

	Total population	Population according to age of onset		
		Before 12 mo	1yr-16yr	After 16yr
<b>Brain MRI</b>	<b>n=527</b>	<b>n=290</b>	<b>n=132</b>	<b>n=96</b>
Leigh, Leigh-like syndrome	177 (33.6)	125 (43.1)	37 (28.0)	11 (11.5)
White matter involvement	156 (29.6)	81 (27.9)	30 (22.7)	42 (43.8)
Posterior fossa atrophy / hypoplasia	127 (24.1)	70 (24.1)	39 (29.5)	16 (16.7)
Cerebral atrophy	55 (10.4)	33 (11.4)	13 (9.8)	9 (9.4)
Leukodystrophy	33 (6.3)	17 (5.9)	11 (8.3)	5 (5.2)
Stroke, stroke-like	27 (5.1)	8 (2.8)	10 (7.6)	9 (9.4)
Thalamic lesions	25 (4.7)	22 (7.6)	1 (0.8)	1 (1.0)
Posterior cortical lesions	24 (4.6)	12 (4.1)	7 (5.3)	3 (3.1)
Corpus callosum lesions	17 (3.2)	15 (5.2)	0 (0.0)	2 (2.1)
Internal capsule lesions	16 (3.0)	9 (3.1)	2 (1.5)	5 (5.2)
Cerebellar hypersignal	10 (1.9)	6 (2.1)	3 (2.3)	1 (1.0)
Abnormalities of gyration	8 (1.5)	7 (2.4)	1 (0.8)	0 (0.0)
Calcifications	4 (0.8)	2 (0.7)	1 (0.8)	1 (1.0)
Others	28 (5.3)	15 (5.2)	5 (3.8)	8 (8.3)
Normal	126 (23.9)	52 (17.9)	43 (32.6)	29 (30.2)
<b>MRI spectroscopy</b>	<b>n=175</b>	<b>n=116</b>	<b>n=44</b>	<b>n=11</b>
Presence of lactate peak	106 (60.6)	79 (68.1)	22 (50.0)	2 (18.2)
Absence of lactate peak	69 (39.4)	37 (31.9)	22 (50.0)	9 (81.8)
<b>Metabolic screening</b>				
<b>Blood</b>				
<b>Standard test</b>	<b>n=525</b>	<b>n=309</b>	<b>n=144</b>	<b>n=66</b>
Hyperlactacidemia	266 (50.7)	168 (54.4)	65 (45.1)	30 (45.5)
Hypoglycaemia	27 (5.1)	20 (6.4)	3 (2.1)	3 (4.5)
<b>Lactate/Pyruvate</b>	<b>n=385</b>	<b>n=232</b>	<b>n=105</b>	<b>n=43</b>
Elevated	193 (50.1)	121 (52.2)	47 (44.8)	24 (55.8)
Normal	192 (49.9)	111 (47.8)	58 (55.2)	19 (44.2)
<b>OH-but/Ac-Ac</b>	<b>n=244</b>	<b>n=158</b>	<b>n=64</b>	<b>n=18</b>
Elevated	66 (27.0)	43 (27.2)	19 (29.7)	4 (22.2)
Normal	178 (73.0)	115 (72.8)	45 (70.3)	14 (77.8)
<b>Plasma amino acids</b>	<b>n=300</b>	<b>n=196</b>	<b>n=77</b>	<b>n=23</b>
Elevated alanine and/or proline	26 (8.7)	11 (5.6)	12 (15.6)	3 (13.0)
Other abnormalities	9 (3.0)	4 (2.0)	4 (5.2)	1 (4.4)
Normal	265 (88.3)	181 (92.4)	61 (79.2)	19 (82.6)
<b>Plasma carnitine/acylcarnitine</b>	<b>n=208</b>	<b>n=131</b>	<b>n=58</b>	<b>n=15</b>
Decreased	10 (4.8)	6 (4.6)	4 (6.9)	0 (0.0)
Elevated	3 (1.4)	3 (2.3)	0 (0.0)	0 (0.0)
Normal	195 (93.8)	122 (93.1)	54 (93.1)	15 (100)
<b>CSF lactate</b>	<b>n=146</b>	<b>n=98</b>	<b>n=36</b>	<b>n=11</b>
Hyperlactatorachia	110 (75.3)	75 (76.5)	30 (83.3)	5 (45.5)
Normal	36 (24.7)	23 (23.5)	6 (16.7)	6 (54.5)
<b>Urinary organic acids</b>	<b>n=360</b>	<b>n=237</b>	<b>n=93</b>	<b>n=24</b>
Elevated Krebs cycle intermed.	15 (4.2)	9 (3.8)	5 (5.4)	1 (4.2)
Elevated Urinary 3-MGC acid	9 (2.5)	7 (3.0)	2 (2.2)	0 (0.0)
Other abnormalities	7 (1.9)	5 (2.1)	1 (1.1)	1 (4.2)
Normal	329 (91.4)	216 (91.1)	85 (91.4)	22 (91.7)

Sup. table 2 : Paraclinical investigations