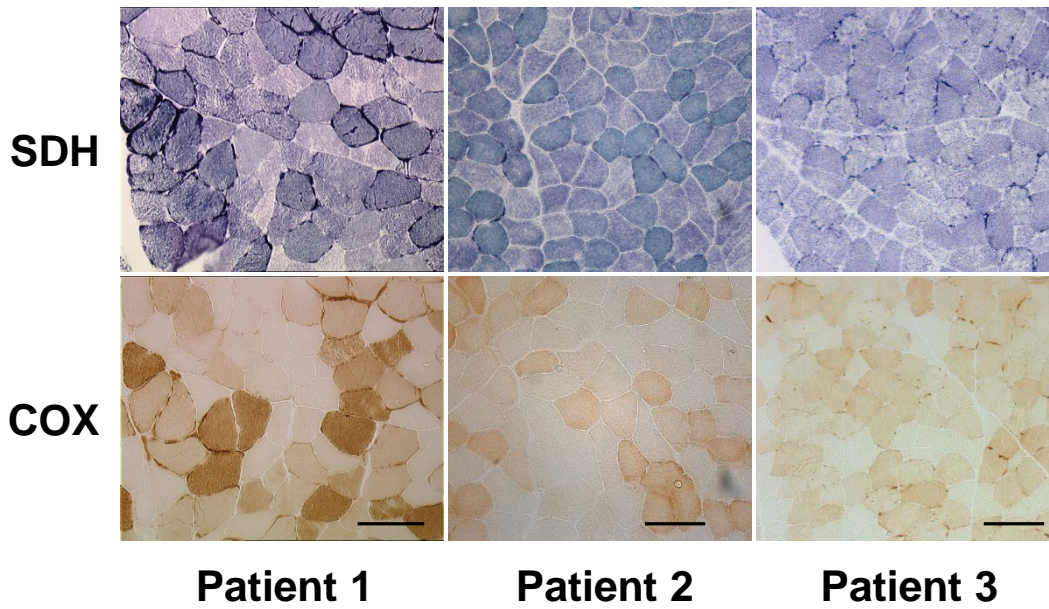


Figure 3: Denaturing gradient gel electrophoresis showing heteroplasmy of the m.1644G>A mutation



Supplemental Figure 1: Cytochrome c oxidase defect with a mosaic pattern coexisted with variable mitochondrial proliferation in Patients 1, 2 and 3 muscle