

	Total population	Population according to age of onset		
		Before 12 mo	1yr-16yr	After 16yr
Muscle histology	n=637	n=292	n=161	n=174
Lipid myopathy	191 (30.0)	100 (34.2)	46 (28.6)	42 (24.1)
RRF	184 (28.9)	42 (14.4)	54 (33.5)	85 (48.9)
COX-negative fibers	176 (27.6)	31 (10.6)	43 (26.7)	99 (56.9)
Minimal abnormalities	29 (4.6)	12 (4.1)	10 (6.2)	7 (4.0)
Normal	273 (42.9)	152 (52.1)	68 (42.2)	49 (28.2)
Spectrophotometry analysis				
Muscle	n=653	n=331	n=165	n=144
Isolated deficiency	204 (31.2)	94 (28.4)	63 (38.2)	45 (31.3)
Multiple deficiency	184 (28.2)	113 (34.1)	34 (20.6)	33 (22.9)
Normal	265 (40.6)	124 (37.5)	68 (41.2)	66 (45.8)
Fibroblasts	n=238	n=153	n=58	n=20
Isolated deficiency	68 (28.6)	37 (24.2)	24 (41.4)	7 (35.0)
Multiple deficiency	31 (13.0)	17 (11.1)	6 (10.3)	8 (40.0)
Normal	139 (58.4)	99 (64.7)	28 (48.3)	5 (25.0)
Liver	n=122	n=102	n=15	n=2
Isolated deficiency	38 (31.1)	32 (31.4)	6 (40.0)	0 (0.0)
Multiple deficiency	32 (26.3)	23 (22.5)	6 (40.0)	2 (100.0)
Normal	52 (42.6)	47 (46.1)	3 (20.0)	0 (0.0)
Heart	n=10	n=6	n=4	n=0
Isolated deficiency	3 (30.0)	2 (33.3)	1 (25.0)	-
Multiple deficiency	3 (30.0)	2 (33.3)	1 (25.0)	-
Normal	4 (40.0)	2 (33.3)	2 (50.0)	-
Kidney	n=4	n=3	n=0	n=0
Isolated deficiency	1 (25.0)	1 (33.3)	-	-
Multiple deficiency	2 (50.0)	1 (33.3)	-	-
Normal	1 (25.0)	1 (33.3)	-	-
Lymphocytes	n=3	n=1	n=2	n=0
Isolated deficiency	1 (33.3)	1 (100.0)	0 (0.0)	-
Multiple deficiency	2 (66.7)	0 (0.0)	2 (100.0)	-
Normal	0 (0.0)	0 (0.0)	0 (0.0)	-

Sup. table 3 : Histological and biochemical analyses of tissue samples