

Supplementary Material

Novel *NDUFA13* mutations associated with OXPHOS deficiency and Leigh syndrome: A second family report.

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Supplementary Table 1: Next generation sequencing “OXPHOS panel”. Structural subunits and assembly factor genes of OXPHOS system included:

<i>ACAD9</i>	<i>ATP5J</i>	<i>COA1</i>	<i>COX18</i>	<i>COX7A1</i>	<i>LYRM7</i>	<i>NDUFA8</i>	<i>NDUFB11</i>	<i>NDUFS2</i>	<i>PET112</i>	<i>TMEM126B</i>
<i>ATP5A1</i>	<i>ATP5J2</i>	<i>COA3</i>	<i>COX19</i>	<i>COX7A2</i>	<i>NDUFA1</i>	<i>NDUFA9</i>	<i>NDUFB2</i>	<i>NDUFS3</i>	<i>SCO1</i>	<i>TMEM70</i>
<i>ATP5B</i>	<i>ATP5L</i>	<i>COA4</i>	<i>COX20</i>	<i>COX7B</i>	<i>NDUFA10</i>	<i>NDUFAB1</i>	<i>NDUFB3</i>	<i>NDUFS4</i>	<i>SCO2</i>	<i>TTC19</i>
<i>ATP5C1</i>	<i>ATP5L2</i>	<i>COA5</i>	<i>COX4I1</i>	<i>COX7B2</i>	<i>NDUFA11</i>	<i>NDUFAB1</i>	<i>NDUFB4</i>	<i>NDUFS5</i>	<i>SDHA</i>	<i>UQCC1</i>
<i>ATP5D</i>	<i>ATP5O</i>	<i>COA6</i>	<i>COX4I2</i>	<i>COX7C</i>	<i>NDUFA12</i>	<i>NDUFAB2</i>	<i>NDUFB5</i>	<i>NDUFS6</i>	<i>SDHAF1</i>	<i>UQCC2</i>
<i>ATP5E</i>	<i>ATP5S</i>	<i>COA7</i>	<i>COX5A</i>	<i>COX8A</i>	<i>NDUFA13</i>	<i>NDUFAB3</i>	<i>NDUFB6</i>	<i>NDUFS7</i>	<i>SDHAF2</i>	<i>UQCR10</i>
<i>ATP5F1</i>	<i>ATPAF1</i>	<i>COX10</i>	<i>COX5B</i>	<i>COX8C</i>	<i>NDUFA2</i>	<i>NDUFAB4</i>	<i>NDUFB7</i>	<i>NDUFS8</i>	<i>SDHB</i>	<i>UQCR11</i>
<i>ATP5G1</i>	<i>ATPAF2</i>	<i>COX11</i>	<i>COX6A1</i>	<i>CYC1</i>	<i>NDUFA3</i>	<i>NDUFAB5</i>	<i>NDUFB8</i>	<i>NDUFV1</i>	<i>SDHC</i>	<i>UQCRB</i>
<i>ATP5G2</i>	<i>ATPIF1</i>	<i>COX14</i>	<i>COX6A2</i>	<i>ECSIT</i>	<i>NDUFA4</i>	<i>NDUFAB6</i>	<i>NDUFB9</i>	<i>NDUFV2</i>	<i>SDHD</i>	<i>UQCRC1</i>
<i>ATP5G3</i>	<i>BCS1L</i>	<i>COX15</i>	<i>COX6B1</i>	<i>FASTKD2</i>	<i>NDUFA5</i>	<i>NDUFAB7</i>	<i>NDUFC1</i>	<i>NDUFV3</i>	<i>SURF1</i>	<i>UQCRC2</i>
<i>ATP5H</i>	<i>CMC1</i>	<i>COX16</i>	<i>COX6B2</i>	<i>FOXRED1</i>	<i>NDUFA6</i>	<i>NDUFB1</i>	<i>NDUFC2</i>	<i>NUBPL</i>	<i>TACO1</i>	<i>UQCRFS1</i>
<i>ATP5I</i>	<i>CMC2</i>	<i>COX17</i>	<i>COX6C</i>	<i>LRPPRC</i>	<i>NDUFA7</i>	<i>NDUFB10</i>	<i>NDUFS1</i>	<i>OXA1L</i>	<i>TIMMDC1</i>	<i>UQCRH</i>
										<i>UQCRQ</i>

Selection of these genes was carried out by literature and databases search in November 2015.

Supplementary Table 2: Amplification PCR primers for exon 2 and exon 3 of the *NDUFA13* gene.

Exon	Foward Primer	Reverse Primer
2	TGTCACAGTCCAAGCAGCCT	CAACAGCGGAATGCTACAGG
3	GAAAGTAGAAAGCCAACAGTC	AACTCCTAGCTTTTCTCTTGC

Thermocycling program: I) 5 minutes at 95 °C II) 30 cycles: 30 seconds at 94 °C, 30 seconds at 58 °C and 40 seconds at 72 °C III) 10 minutes of 72 °C