

	Total population	Population according to age of onset		
	n=743	Before 12 mo n=356	1yr-16yr n=192	After 16yr n=180
<b>Men</b>	<b>395 (53.2)</b>	<b>179 (50.3)</b>	<b>113 (58.9)</b>	<b>92 (51.1)</b>
<b>Women</b>	<b>348 (46.8)</b>	<b>177 (49.7)</b>	<b>79 (41.1)</b>	<b>88 (48.9)</b>
Consanguinity	73 (9.8)	50 (14)	15 (7.8)	6 (3.3)
Affected maternal relatives	44 (5.9)	6 (1.7)	20 (10.4)	17 (9.4)
<b>CNS involvement</b>	<b>540 (72.7)</b>	<b>299 (84.0)</b>	<b>136 (70.8)</b>	<b>94 (52.2)</b>
Psychomotor retardation	268 (36.1)	198 (55.6)	58 (30.2)	6 (3.3)
Epilepsy/myoclony	239 (32.2)	153 (43.0)	55 (28.6)	27 (15.0)
Cerebellar ataxia	187 (25.2)	66 (18.5)	71 (37.0)	45 (25.0)
Encephalopathy	169 (22.7)	134 (37.6)	25 (13.0)	7 (3.9)
Movement disorders	129 (17.4)	82 (23.0)	31 (16.1)	12 (6.7)
Regression	109 (14.7)	72 (20.2)	29 (15.1)	6 (3.3)
Stroke-like episodes	48 (6.5)	10 (2.8)	18 (9.4)	20 (11.1)
Migraine-like headaches	32 (4.3)	5 (1.4)	13 (6.8)	14 (7.8)
Pyramidal symptoms	31 (4.2)	10 (2.8)	10 (5.2)	11 (6.1)
Cognitive impairment	29 (3.9)	0 (0.0)	12 (6.3)	17 (9.4)
Psychiatric disorders	15 (2.0)	5 (1.4)	2 (1.0)	8 (4.4)
Central respiratory depression	12 (1.6)	11 (3.1)	1 (0.5)	0 (0.0)
Cortical blindness	9 (1.2)	7 (2.0)	1 (0.5)	1 (0.6)
Bulbar involvement	9 (1.2)	5 (1.4)	0 (0.0)	4 (2.2)
Deterioration under valproate	6 (0.8)	3 (0.8)	2 (1.0)	1 (0.6)
Others...	10 (1.3)	6 (1.7)	0 (0.0)	4 (2.2)
<b>Muscular involvement</b>	<b>418 (56.3)</b>	<b>164 (46.1)</b>	<b>123 (64.1)</b>	<b>124 (68.9)</b>
Hypotonia	168 (22.6)	133 (37.4)	30 (15.6)	3 (1.7)
Myopathy	162 (21.8)	42 (11.8)	50 (26.0)	68 (37.8)
CPEO	137 (18.4)	26 (7.3)	52 (27.1)	58 (32.2)
Others...	121 (16.3)	13 (3.7)	48 (25.0)	57 (31.7)
<b>Growth failure</b>	<b>167 (22.5)</b>	<b>132 (37.1)</b>	<b>31 (16.1)</b>	<b>1 (0.6)</b>
Postnatal	136 (18.3)	105 (29.5)	27 (14.1)	1 (0.6)
Antenatal	52 (7.0)	43 (12.1)	8 (4.2)	0 (0.0)
<b>Ocular involvement</b>	<b>138 (18.6)</b>	<b>65 (18.3)</b>	<b>43 (22.4)</b>	<b>29 (16.1)</b>
Optic atrophy	52 (7.0)	21 (5.9)	22 (11.5)	9 (5.0)
Pigmentary retinopathy	35 (4.7)	20 (5.6)	10 (5.2)	4 (2.2)
Cataract	22 (3.0)	8 (2.2)	4 (2.1)	9 (5.0)
Others...	43 (5.8)	20 (5.6)	11 (5.7)	12 (6.7)
<b>Heart involvement</b>	<b>131 (17.6)</b>	<b>77 (21.6)</b>	<b>28 (14.6)</b>	<b>25 (13.9)</b>
Hypertrophic cardiomyopathy	58 (7.8)	39 (11.0)	11 (5.7)	8 (4.4)
Dilated cardiomyopathy	34 (4.6)	21 (5.9)	9 (4.7)	4 (2.2)
Restrictive cardiomyopathy	4 (0.5)	1 (0.3)	2 (1.0)	1 (0.6)
Others...	43 (5.8)	19 (5.3)	9 (4.7)	14 (7.8)
<b>Liver involvement</b>	<b>123 (16.6)</b>	<b>99 (27.8)</b>	<b>12 (6.3)</b>	<b>11 (6.1)</b>
<b>Deafness</b>	<b>113 (15.2)</b>	<b>37 (10.4)</b>	<b>40 (20.8)</b>	<b>32 (17.8)</b>
<b>Peripheral neuropathy</b>	<b>94 (12.7)</b>	<b>21 (5.9)</b>	<b>27 (14.1)</b>	<b>44 (24.4)</b>
<b>Digestive disorders</b>	<b>58 (7.8)</b>	<b>35 (9.8)</b>	<b>17 (8.9)</b>	<b>6 (3.3)</b>
Transit disorders	39 (5.2)	24 (6.7)	11 (5.7)	4 (2.2)
Others...	25 (3.3)	16 (4.5)	7 (3.6)	2 (1.1)
<b>Blood disorders</b>	<b>53 (7.1)</b>	<b>42 (11.8)</b>	<b>10 (5.2)</b>	<b>1 (0.6)</b>
Anemia	43 (5.8)	34 (9.6)	8 (4.2)	1 (0.6)
Neutropenia	10 (1.3)	9 (2.5)	1 (0.5)	0 (0.0)
Others...	9 (1.2)	8 (2.2)	1 (0.5)	0 (0.0)
<b>Renal involvement</b>	<b>59 (7.9)</b>	<b>41 (11.5)</b>	<b>13 (6.8)</b>	<b>3 (1.7)</b>
<b>Diabetes</b>	<b>45 (6.1)</b>	<b>9 (2.5)</b>	<b>14 (7.3)</b>	<b>21 (11.7)</b>
<b>Endocrine disorders</b>	<b>19 (2.6)</b>	<b>7 (2.0)</b>	<b>5 (2.6)</b>	<b>6 (3.3)</b>
<b>Malformative disorders</b>	<b>16 (2.2)</b>	<b>15 (4.2)</b>	<b>0 (0.0)</b>	<b>0 (0.0)</b>
<b>Microcephaly</b>	<b>11 (1.5)</b>	<b>11 (3.1)</b>	<b>0 (0.0)</b>	<b>0 (0.0)</b>
<b>Lipomas</b>	<b>5 (0.7)</b>	<b>0 (0.0)</b>	<b>2 (1.0)</b>	<b>3 (1.7)</b>
<b>Others...</b>	<b>25 (3.3)</b>	<b>12 (3.3)</b>	<b>3 (1.5)</b>	<b>10 (5.5)</b>

Sup. table 1 : Clinical characteristics of 743 patients suspected of manifesting a mitochondrial disorder