

Supplementary Table 3

Overview of patients with multiple heteroplasmic mutations in the coding region and the D-loop

Patient	Mutations	Gender	Age at biopsy	Phenotype*
p1	del8809–13035 / T16297C	Female	34	CPEO
p2	del12103–14411 / del71	Male	60	CPEO
p3	A3243G / A185G / A189G	Female	48	MELAS
p4	A3243G / A185G / A189G	Male	36	CPEO
p5	A3243G / T72C	Female	42	MELAS
p6	G5835A / T72C	Male	22	CPEO
p7	del10311–15571 / T72C	Male	53	CPEO
p8	del9098–14651 / T72C	Male	48	CPEO
p9	del10198–13755 / T72C	Male	44	CPEO
p10	del11032–13232 / T72C	Female	78	CPEO

* CPEO, chronic progressive external ophthalmoplegia; MELAS, mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes