Do Not Replace Personal Patient/Doctor Interactions by Electronic Visits in Myotonic Dystrophy Patients

Josef Finsterer^a Fulvio Alexandre Scorza^b

^aKlinik Landstrasse, Messerli Institute, Vienna, Austria; ^bDisciplina de Neurociência, Universidade Federal de São Paulo/Escola Paulista de Medicina (UNIFESP/EPM), São Paulo, Brazil

With interest we read the article by Wenninger et al. [1] about an online survey on current symptoms among 207 patients with myotonic dystrophy type-1 (MD1) and 187 patients with myotonic dystrophy type-2 (MD2). It was concluded that the results are consistent with the results of clinical studies and online registries and that systematic online gathering of data from patients with rare diseases is useful and provides realistic and appropriate results [1]. The study has a number of limitations and raises the following comments and concerns.

The main shortcoming of the study is that the clinical findings were not related to the genotype. Since severity of the phenotype correlates with the expansion size of the CTG-repeat (MD1) and, respectively, the CCTG-repeat (MD2), it is crucial to include these genetic data. Genetic data could, to some extent, certify that the answers provided are in accordance with what could be expected from the genetic background. It would be also useful to correlate the individual disease duration with quantitative parameters of disease severity.

A second shortcoming is that it is unclear if the patient himself, any of his relatives, or any of his caregivers responded to the questionnaire. If the patient was asked to respond himself, it is crucial to consider his mental status.

For children with MD1/MD2, their parents were allowed to fill in the questionnaire. We should know who filled in the form for those who were mentally unable to perform the task. If mentally handicapped patients were excluded, the study has a bias toward mentally healthy patients. Thus, a major spectrum of the phenotype may not be covered by the survey. Were patients asked for cognitive impairment?

On the one side, the authors concluded that the online survey provides realistic and appropriate results, whereas on the other side the validity of the data is assessed as not reliable due to the absence of an assessor and the possibility of fraud. This contradiction should be solved.

A further shortcoming is that patient information about symptoms is not always reliable. This is not only due to cognitive impairment but may be also due to misinterpretation of a symptom as normal, extinction of symptoms, or overestimation of complaints [2]. This is why the clinician needs to actively ask for the presence or absence of particular symptoms and needs to carefully guide the patient and interpret his answers.

Electronic surveys have also the disadvantage that they could entrap the treating physicians to relinquish from personal visits, which was obviously an intention of the

survey. Since MD1/MD2 patients can die suddenly from ventricular arrhythmias or acute heart failure [3], it is crucial that these patients are regularly seen by their treating physicians and that appropriate investigations are routinely carried out during regular follow-up visits. Additionally, MD1/MD2 patients may not always be aware of seizures: they might have missed because of living alone or not experiencing manifest post-ictal complications [4]. This is another reason why multi-morbid patients need to be physically seen and investigated. Personal contacts have also the advantage that not only acutely arising questions can be asked by the physician or the patient, but that the physician can also inspect the patient already during verbal interaction with the patient. Thus, personal contact helps collect much more useful information than an inflexible questionnaire.

It remains unclear if only patients with a genetically confirmed diagnosis were contacted or also patients with suspected MD1/MD2. Did patients need to know their diagnosis? Missing in the questionnaire are questions about sleep quality, palpitations, exertional dyspnea, leg edema, and weight loss/gain.

Overall, a stiff questionnaire cannot replace personal guidance and assessment of individual history by a physician who will implement his personal experience and considerations in the course and outcome of a present patient/doctor interaction. We should not shift our responsibility to the patient or his caregivers and should

admit that personal contact is also a social contact for the patient and the physician and thus can improve the patient's overall well-being.

Statement of Ethics

Ethical standards were met.

Conflict of Interest Statement

The authors have no conflicts of interest to disclose.

Funding Sources

No funding was received.

Author Contributions

J.F.: design, literature search, discussion, first draft, critical comments, and final approval. F.S.: literature search, discussion, critical comments, and final approval.

Informed Consent

Informed consent was obtained. The study was approved by the Institutional Review Board.

References

- 1 Wenninger S, Stahl K, Montagnese F, Schoser B. Utility and results from a patient-reported online survey in myotonic dystrophies types 1 and 2. *Eur Neurol*. 2020;83:523–33.
- 2 Sui J, Humphreys GW. The self survives extinction: self-association biases attention in patients with visual extinction. *Cortex*. 2017; 95:248–56.
- 3 Itoh H, Hisamatsu T, Tamura T, Segawa K, Takahashi T, Takada H, et al. Cardiac conduction disorders as markers of cardiac events in myotonic dystrophy type 1. *J Am Heart Assoc*. 2020;9:e015709.
- 4 Salinsky M, Binder L, Storzbach D, Parko K, Rutecki P, Goy E, et al. Validity testing in veterans with epileptic seizures and psychogenic nonepileptic seizures. *Epilepsy Behav*. 2020; 111:107246.