Table 1- Online version only

Table 1: Method of diagnosis of each patient according to modified Walker Criteria (Mod Walk Criteria III by Bernier 2002 (8).

Sub No.	Clinical Cytopathy (multi-system disorder)	Syndromic Presentation (MELAS, Leigh, etc.)	>2% subsarcolemmal mitochondrial aggregates (muscle)	Widespread EM abnormalities (any tissue)	>2% COX-neg fibers (muscle)	<20% RC activity (any tissue)	20-30% RC activity (any tissue)	Probable pathogenic DNA mutation	Metabolic indicators of impaired RC function	TOTAL MAJOR	TOTAL MINOR	M Walk (DEF OR PROB)**
	Maj	Maj	Min	Min	Maj	Maj	Min	Min	Min			
1	X							X	X	1	2	Def
2	X							X	X	1	2	Def
3	X		X	X				X	X	1	4	Def
4	X								X	1	1	Prb
5	X							X	X	1	2	Def
6	X								X	1	1	Prb
7	X								X	1	1	Prb
8	X				X				X	1	2	Def
9	X		X						X	2	1	Def
10	X		X				X		X	1	3	Def
11	X		X						X	1	2	Def
12		X							X	1	1	Prb
13	X					X			X	1	2	Def
14	X					X			X	1	2	Def
15	X								X	1	1	Prb
16	X			X					X	1	2	Def
17		X	X						X	1	2	Def
18	X		X	X			X			1	3	Def
19	X		X	X						1	2	Def
20	X			X				X		1	2	Def
21	X							X	X	1	2	Def
22				X		X			X	1	2	Def
23		X							X	1	1	Prb
24	X							X	X	1	2	Def
25	X					X			X	2	1	Def
26	X							X	X	1	2	Def

The patients are classified as having definite (Def) or probable (prb) mitochondrial disorder.

The subclasses used for diagnosis are listed in the table.

Definite criteria is defined by identifying 2 major or 1 major and 2 minor criteria while probable mitochondrial disorder is classified by identifying 1 major with 1 minor or at least 3 minor criteria.

** None of subjects belonged in the category of: >2% of ragged-red fibers (Major), antibody-based RC defect (Minor), <30% (Major) or <40% (Minor) RC activity in 2 tissues, and undisputed pathogenic DNA mutation (Major), therefore these 5 markers are not shown in the table.

Sub= subjects, RC= respiratory chain, EM= electron microscopic, DNA= Deoxyribo nucleic acid

