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**The study of DNA alterations and Immunological Proteins Profile in patients with suspected Mitochondrial Disorders**

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**ABSTRACT**

Mitochondria contain their own DNA called mtDNA, that encodes four of the five respiratory chain (RC) complexes i.e. MTND1-6, MTND4L (Complex I), MTCYB (Complex III), MTCO1-3 (Complex IV), MTATP6 & MTATP8 (Complex V). Mutations of these genes caused oxidative phosphorylation (OXPHOS) system deficiencies which results in devastating mitochondrial diseases (MD) and cancers. The use of mtDNA as a potential biomarker for diagnosing and staging of human cancer and mitochondrial disorders has become fast hot spots of research. Our aim was to improve our diagnostic testing for patients with positive clinical symptoms of MD using a combination of DNA sequencing and immunological protein analyses (western-blot). Blood from 4 high-risk patients for MD were collected in EDTA tube, extracted and purified with Qiagen genomic DNA extraction kit followed by DNA sequencing. Skin from these patients was cultured for fibroblast, harvested and processed into lysates which contained 20-50µg of protein. These lysates were subjected to electrophoresis and immuno-detection of OXPHOS proteins against antibodies (ab) specific for each complexes (abCI, abCII, abCIII abCIV & abCV cocktail). Western-Blot for #Patient-1 revealed Complex-1 deficiency and homozygous mutation was detected at nucleotide 3380 (G to A) in MTND 1 gene. Western-Blot for #Patient-2 showed subtle Complex IV deficiency and sequencing showed 1 heterozygous mutation at c.574(C to T) p. (Arg192Trp) in exon 6 and a polymorphic variant at c.604 (G to C) p.(Asp202His) in exon 7, confirming SURF-1 carrier with CIV deficiency. #Patient-3 has normal western-blot but sequencing showed a hemizygous mutation at c>1132(C to T) p (Arg378Cys) in exon 11 confirming Pyruvate Dehydrogenase-I (PDHAI) deficiency. #Patient-4 has normal western-blot and no mutation was found in sequencing. The DNA alterations and protein immunological analyses improve our diagnostic testing for high-risk patients for mitochondrial disorders.