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Background

- ✓ The myotonic disorders are a heterogeneous group of genetically determined diseases that are unified by the presence of myotonia, which is defined as failure of muscle relaxation after activation¹
- ✓ The presentation of these disorders ranges from asymptomatic electrical myotonia, as seen in some forms of Myotonia Congenita (MC), to severe disability with muscle weakness, cardiac conduction defects and other systemic features, as in Myotonic Dystrophy type I (MD1)¹
- ✓ Currently, there is no standardized instrument to assess symptom severity. It has been used, for clinical trials, an automated interactive telephone-based voice response diary (IVR) to record frequency and severity of stiffness²
- ✓ Recently, a commercially available quantitative measure of handgrip myotonia (QMA) has been used in patients with MD1³ and in MC⁴, to calculate peak force and relaxation times following forced handgrips
- ✓ More recently, a device based on a sensor-engineered glove (SEGT) has been developed to perform quantitative analysis of motor performances during finger opposition sequences. This instrument has been used to objectively evaluate hand function and motor cortex connectivity and plasticity in healthy controls and subjects affected by various neurological diseases⁵
- ✓ Herein, we applied the sensor-engineered glove test (SEGT) to a population of subjects affected by MC and MD1, to assess myotonia and "warm-up" phenomenon

Patients & Methods

- ✓ SEGT was applied on 20 patients and compared with a cohort of healthy controls, evaluating touch duration and inter-tapping interval.
- ✓ Subjects:
 - 10 (MC) (5 on mexiletine; 5 drug-free)
 - 10 MD1
- ✓ Protocol:
 1. Finger tapping (FT) between thumb-index, making the fist, started after 5 minutes of rest and lasting for one minute, to assess time to fully release the hand;
 2. After 2 minutes of ice-application at the hand, the same protocol was repeated, without the initial rest.

Results

MC Patients on Mexiletine	Handgrip myotonia	Warm-up	Effects of cold
1. M, 17-yrs	Yes	Yes	Worsening
2. M, 20-yrs	Yes	Yes	None
3. F, 45-yrs	Yes	Yes	None
4. M, 47-yrs	Yes - slight	Yes	Worsening
5. M, 57-yrs	Yes - slight	Yes	None

