DystoniaNet: Neural Biomarker-Based Platform for Dystonia Diagnosis using Deep Learning

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Objective: Development and validation of a deep learning algorithmic platform for the identification of a microstructural biomarker for objective, accurate, and rapid diagnosis of dystonia.

Background: Dystonia is a neurological movement disorder causing involuntary muscle contractions leading to abnormal movements and postures. The diagnosis of dystonia is challenging due to the absence of an objective biomarker of the disorder. It is estimated that patients receive the final diagnosis, on average, 10.4 years after symptom onset [1, 2], while clinicians consistently show poor agreement rates in diagnosing dystonia [3, 4]. Previous neuroimaging studies have reported microstructural brain abnormalities in dystonia patients, which may contribute to its pathophysiology [5]. Using structural brain MRI scans in a large cohort of dystonia patients and matched healthy controls, we developed and validated a deep learning algorithmic platform for identification of a dystonia neural biomarker and its utilization for objective, accurate and rapid diagnosis of dystonia.

Methods: Structural MRI data were acquired from 521 individuals, including 301 patients with three forms of dystonia (279 laryngeal, 12 cervical, 10 blepharospasm) and 220 healthy controls. DystoniaNet, a deep learning algorithm based on 3D convolutional layers, was developed to automatically identify a microstructural biomarker of dystonia from raw brain MRIs, and then used for automatic diagnosis of dystonia. The performance of DystoniaNet was validated using out-of-sample patients.

Results: The microstructural biomarker was identified in the corpus callosum, thalamic radiation, inferior fronto-occipital fasciculus and temporal gyrus, all of which have been previously reported to be abnormal in patients with various forms of dystonia. Based on this biomarker, DystoniaNet achieved a high diagnostic accuracy of 97.5% with a low referral rate of 1.2% across different forms of dystonia. The diagnostic decision of DystoniaNet was made in 0.36 seconds.

Conclusions: DystoniaNet achieved nearly three-fold improvement in diagnosing dystonia compared to a 34% agreement rate between physicians [3]. DystoniaNet may be implemented as a biomarker-based, objective, and generalizable algorithmic platform to enhance clinical decision making for dystonia diagnosis.

References:

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