

Centre de génétique moléculaire et chromosomique Pitié Salpêtrière
Diagnostic des maladies mitochondriales –Dr Claude Jardel (claude.jardel@aphp.fr)
Panel NGS MITOCAPTURE_V2_avril 2016

Gènes nucléaires				ADNmt (NC_012920)		
<i>AARS2</i>	NM_020745	<i>MFN2</i>	NM_014874	<i>SDHAF2</i>	NM_017841	<i>MT-TF</i>
<i>ACAD9</i>	NM_014049	<i>MPV17</i>	NM_002437	<i>SDHB</i>	NM_003000	<i>MT-RNR1</i>
<i>ADCK3</i>	NM_020247	<i>MRPS16</i>	NM_016065	<i>SDHC</i>	NM_003001	<i>MT-TV</i>
<i>ADCK4</i>	NM_024876	<i>MRPS22</i>	NM_020191	<i>SDHD</i>	NM_003002	<i>MT-RNR2</i>
<i>AFG3L2</i>	NM_006796	<i>MTFMT</i>	NM_139242	<i>SERAC1</i>	NM_032861	<i>MT-TL1</i>
<i>AGK</i>	NM_018238	<i>MTO1</i>	NM_001123226	<i>SLC19A3</i>	NM_025243	<i>MT-ND1</i>
<i>AIFM1</i>	NM_001130846	<i>MTPAP</i>	NM_018109	<i>SLC25A19</i>	NM_001126121	<i>MT-TI</i>
<i>ATP5A1</i>	NM_001001937	<i>NDUFA1</i>	NM_004541	<i>SLC25A20</i>	NM_000387	<i>MT-TQ</i>
<i>ATP5E</i>	NM_006886	<i>NDUFA10</i>	NM_004544	<i>SLC25A22</i>	NM_001191060	<i>MT-TM</i>
<i>ATPAF2</i>	NM_145691	<i>NDUFA11</i>	NM_001193375	<i>SLC25A3</i>	NM_005888	<i>MT-ND2</i>
<i>BCS1L</i>	NM_001257342	<i>NDUFA11</i>	NM_175614	<i>SLC25A4</i>	NM_001151	<i>MT-TW</i>
<i>BOLA3</i>	NM_212552	<i>NDUFA12</i>	NM_018838	<i>SLC25A46</i>	NM_138773	<i>MT-TA</i>
<i>C10ORF2</i>	NM_021830	<i>NDUFA2</i>	NM_002488	<i>SPG7</i>	NM_003119	<i>MT-TN</i>
<i>C20orf72</i>	NM_052865	<i>NDUFA9</i>	NM_005002	<i>SUCLA2</i>	NM_003850	<i>MT-TC</i>
<i>CARS2</i>	NM_024537	<i>NDUFAF1</i>	NM_016013	<i>SUCLG1</i>	NM_003849	<i>MT-TY</i>
<i>CHCHD10</i>	NM_213720	<i>NDUFAF2</i>	NM_174889	<i>SURF1</i>	NM_003172	<i>MT-CO1</i>
<i>CHKB</i>	NM_005198	<i>NDUFAF3</i>	NM_199069	<i>TACO1</i>	NM_016360	<i>MT-TS1</i>
<i>COA5</i>	NM_001008215	<i>NDUFAF4</i>	NM_014165	<i>TARS2</i>	NM_025150	<i>MT-TD</i>
<i>COA6</i>	NM_001206641	<i>NDUFAF5</i>	NM_024120	<i>TAZ</i>	NM_000116	<i>MT-CO2</i>
<i>COQ2</i>	NM_015697	<i>NDUFAF6</i>	NM_152416	<i>TIMM8A</i>	NM_004085	<i>MT-TK</i>
<i>COQ4</i>	NM_016035	<i>NDUFB3</i>	NM_002491	<i>TK2</i>	NM_004614	<i>MT-ATP8</i>
<i>COQ6</i>	NM_182476	<i>NDUFB9</i>	NM_005005	<i>TMEM240</i>	NM_001114748	<i>MT-ATP6</i>
<i>COQ9</i>	NM_020312	<i>NDUFS1</i>	NM_001199984	<i>TMEM70</i>	NM_017866	<i>MT-CO3</i>
<i>COX10</i>	NM_001303	<i>NDUFS2</i>	NM_004550	<i>TSFM</i>	NM_001172696	<i>MT-TG</i>
<i>COX14</i>	NM_001257133	<i>NDUFS3</i>	NM_004551	<i>TTC11</i>	NM_016068	<i>MT-ND3</i>
<i>COX15</i>	NM_078470	<i>NDUFS4</i>	NM_002495	<i>TTC19</i>	NM_017775	<i>MT-TR</i>
<i>COX20</i>	NM_198076	<i>NDUFS6</i>	NM_004553	<i>TUFM</i>	NM_003321	<i>MT-ND4L</i>
<i>COX4I2</i>	NM_032609	<i>NDUFS7</i>	NM_024407	<i>TYMP</i>	NM_001257989	<i>MT-ND4</i>
<i>COX6B1</i>	NM_001863	<i>NDUFS8</i>	NM_002496	<i>UQCC2</i>	NM_032340	<i>MT-TH</i>
<i>CYC1</i>	NM_001916	<i>NDUFV1</i>	NM_007103	<i>UQCC3</i>	NM_001085372	<i>MT-TS2</i>
<i>DARS2</i>	NM_018122	<i>NDUFV2</i>	NM_021074	<i>UQCRB</i>	NM_001254752	<i>MT-TL2</i>
<i>DGUOK</i>	NM_080916	<i>NUBPL</i>	NM_025152	<i>UQCRC2</i>	NM_003366	<i>MT-ND5</i>
<i>DNA2</i>	NM_001080449	<i>OPA1</i>	NM_130837	<i>UQCRQ</i>	NM_014402	<i>MT-ND6</i>
<i>DNM1L</i>	NM_012062	<i>PDHX</i>	NM_003477	<i>VARA2</i>	NM_001167734	<i>MT-TE</i>
<i>EARS2</i>	NM_001083614	<i>PDSS1</i>	NM_014317	<i>WARS2</i>	NM_015836	<i>MT-CYB</i>
<i>FBXL4</i>	NM_012160	<i>PDSS2</i>	NM_020381	<i>WFS1</i>	NM_006005	<i>MT-TT</i>
<i>FOXRED1</i>	NM_017547	<i>PET100</i>	NM_001171155	<i>YARS2</i>	NM_001040436	<i>MT-TP</i>
<i>HARS2</i>	NM_012208	<i>PNPT1</i>	NM_033109			
<i>IARS2</i>	NM_018060	<i>POLG</i>	NM_002693			
<i>IBA57</i>	NM_001010867	<i>POLG2</i>	NM_007215			
<i>ISCA1</i>	NM_030940	<i>PUS1</i>	NM_025215			
<i>ISCA2</i>	NM_194279	<i>QIL1</i>	NM_205767			
<i>ISCU</i>	NM_213595	<i>RARS2</i>	NM_020320			
<i>LARS</i>	NM_020117	<i>RRM2B</i>	NM_001172477			
<i>LARS2</i>	NM_015340	<i>SARS2</i>	NM_001145901			
<i>LRPPRC</i>	NM_133259	<i>SCO1</i>	NM_004589			
<i>LYRM4</i>	NM_001164840	<i>SCO2</i>	NM_005138			
<i>LYRM7</i>	NM_181705	<i>SDHA</i>	NM_004168			
<i>MARS2</i>	NM_138395	<i>SDHAF1</i>	NM_001042631			