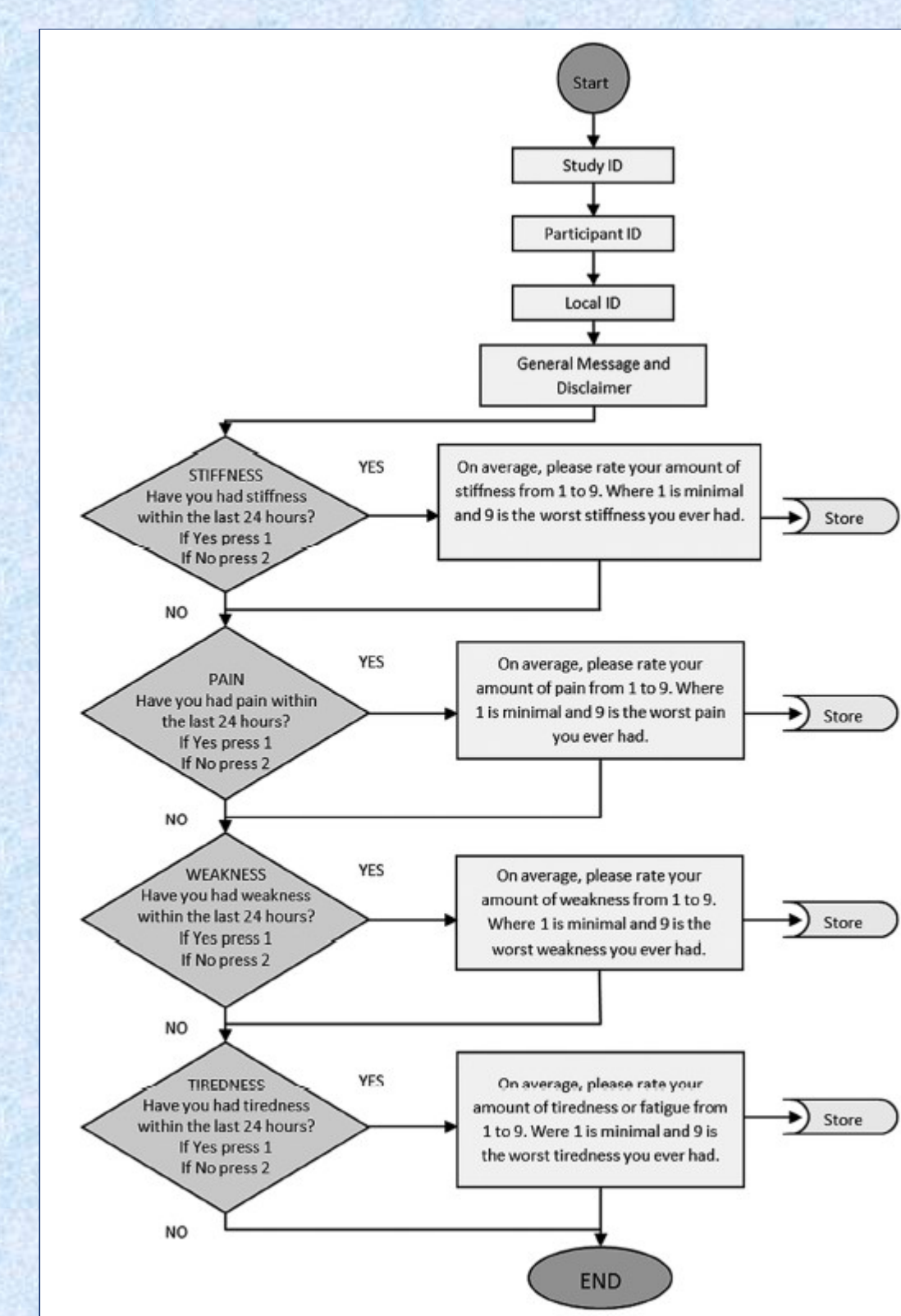
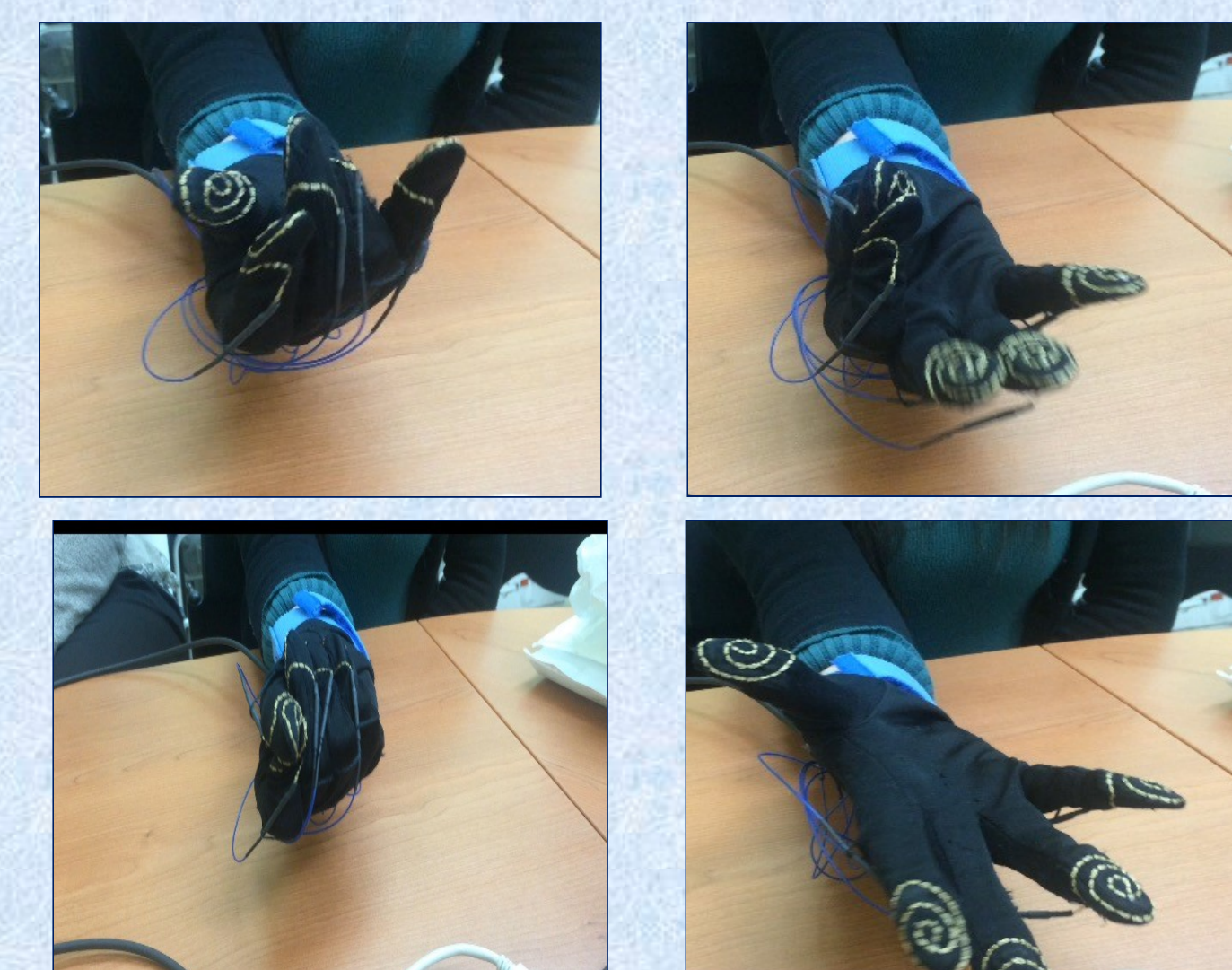
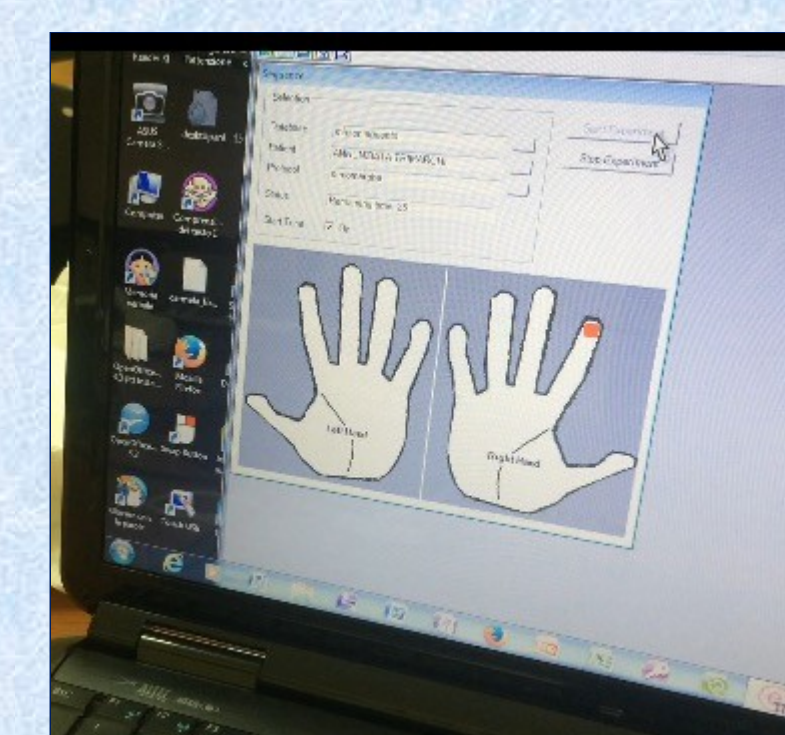
SEGT parameters MC pts on Mexiletine	Patient 1 M, 17-yrs	Patient 2 M, 20-yrs	Patient 3 F, 45-yrs	Patient 4 M, 47-yrs	Patient 5 M, 57-yrs
TD Global					
1° test	900,28	214,43	400,12	513,09	744,61
2° test after ice	578,67	380,6	406,21	662,43	874,63
ITI					
1° test	598,79	557,93	604,84	538,98	827,03
2° test after ice	642,29	418,88	493,38	584,48	597,9

