Supplement Table 1: Mitochondrial disease diagnosis by type reported by MDCR survey participants		
Diagnosis Type	N[a]	n[b](%)
Other	172	
Definite	172	123 (71.51)
Probable	172	43 (25.00)
Possible	172	6 (3.49)
MELAS (Mitochondrial Encephalopathy Lactic Acidosis with Stroke-like Episodes)	52	
Definite	52	34 (65.38)
Probable	52	16 (30.77)
Possible	52	2 (3.85)
CPEO (Chronic Progressive External Ophthalmoplegia)	46	
Definite	46	41 (89.13)
Possible	46	3 (6.52)
Probable	46	2 (4.35)
Complex I Deficiency	39	
Definite	39	29 (74.36)
Probable	39	8 (20.51)
Possible	39	2 (5.13)
CoQ Deficiency	38	
Definite	38	22 (57.89)
Probable	38	13 (34.21)
Possible	38	3 (7.89)
KSS (Kearns-Sayre Syndrome)	30	
Definite	30	26 (86.67)
Possible	30	2 (6.67)
Probable	30	2 (6.67)
Mitochondrial DNA Depletion Syndrome	29	
Definite	29	20 (68.97)
Probable	29	5 (17.24)
Possible	29	4 (13.79)

Complex IV Deficiency	27	
Definite	27	19 (70.37)
Probable	27	8 (29.63)
Complex III Deficiency	25	
Definite	25	20 (80.00)
Probable	25	4 (16.00)
Possible	25	1 (4.00)
Encephalopathy		
Definite	24	11 (45.83)
Probable	24	9 (37.50)
Possible	24	4 (16.67)
Multiple Respiratory Chain Enzyme Deficiencies	21	
Definite	21	14 (66.67)
Probable	21	6 (28.57)
Possible	21	1 (4.76)
MNGIE (Mitochondrial Neurogastrointestinal Encephalomyopathy)	19	
Definite	19	10 (52.63)
Possible	19	5 (26.32)
Probable	19	4 (21.05)
LHON (Leber Hereditary Optic Neuropathy)	18	
Definite	18	15 (83.33)
Probable	18	3 (16.67)
MERRF (Myoclonus Epilepsy Ragged-red Fibers)	14	
Definite	14	7 (50.00)
Probable	14	6 (42.86)
Possible	14	1 (7.14)
Encephalomyopathy	12	
Definite	12	6 (50.00)
Probable	12	5 (41.67)

Possible	12	1 (8.33)
MILS (Maternally Inherited Leigh Syndrome)	12	
Definite	12	9 (75.00)
Probable	12	2 (16.67)
Possible	12	1 (8.33)
NARP (Neuropathy, Ataxia and Retinitis Pigmentosa)	12	
Probable	12	6 (50.00)
Definite	12	5 (41.67)
Possible	12	1 (8.33)
Sensory Ataxia Neuropathy	12	
Definite	12	10 (83.33)
Probable	12	2 (16.67)
Complex V Deficiency	7	
Definite	7	4 (57.14)
Probable	7	3 (42.86)
SANDO (Sensory Ataxia, Neuropathy, Dysarthria, Ophthalmoplegia)	7	
Definite	7	6 (85.71)
Probable	7	1 (14.29)
Complex II (SDH) Deficiency	6	
Definite	6	5 (83.33)
Probable	6	1 (16.67)
LHON-Plus (Leber Heredity Optic Neuropathy Plus)	6	
Definite	6	3 (50.00)
Probable	6	3 (50.00)
Alpers syndrome	5	
Definite	5	4 (80.00)
Possible	5	1 (20.00)
MIDD (Maternally Inherited Diabetes and Deafness)	5	

Definite	5	4 (80.00)
Possible	5	1 (20.00)
Dysarthria Ophthalmoplegia	4	
Definite	4	3 (75.00)
Probable	4	1 (25.00)
Pearson syndrome	4	
Definite	4	2 (50.00)
Possible	4	1 (25.00)
Unlikely	4	1 (25.00)
FBSN (Familial Bilateral Striatal Necrosis)	1	
Definite	1	1 (100.00)
Hepatocerebral disease	1	
Definite	1	1 (100.00)
Leukoencephalopathy	1	
Definite	1	1 (100.00)

Note: Responses to ten multiple choice survey questions, where multiple responses could be selected, where used for this table. Additional responses, where diagnosis was specified in free text field, were not considered.
a. N = number of subjects with a specific diagnosis. This value is used as the denominator for the percentage calculations.
b. n = Number of subjects with the specified characteristic.