Table 2- Online version only

Table 2: Descriptive characteristics of the study cohort

Sub	Age (yrs)	Sex	Mito Disease	Signs and Symptoms	GI Symptoms	Ht, Wt, BMI (%)	GE Time	LP	ITT	Repeat study	Type of Prokinetic	Repeat GE time	Repeat ITT
1	13	F	Def	Hypotonia, HA, ptosis,myopathy,	AP,Reflux	60, 96, 96	87	30	N	Y	Bet	80, LP16	6
2	12	F	Def	Hypotonia, neuropathy	Reflux	49, 76, 72	50	10	N				
3	5	M	Def	Hypoton, fatigue, EI	Reflux const	17, 35, 68	126	10	N				
4	6	F	Prb	Hypotonia, motor delay	AP, const. on Lactul	36, 97, 99	48	9.5	N				
5	7	M	Def	Fatig, EI.,	AP, on Met	3, 22, 74	229	70	N				
6	4	F	Prb	DD	V,Gag, FI	21, 6, 8	60	11	N				
7	5	F	Prb	Hypotonia, OSA	Reflux, Diarrhea	13, 61, 93	164	45	4	Y	Met	128, LP 40	4
8	13	F	Def	Migraine HA, Ocular weakness,	AP, Reflux , IBS	96, 99, 99	167	50	6	Y	Met	60, LP 17	4
9	8	M	Def	Hypotonia, Sz	AP, Reflux	92, 99, 99	80	42	N	Y	Bet, Mom refused Met	110, LP Unk	N
10	5	M	Def	Sz, Hypotonia	Reflux,const	1, 1, 44	59	none	N				
11	16	M	Def	Prog spasticity in wheelchair, ataxia, nystagmus,dysarthia	const, V,Asp thin Liq on MBS	1, 46,83	345	none	6	Y	Met	6 hrs	6
12	16	M	Prb	Migraine, pancreatitis in past	const., AP	67,37,30	107	18	4				
13	3	M	Def	DD, Hypotonia	Vomt, Reflux	5, 89,99	45	none					
14	3	M	Def	Hypotonia, cardiomyopathy, ptosis	const, AP,	42,5,1	147	29	6				
15	14	M	Prb	DD, PDD, Hypotonia, epilepsy, myoclonus	Chronic diarrhea	3,1,1	124	unk	6		none, only diarrhea		
16	5	M	Def	DD,PDD, ADHD, hypotonia	AP	16,16,35	69	unk	4				
17	10	M	Def	Bilat BG, Hyptonia, dystonia, SD	AP	29,86,95	66	13	6				
18	4	M	Def	Cortical visual impairment, DD, SD, myoclonus,chorea, dystonia	AP, Reflux, const, on Nex and Mir	1,39,96	107	unk	6	Y	Beth , Met CI	177, LP Unk	
19	4	M	Def	EI, muscle weakness, DD, infantile cardiac arrhythmias	AP, Reflux	99,92,57	67	unk	N				
20	11	M	Def	hypotonia, EI , ADHA	Const, BM 1/wk.	26,21,30	99	unk	4	Y	Met	70, LP Unk	N
21	7	. M	Def	ADHD, hypotonia, elevated transaminases	AP	21,43,67	85	unk	N				
22	4	M	Def	PDD, Cortical visual impairment, Sz,	PO int, G tube in px	86,59,21	70	30	N				
23	6	M	Prb	Ptosis, visual impairment, proximal myopathy	N, V, Const, FTT	50,1,1	99	30	N	Y	Met	199, LP unk	N
24	3	M	Def	LD	Reflux on Beth & Lans	27,42,58	148	40	N				
25	3	M	Def	Hypotonia, autonomic dysfunction, elevated transminases in past	AP, Reflux, early fullness	8,14,6	196	unk	6	Y		200	N
26	14	M	Def	Ex intol, fatig, ptosis, OSA	Reflux on Met	99,99,99	195	24	6				

Table 2 footnotes:

Abbreviations: S and S = signs and symptoms, Ht = height, Wt = weight, BMI = body mass index, GET = gastric emptying time (time taken for half of the stomach to empty post ingestion of the radiolabeled meal), ITT = small intestinal transit times (time taken for the radioisotope to be seen in the cecum post ingestion), M = male, F = female, N = normal, LP = lag phase (time taken for the stomach to start to empty post ingestion of the radioisotope containing meal), Met = metoclopromide, Eryth = erythromycin, Beth = bethanechol, Lan = lansoprozole, Px = proximal, Dx = distal, G tube = gastrostomy tube, Const = constipation, BM = bowel movement, DD = development delay, FTT = failure to thrive, Y = yes, Def = definite, Prb = prob, HA = headache, EI = exercise intolerance, OSA = obstructive sleep apnea, Sz = seizure, PDD = pervasive developmental delay, ADHD = attention deficit hyperactive disorder, LD = learning disability, SD = speech delay, OT = occupational therapy, PT = physical therapy, prog = progressive, Unk = unknown.

All subjects consumed solid radiolabeled meal except subject #6 (consumed semisolid meal). GE time of 60 minutes is prolonged in this subject (normal for semisolid diet is < 60 minutes). Subject 5 and 26 were on metoclopramide from before. Subject 24 was on betanechol for reflux. These prokinetic medications were stopped 48h before study and then restarted. Scintigraphy was not repeated in these subjects. Annotations in numbers underlined signify deviation from the standard reference range (abnormal).