MC Patients (no Mexiletine)	Handgrip myotonia	Warm-up	Effects of cold
M, 36-yrs	Yes	Yes	Worsening
F, 56-yrs	No, only generalized stiffness	No	Worsening
F, 22-yrs	Yes	Yes	Worsening
M, 19-yrs	Yes	Yes	None
M, 44-yrs	Yes	Yes	None

SEGT parameters MC, no Mexiletine	Patient 1 M, 36-yrs	Patient 2 F, 56-yrs	Patient 3 F, 22-yrs	Patient 4 M, 19-yrs	Patient 5 M, 44-yrs
TD Global					
1° test	391,07	477,36	700,02	577,97	637,87
2° test after ice	800,76	514,07	521,97	537,65	632,21
ITI					
1° test	1.056,37	535,81	1.284,85	1.396,07	598,7
2° test after ice	2.499,63	921,35	1.350,55	595,53	410,77

MD1 Patient	Handgrip myotonia	Warm-up	Effects of cold
Pt 1. F, 15-yrs	Yes	Yes	Not reported
Pt 2. M, 18-yrs	Yes	Yes	Worsening*
Pt 3. F, 47-yrs	Yes	Yes	Worsening
Pt 4.F, 27-yrs	Yes	Yes	Worsening
Pt 5.F, 21-yrs	Yes	Yes	Not reported
Pt 6.M, 52-yrs	Yes	Yes	Not reported
Pt 7.M, 28-yrs	Yes	Yes	Not reported
Pt 8.F, 29-yrs	Yes	Yes	Not reported
Pt 9. F, 36-yrs	Yes	Yes	Not reported
Pt 10. M, 41-yrs	Yes	Yes	Not reported

SEGT values MD1 pts	Pt 1 F, 15-yrs	Pt 2 M, 18-yrs	Pt 3 F, 47-yrs	Pt 4 F, 27-yrs	Pt 5 F, 21-yrs	Pt 6 M, 52-yrs	Pt 7 M, 28-yrs	Pt 8 F, 29-yrs	Pt 9 F, 36-yrs	Pt 10 M, 41-yrs
TD Global										
1° test	1.096,07	1.255,19	1.347,85	1.528,47	1.856,80	1.092,24	937,84	1.360,39	1.829,53	1.555,53
2° test after ice	722,21	950,53	1.372,57	2.381,60	2.151,38	875,14	869,35	1.141,52	1.802,58	2.748,08
ITI										
1° test	2.455,00	2.485,73	1.173,05	1.251,38	1.923,21	1.318,00	1.403,96	1.493,83	1.332,44	1.737,88
2° test after ice	1.103,09	2.212,00	1.207,15	1.635,79	1.241,80	1.298,59	1.345,84	1.300,96	1.080,78	1.626,17



Discussion

- ✓ The SEGT is a non-invasive and sensitive test to quantify hand grip myotonia
- ✓ All previously described tests of hand function were a combination of qualitative and semi-quantitative evaluations: dynamometry only evaluated grip or pinch strength, but did not consider dexterity and did not give any information about grasp patterns, precision and accuracy
- ✓ The SEGT describes, in an objective and quantitative way, the spatial and temporal accuracy of finger opposition movements
- ✓ The ability to quantify the relaxation times, to establish the pattern of myotonia on sequential handgrips and to evaluate the influence of cold may be useful to evaluate and to monitor treatment response and efficacy in future trials
- ✓ The SEGT, associated to the IVR, could be a useful tool to give a global assessment of myotonia (objectively and subjectively)
- ✓ The limitation of the study is the small sample size and the random application of the SEGT (i.e. not before, during and after treatment)

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