

Congenital Myasthenic Syndrome Natural History Project

Demographics

▪ Date of birth: __/__/____ Male Female

▪ Genetic mutation:

- | | | |
|---|--------------------------------|-----------------------------|
| <input type="checkbox"/> RAPSYN | <input type="checkbox"/> ChAT | |
| <input type="checkbox"/> DOK-7 | <input type="checkbox"/> AGRIN | |
| <input type="checkbox"/> COLQ | | |
| <input type="checkbox"/> AChR Deficiency: | CHRNA1 | CHRNA1 CHRNA1 CHRNA1 CHRNA1 |
| <input type="checkbox"/> Fast Channel: | CHRNB1 | CHRNB1 CHRNB1 CHRNB1 CHRNB1 |
| <input type="checkbox"/> Slow Channel: | CHRND | CHRND CHRND CHRND CHRND |
| <input type="checkbox"/> Glycosylation: | CHRNE | CHRNE CHRNE CHRNE CHRNE |
| <input type="checkbox"/> Other | GFPT1 | DPAGT1 GMPPB ALG2 ALG14 |

▪ Age of CMS symptom onset:

▪ Age at CMS diagnosis:

▪ Age CMS treatment initiated:

▪ Current CMS treatments (doses):

- Pyridostigmine
- 3,4-Diaminopyridine
- Salbutamol
- Ephedrine
- Fluoxetine
- Quinidine

▪ Is patient on any respiratory support? Yes No

If yes, please tick most appropriate:

NIV	<input type="checkbox"/> < 16 hours/day	<input type="checkbox"/> > 16 hours/day
Ventilated via tracheostomy	<input type="checkbox"/> < 16 hours/day	<input type="checkbox"/> >16 hours/day

Quantitative Myasthenia Gravis Test

Time of Assessment: __: __

To be completed at all ages as able for the patient.

	TEST ITEMS:	NONE	MILD	MODERATE	SEVERE	RAW SCORE	SCORE
	GRADE	0	1	2	3		
1	Diplopia	>60	11-59	1-10	Spontaneous 0		
2	Ptosis (s)	>60	11-59	1-10	Spontaneous 0		
3	Facial muscles (Eyelid closure)	Normal lid closure	Complete, Weak, Some resistance	Complete, without resistance	Incomplete eyelid closure		
4	Swallowing (question)	Normal	Rare episode of choking or trouble swallowing	Frequent trouble swallowing, necessitating change in diet	Unable to swallow / Gastric tube		
5	Counting 1-50 Dysarthria / nasal speech	None at #50	Dysarthria at #30-49	Dysarthria at #10-29	Dysarthria at or before #9		
6	R arm 90° (s) Test both arms together	>240	90-239	10-89	0-9		
7	L arm 90° (s) Test both arms together	>240	90-239	10-89	0-9		
8	Vital Capacity Best of 3 trials	M > 3.5 F > 2.5	M 2.6-3.5 F 1.9-2.5	M 1.5-2.5 F 1.2-1.8	M <1.5 F <1.2	L %P	
9	R Grip (Kg) Support dynamometer	M > 45 F > 30	M 15-44 F 10-29	M 5-14 F 5-9	M 0-4 F 0-4		
10	L Grip (Kg) Support dynamometer	M > 35 F > 25	M 15-34 F 10-24	M 5-14 F 5-9	M 0-4 F 0-4		
11	Head lift 45° (s)	>120	30-119	0-29	Unable 0		
12	R Leg 45° (s) Test separately	>100	31-99	1-30	Unable 0		
13	L Leg 45° (s) Test separately	>100	31-99	1-30	Unable 0		

Total QMG Score: / 39

Comments: (e.g., if patient gave up / pain limiting)

MG-ADL

Time of Assessment: __: __

To be completed at all ages - patient or parent for young child

Rate	0	1	2	3	Score (0,1,2 or 3)
1. Talking	Normal	Intermittent slurring or nasal speech	Constant slurring or nasal speech, but can be understood	Difficult to understand speech	
2. Chewing	Normal	Fatigue with solid food	Fatigue with soft food	Gastric tube	
3. Swallowing	Normal	Rare episode of choking	Frequent choking necessitating changes in diet	Gastric tube	
4. Breathing	Normal	Shortness of breath with exercise	Shortness of breath at rest	Ventilator dependence	
5. Impairment of ability to brush teeth or comb hair	None	Extra effort, but no rest periods needed	Rest periods needed	Cannot do one of these functions	
6. Impairment of ability to arise from a chair	None	Mild, sometimes uses arms	Moderate, always uses arms	Severe, requires assistance	
7. Double vision	None	Occurs, but not daily	Daily, but not constant	Constant	
8. Eyelid droop	None	Occurs, but not daily	Daily, but not constant	Constant	
					MG-ADL score total (items 1-8) =

Repeated Sit-to-Stand in One Minute

To be completed at all ages - based on child’s developmental ability

Time	10 sec	20 sec	30 sec	40 sec	50 sec	60 sec
Total No. STS						
Interval difference						

Total number of STS in one minute:

For children < 2 years

Rate	0	1	2	3	Score (0,1,2 or 3)
Sitting balance	Able to sit independently for more than 5 minutes	Able to achieve independent sitting and maintain (for at least 10 seconds or more)	Able to sit momentarily (less than 10 seconds), when placed	Cannot sit	

For children <2

HAMMERSMITH INFANT NEUROLOGICAL EXAMINATION (HINE)

Adapted May 2021 by Anna Mayhew to improve measurability and provide linkage with WHO MM and RHS

Motor Function	Milestone Progression Score – may perform in any observed order					Score
	0	1	2	3	4	
Voluntary Grasp	No grasp	Uses whole hand	Index finger and thumb but immature grasp	Pincer grasp (thumb and index finger only)		
Ability to Kick (in supine)	No kicking	Kicks horizontal; legs do not lift	Upward (vertical)	Touches leg	Touches toes	
Head control	Unable to maintain upright	Wobbles	All the time upright (At least 30 seconds)			
Rolling	No rolling	Rolling to side (Perpendicular pelvis and shoulders to mat. At least to one side)	Prone to supine (At least in one direction)	Supine to prone (At least in one direction)		
Sitting	Cannot sit	Sits with support at hips	Props	Stable sit (No arms support and for 10 seconds)	Pivots (rotates) (At least 30° pelvis pivot – moves legs)	
Crawling	Does not lift head	On elbow (Maintain head up for at least 3 seconds)	On outstretched hand (Maintain head up for at least 3 seconds)	Crawling flat on abdomen	On hands and knees (At least 3 continuous, consecutive movements in a row)	
Standing	Does not support weight	Supports weight	Stands with support (At least 10 seconds supported standing, hands only -no leaning trunk)	Stands unaided (At least 10 seconds)		
Walking	No walking	Bouncing	Cruising (holding on) (At least 5 steps)	Walking independently (At least 5 steady steps)		
Total Score out of 26						/26