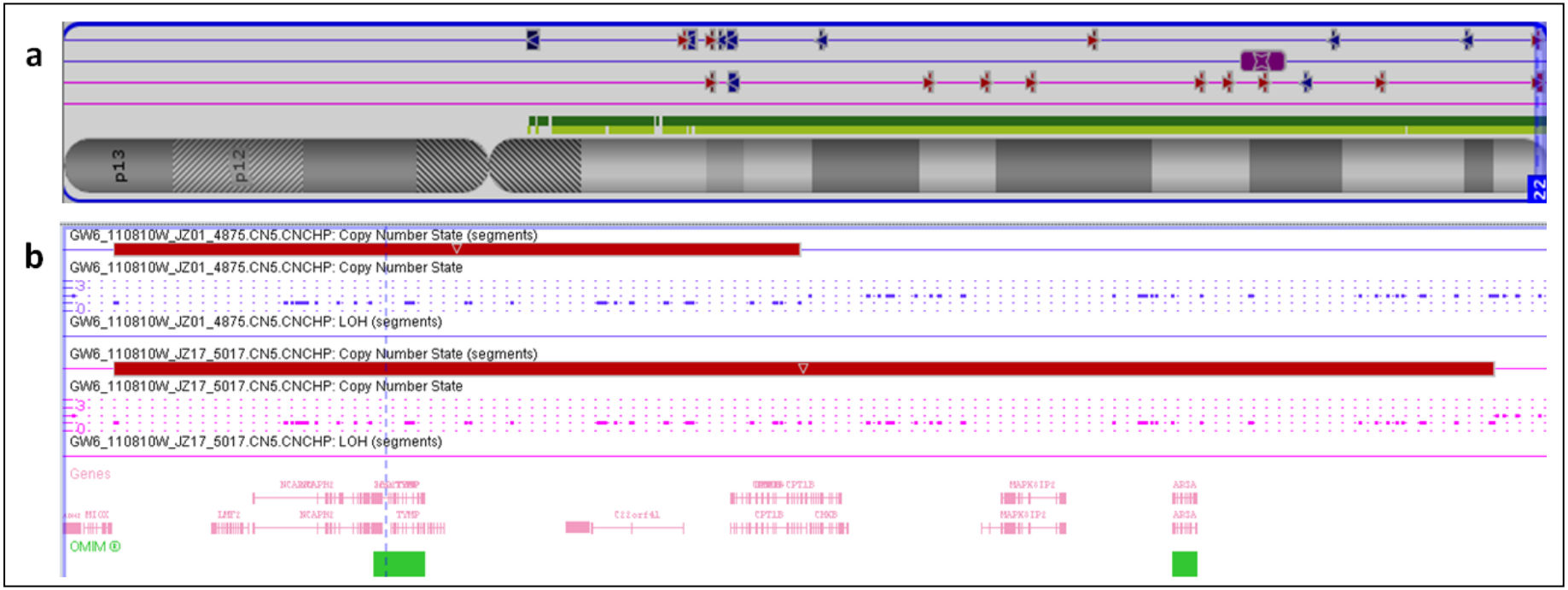
Vondráčková et al. Large copy number variations in combination with point mutations in the *TYMP* and *SCO2* genes found in two patients with mitochondrial disorders

