



PROGETTO

Supervisore::

Prof.ssa Maria Assunta Rocca

Titolo/Title:

Identifying Distinct Diagnostic and Prognostic Phenotypes
in rare central nervous system disimmune disorders

Curriculum:

Neuroscienze e Neurologia Sperimentale

Link alla pagina personale del sito web di
Ateneo o del polo ospedaliero di riferimento:

<https://www.unisr.it/docenti/r/rocca-maria-assunta>

[https://research.hsr.it/en/institutes/institute-of-experimental-
neurology/neuroimaging-of-CNS-white-matter/maria-assunta-
rocca.html](https://research.hsr.it/en/institutes/institute-of-experimental-neurology/neuroimaging-of-CNS-white-matter/maria-assunta-rocca.html)

Descrizione del progetto (max 3.000 caratteri spazi inclusi)

Background/gap of knowledge

The identification of antibodies against aquaporin-4 (AQP4), myelin oligodendrocyte glycoprotein (MOG), and several other central nervous system (CNS) antigens has significantly refined the classification of different clinical and MRI presentations within neuromyelitis optica spectrum disorder (NMOSD), MOG associated disease (MOGAD) and other rare central nervous system (CNS) disimmune disorders. Nonetheless, there is still a clinical and research need for a deeper understanding of the pathophysiological mechanisms underlying these conditions, for diagnostic, prognostic and therapeutic purposes. Among these conditions, AQP4-negative NMOSD likely includes other conditions driven by different pathogenetic processes which generate similar clinical and MRI pictures. Not rarely, CNS affections with a possible disimmune etiopathogenesis cannot be better classified in a known definite entity.

Rationale and hypothesis

Together with laboratory testing of serum and cerebrospinal fluid (CSF) specimens, the MRI of brain, optic nerve and spinal cord is an essential tool in the evaluation of CNS disimmune disorders, and plays a relevant role in the current nosographic characterization, as well as in the treatment choice. Optical coherence tomography (OCT) is another non invasive technique which allows evaluating retinal damage occurring in these conditions. The combined evaluation of clinical, laboratory, MRI and OCT data may allow to identify different disease phenotypes within rare CNS disimmune disorders, and to evaluate medium- and long-term prognosis as well as treatment response.

Objectives and specific aims

This project aims to exploit structural and functional MRI measures in a relatively large population of patients with NMOSD, MOGAD, disimmune encephalitis, secondary CNS vasculitis and not otherwise specified inflammatory CNS conditions to improve their differential diagnosis and to define their prognosis and treatment response.

A retrospective cohort of more than one hundred patients will be combined with a prospective cohort of newly recruited cases. A follow-up evaluation after one year will be evaluated, when available. Matched comparison groups including patients with multiple sclerosis and healthy subjects will also be included, with an expected total of more than 500 visits.

By evaluating conventional and advanced MRI data, an extensive fluid biomarkers assessment and OCT data, with cross-sectional and longitudinal approaches, this project aims to define specific disease phenotypes of rare CNS disimmune disorders with predictable clinical prognosis and treatment response.

Expected outcomes



To better characterize the diagnostic and prognostic relevance of conventional and advanced MRI, OCT and fluid biomarkers in different rare CNS disimmune disorders. The obtained findings will be validated in international cohorts from European collaborating neurological centers.

Competenze che deve acquisire lo studente (Max 600 caratteri spazi inclusi):

During the project, the student will acquire the following skills:

- collection of clinical data from CNS disimmune disorders patients with different characteristics;
- interpretation of clinical, OCT and brain and spinal cord MRI findings;
- post-processing of structural and functional MRI data from patients with CNS disimmune disorders and healthy controls;
- defining associations between clinical and OCT data, fluid biomarkers profiles and MRI measures in CNS disimmune disorders patients;
- presentation of works at National and International congresses;
- drafting of research reports and articles.

Bibliografia (max. 15)

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PROPOSTA PROGETTO DI RICERCA**

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