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

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Background – Diagnosing dystonia is a challenging task

Dystonia is a rare movement disorder characterized by involuntary muscle contractions, leading to abnormal movements and postures. **Neural markers of dystonia are non-existent**, resulting in poor agreement rate between clinicians [1,2] and significant delays in diagnosis and treatment [3,4].

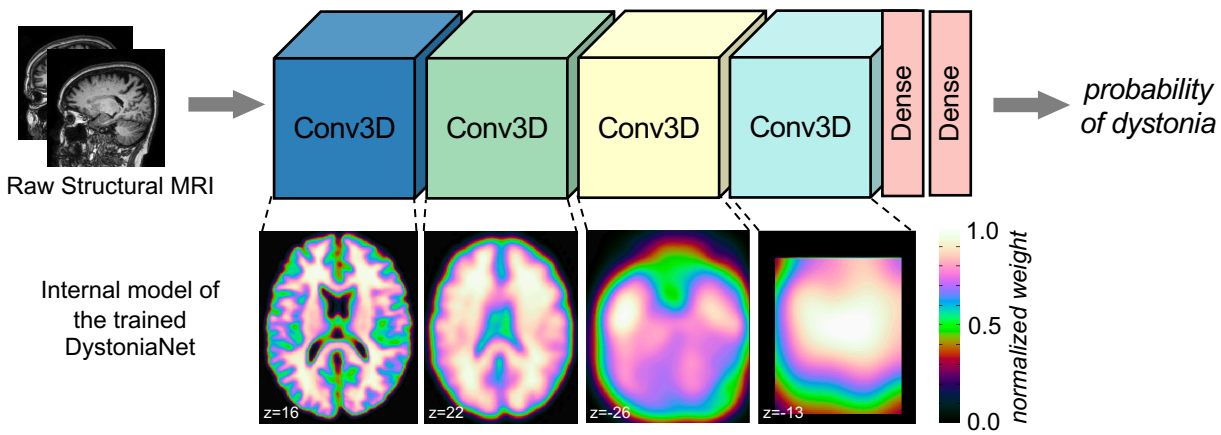
Objective – Identify and validate a biomarker for dystonia diagnosis

- Develop a deep learning algorithm (**DystoniaNet**) for the identification of diagnostic biomarker.
- Validate biomarker in a **large cohort** of 612 subjects.

Methods – A deep learning platform and a large cohort

Structural MRI data were acquired in **612 subjects**, including 392 patients with three different forms of isolated focal dystonia and 220 healthy controls:

- **Training set**: 160 patients with laryngeal dystonia and 160 age/sex-matched healthy controls
- **Optimization set**: 60 patients with laryngeal dystonia and 60 healthy controls
- **Validation set**: 172 patients with laryngeal dystonia, blepharospasm, cervical dystonia



The architecture of DystoniaNet included four convolutional layers and two dense layers that extracted features from raw structural brain MRIs and predicted the probability of dystonia. Axial brain slices depict the average normalized weight of the 3D feature maps extracted from the corresponding convolutional layers of DystoniaNet trained on 160 patients and 160 controls.

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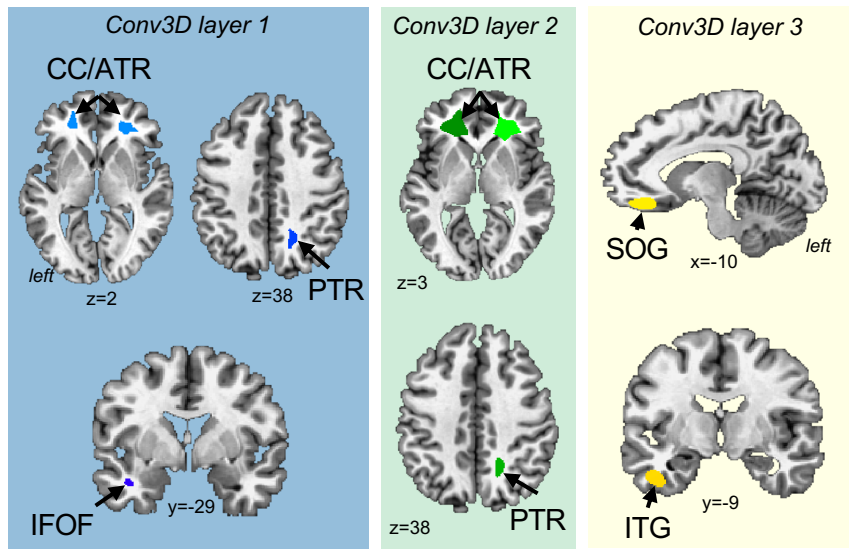
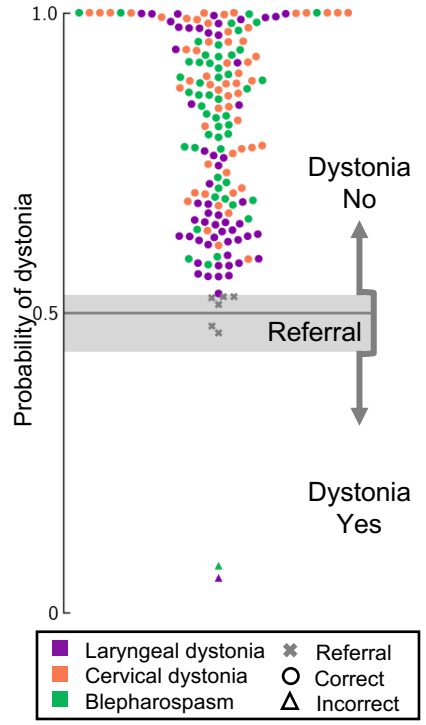
Results – Objective biomarker for dystonia diagnosis

Structural neural biomarker for automatic diagnosis of dystonia as identified by DystoniaNet



Axial and sagittal brain slices visualize the most discriminative features in the first three 3D convolutional layers of DystoniaNet in standard Talairach-Tournoux space. CC/ATR – corpus callosum/anterior thalamic radiation of corona radiata; PTR – posterior thalamic radiation of corona radiata; IFOF – inferior fronto-occipital fasciculus; SOG – superior orbital gyrus; ITG – inferior temporal gyrus.

Accuracy = 98.8%
Referral rate = 3.5%



Conclusions – DystoniaNet delivers objective diagnosis of dystonia

- DystoniaNet identified a neural biomarker including regions previously reported as abnormal in patients with dystonia [5] and achieved nearly **three-fold improvement in diagnosing dystonia** compared to a 34% agreement rate between physicians [1].
- DystoniaNet may be implemented as a biomarker-based, objective, and generalizable algorithmic platform to enhance clinical decision-making for dystonia diagnosis

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