

MALDI-IMS LIPID FINGERPRINTING: A MACHINE-LEARNING METHODOLOGY FOR ACCURATE ALS AND MYOPATHY DIAGNOSIS

An *in vitro* diagnostic methodology using Lipid Imaging Mass Spectrometry (LIMS) on muscle biopsies to achieve reliable early detection, differential diagnosis, and patient stratification for Amyotrophic Lateral Sclerosis (ALS) and myopathies

Research goal

To establish a diagnostic methodology based on muscle lipid phenotypes to overcome the critical bottleneck of late and complex clinical diagnosis in neuromuscular diseases. By mapping specific lipid signatures directly from skeletal muscle biopsies via Matrix-Assisted Laser Desorption Ionization Mass Spectrometry Imaging (MALDI-IMS), the system trains machine-learning classifiers to identify and differentiate ALS from mimicking myopathies. This tool facilitates objective early detection, precise disease staging, and quantifiable tracking of therapeutic efficacy in clinical trials

Problem to solve

Amyotrophic Lateral Sclerosis (ALS) is a rapidly progressive, fatal neurodegenerative condition characterized by a devastatingly complex, slow diagnostic pathway primarily reliant on subjective clinical and electrophysiological exclusion methods. This diagnostic delay creates a massive window where the disease progresses unhindered and effectively blocks early therapeutic intervention. Furthermore, clinicians face a profound lack of objective biomarkers to accurately stratify highly heterogeneous patient cohorts or to clearly differentiate ALS from mimicking myopathies, which heavily compromises the success rates of upcoming clinical trials.

Innovative Aspects

- **Early Pre-Symptomatic Detection:** Successfully identifies disease-specific metabolic and lipid updates at early stages, as validated in robust hSOD1G93A mouse configurations.
- **High-Performance Machine Learning:** Evaluated through advanced regression models (Random Forest, SVM, Naive Bayes), achieving exceptional classification accuracy, sensitivity, and optimal specificity across human patient cohorts.
- **Clear-Cut Differential Staging:** Distinctively isolates pure ALS signatures from structural myopathies or muscular dystrophies using a highly refined minimum 5-lipid biomarker panel.
- **Sex-Specific Precision Parameters:** Novel integration of gender-dimensional screening metrics to capture distinct metabolic profiles across males and females, significantly reducing diagnostic blind spots.

Market opportunity

This diagnostic methodology turns a complex clinical bottleneck into a standardized screening solution, offering significant value to CROs and pharmaceutical entities looking to optimize patient stratification in neuromuscular clinical trials. Operating on standard hospital mass spectrometry setups without requiring complex chemical extraction, it serves as a highly scalable candidate for a rapid companion diagnostic tool to evaluate treatment responses.

Intellectual Property:

Priority European patent application filed (September 19, 2024) **EP26382477.3**

Development stage:



Available for: *Licensing, co-development*

Aims

Looking for a partner interested in a license and/or a collaboration agreement to develop and exploit this asset.



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