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MRI Parameters in Myotonic Dystrophy type-1 and type-2 can be Highly Dependent on CTG/CCTG-Repeat Size

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LETTER TO THE EDITOR

We read with interest the article by Maj *et al*. about a retrospective functional MRI (fMRI) study of 19 patients with myotonic dystrophy type -1 (MD1) and 16 patients with myotonic dystrophy type-2 (MD2) on impairment of white matter tracts, by measuring fractional anisotropy (FA), mean diffusivity (MD), radial diffusivity (RD), and axial diffusivitv (AD) [Maj, E. *et al*., 2023]. FA was decreased in most white matter tracts in MD1 and less in MD2 patients, MD and RD values were increased in 47 tracts in MD1, and AD values were increased in MD1 compared to MD2 patients [Maj, E. *et al*., 2023]. It was concluded that MD1 patients are more likely than MD2 patients to develop diffuse disintegration of white matter pathways [Maj, E. *et al*., 2023]. The study is impressive, but some points should be discussed.

The major limitation of the study is that fMRI parameters (FA, MD, RD, AD) were not correlated with the CTG-repeat expansion in the 19 MD1 patients or with the CCTG-repeat expansion in the 16 MD patients. Since the severity of the phenotype depends not only on age, gender, and disease stage, but also on the severity of the genetic defect, it is important to know whether the fMRI parameters correlated with the CTG- or the CCTG-repeat expansion. The extent of WMLs can be completely different in an MD1 patient with 300 CTG-repeats and a patient with 1200 CTG-repeats. Since the method section indicates that only genetically confirmed patients were included, CTG and CCTG-repeat sizes must be available, and it is unclear why this particular analysis was not performed.

A second limitation of the study is that the fMRI parameters were not correlated with the degree of global, regional, or focal brain atrophy [Maj, E. *et al*., 2023]. MD1 patients in particular are known to develop decrease brain volume as the disease progresses [Cabada, T. *et al*., 2017]. Therefore, the results can only be interpreted in the context of the extent of global or regional atrophy in the included patients.

We disagree with the statement in the introduction that white matter lesions (WMLs) in MD1 are located in the frontal and temporal lobes [Maj, E. *et al*., 2023]. WMLs in MD1 occur throughout the brain and can even affect the brainstem [Igreja, L. *et al*., 2023]. We also disagree with the statement that cerebral lesions in MD1 mainly affect the white matter [Maj, E. *et al*., 2023]. Depending on the stage of the disease, white and grey matter lesions can be found in MD1, as documented in previous fMRI studies [Labayru, G]. Grey matter lesions have been found particularly the thalamus [Labayru, G. *et al*., 2022].

Because the mean age was significantly lower in MD1 compared to MD2 patients [Maj, E. *et al*., 2023] and because FA parameters may depend on age, the comparison between the two patient groups is of limited significance and the results should be interpreted with caution.

When MD1 was compared with healthy controls-1 (HC1), FA was decreased and MD, RD, and AD were increased in most tracts examined [Maj, E. *et al*., 2023]. However, no differences between fMRI parameters were found when comparing HC1 with MD1 [Maj, E. *et al*., 2023]. This discrepancy should be resolved.

In summary, the excellent study has limitations that should be addressed before drawing final conclusions. Clarifying the weaknesses would strengthen the conclusions and could improve the study. fMRI results in MD1 and MD2 patients need to be interpreted in relation to the genetic background and degree of cerebral atrophy. Before comparing MD1 with MD2 patients, it must be ensured that they are compatible in terms of age, gender, and disease duration.

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