

Supplemental table 2 Major clinical features at onset and later during disease course			
Clinical feature	Number of patients with feature *	Number of patients with feature at onset **	Number of patients having developed feature after onset ***
Ptosis	70/79 (88.6%)	38/63 (60.3%)	16/25 (64.0%)
PEO	66/78 (84.6%)	25/57 (43.9%)	20/32 (62.5%)
Limb weakness	41/74 (55.4%)	6/58 (10.3%)	19/52 (36.5%)
Pigmentary retinopathy	37/73 (50.7%)	4/58 (6.9%)	18/54 (33.3%)
Hearing impairment	31/75 (41.3%)	4/68 (5.9%)	20/64 (31.3%)
Visual impairment	31/66 (47.0%)	2/54 (3.7%)	17/52 (32.7%)
Fatigue	26/69 (37.7%)	9/60 (15.0%)	8/51 (15.7%)
Cardiac conduction block	25/75 (33.3%)	0/66 (0.0%)	16/66 (24.2%)
Cerebellar ataxia	25/73 (34.2%)	3/68 (4.4%)	17/65 (26.2%)
Muscular atrophy	21/71 (29.6%)	2/60 (3.3%)	8/58 (13.8%)
Anemia	16/75 (21.3%)	13/75 (17.3%)	3/62 (4.8%)
Strabismus	16/69 (23.2%)	5/63 (7.9%)	5/58 (8.6%)
Renal disorder	15/70 (21.4%)	2/69 (2.9%)	12/67 (17.9%)
Diabetes mellitus, type 1	14/77 (18.2%)	2/77 (2.6%)	12/75 (16.0%)

* Number before slash is patients with the feature at some time during disease course, number after slash is patients with known data for the feature.

** Number before slash is patients with the feature as part of disease onset, number after slash is patients with known data for the feature at disease onset.

*** Number before slash is patients developing the feature during disease course, after onset, number after slash is patients known to be susceptible to develop the feature during disease course, after onset.

The reason the numbers do not add up between the columns is that in retrospect it is not known for all patients whether the feature was present at disease onset or developed later during disease course.