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Profiling morphologic MRI features of motor neuron disease caused by TARDBP mutations

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State of the art. Mutations in the *TARDBP* gene are a rare cause of genetic motor neuron disease (MND). Our objective was to investigate distinctive clinical and MRI features of a relatively sized sample of MND patients carrying *TARDBP*mutations.
Methodology. Eleven *TARDBP* mutated patients and eleven sporadic MND (sMND) patients matched by age, sex, clinical presentation and disease severity were enrolled, along with 22 healthy controls. Patients underwent clinical and cognitive evaluations, as well as 3D T1-weighted and diffusion tensor (DT) MRI on a 3 Tesla scanner. Grey matter (GM) atrophy was investigated using voxel-based morphometry (VBM). GM volumes and DT MRI values of white matter (WM) tracts were also obtained.
Results. Greater impairment at naming tasks was observed in *TARDBP* mutation carriers, compared with sMND. VBM analysis showed significant atrophy of the lateral parietal cortex in TARDBP patients, compared with controls. Distinctive reductions of GM volumes were found in the left precuneus and right angular gyrus of TARDBP patients. Significant decreased fractional anisotropy of the right CST and increased axial diffusivity of the left ILF was detected only in *TARDBP* mutation carriers.
Conclusions. TARDBP patients showed a distinctive pattern of parietal cortical atrophy and greater damage of motor and extra-motor WM tracts compared with controls, that sMND patients matched for disease severity and clinical presentation were lacking. TDP-43 pathology due to *TARDBP* mutations may cause deeper morphologic alterations in both GM and WM.
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Conflicts of interest

N/A