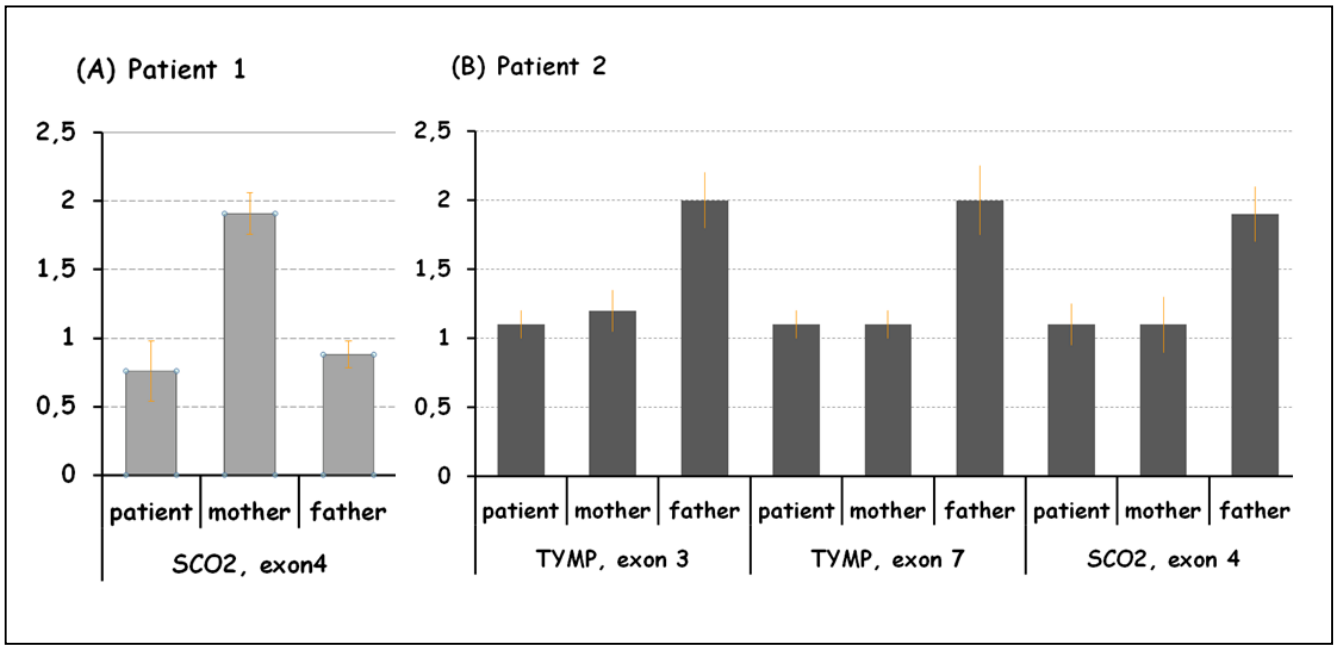
Supplementary Figures



Supplementary figure 1

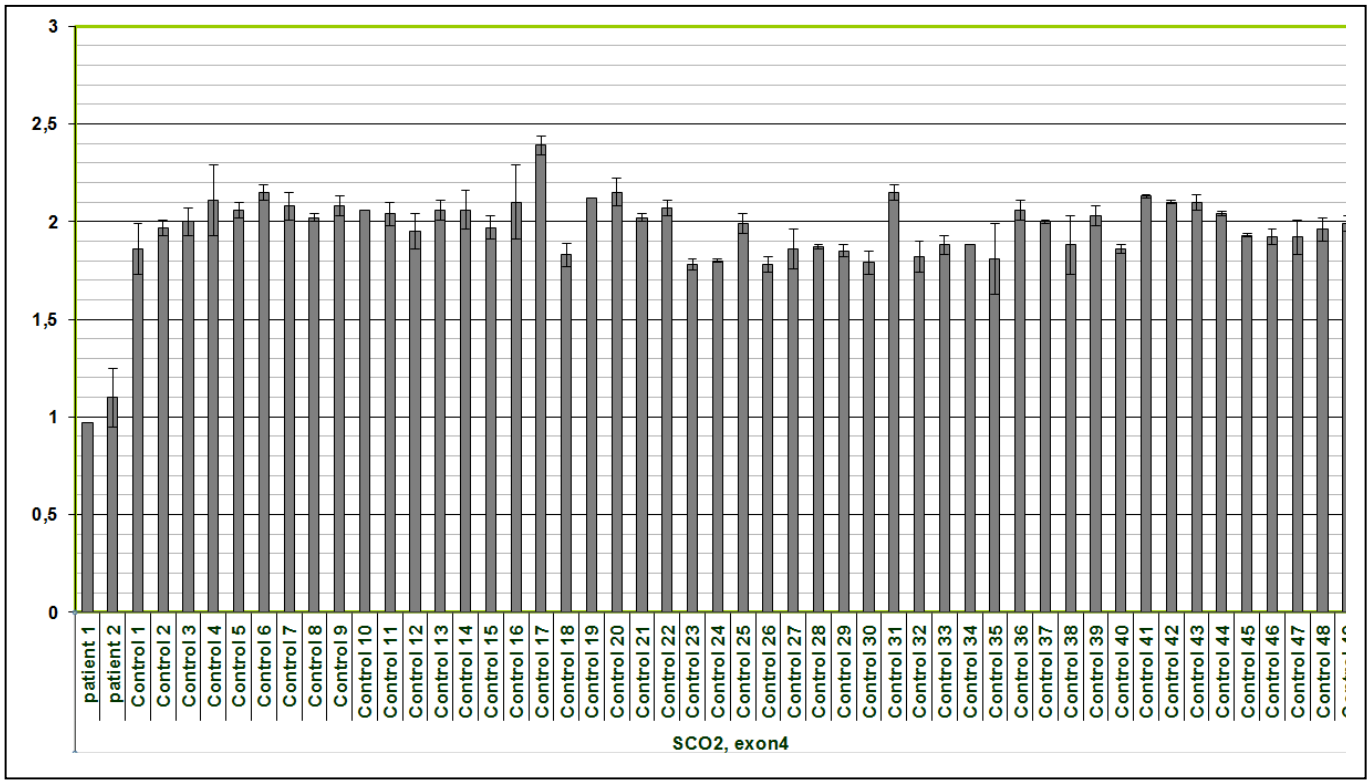
The output data of the Chromosome Analysis Suite (Affymetrix) of two patients with mitochondrial disease. (**a**) The blue region surrounds the causal large heterozygous deletions on chromosome 22q13.33 found in the patients. Blue arrows indicate losses of DNA, and red arrows indicate gains of DNA. (**b**) The thick red lines show large heterozygous deletions. In *SCO2* patient 1, the 87-kb deleted region covers eight genes: *LMF2*, *NCAPH2*, *SCO2*, *TYMP*, *ODF3B*, *KLHDC7B*, *c22orf41*, and *CPT1B* (violet dotted lines). In MNGIE patient 2, the 175-kb deleted region covers

twelve genes: *LMF2*, *NCAPH2*, *SCO2*, *TYMP*, *ODF3B*, *KLHDC7B*, *c22orf41*, *CPT1B*, *CHKB*, *LOC100144603*, *MAPKIP2*, and *ARSA* (pink dotted lines).



Supplementary figure 2

A real-time PCR copy-number variation assay was used to verify the absolute copy number status in two affected patients harbouring two different large heterozygous deletions: (A) in SCO2-deficient patient, (B) in MNGIE patient.



Supplementary figure 3

A real-time PCR copy-number variation assay of the *SCO2* gene was carried out in 50 control samples. With the exception of our two patients and/or their parents, no other deletions of the *SCO2* gene were detected in the Czech population.