

Supplementary Table e-1: Results of Congenital Myasthenic Syndrome rating scale performed in patients with *SYT2* mutation

<i>Examined Parameters / Scores</i>		<i>Patient II.2 (44 y female)</i>			<i>Patient III.2 (27y female)</i>		
		Initial Visit	1 month on 3,4 DAP	2 weeks NO 3,4 DAP	Initial Visit	1 month on 3,4 DAP	2 weeks NO 3,4 DAP
Swallowing ability / 0-9 (Texture of food (0-3), Meal time (0-3), Swallowing/choking (0-3))		0/9	0/9	0/9	0/9	0/9	0/9
Respiratory function / Forced Vital Capacity (FVC)	Sitting FVC / Predicted %	3.87 / 112%	3.78 / 113%	3.63/ 109%	3.64 / 99%	not performed	3.60 / 98%
	Supine FVC / Predicted %	not performed	not performed	3.16/ 95%	not performed	not performed	3.07/ 83%
Oculomotor weakness	Double vision R/L eye / Lateral gaze 1 minute test	1 min/ 1 min	1 min/ 1 min	1 min/58sec	28 sec/ 26 sec	1 min/ 1 min	12.3 sec/ 15.8 sec
	Ptosis / Upward gaze 1 minute test	1 min	1 min	1 min	45 sec increased blinking	1 min	1 min
	Eyelid closure / 0-3	0	0	0	0	0	0
Bulbar motor weakness	Oro-facial motor function/ Timed slurp test	6:30 sec	5:39 sec	6:10 sec	11:36 sec	7:20 sec	9:87 sec
	Speech weakness/ Count up to 50	no change	no change	no change	no change	no change	no change
General and limb muscle weakness	Rise from floor / time	unable	9:09 sec with chair support	7:51 sec with chair support	unable	unable	5:47 sec
	Sustained arm raise / 4 minute abduction at 90°	4 min	4 min	4 min	4 min	not performed	4 min
	Sustained head raise /2 minute supine head flexion at 45°	43 sec	2 min	1:38 min	1:30 min	2 min	1:58 min
	Sustained leg raise / 1:40 minute supine hip-knee flexion at 45°	1:40 min	1:40 min	1:40 min	1:34 min	1:40 min	1:38 min
	Squats / number in 1 minute	unable	unable	unable	unable	unable	unable
	Repeated sit to stand / number in 1 minute	unable	9	13	unable	not performed	16
	Repeated rise from floor / time	unable	9:69 sec with chair support	10:62 with chair support	unable	5:00 sec with hand support	5:45 sec