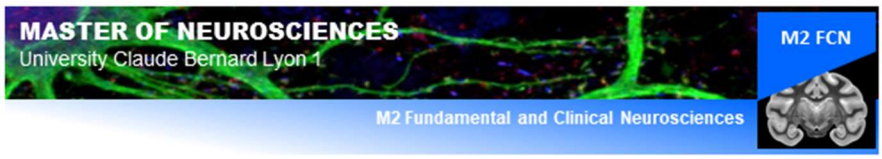

Université Claude Bernard Lyon 1

MASTER 2 Fundamental and Clinical Neurosciences

Internship proposal 2025-2026

(internship from January to June 2026)

Host laboratory: *Name + address*
Institut NeuroMyoGène, Laboratoire Physiopathologie et Génétique du Neurone et du Muscle ; Faculté de médecine, 8 av Rockefeller, 69008 Lyon. https://pgnm.inmg.fr/

Host team : *team name + website*
Equipe Schaeffer, https://pgnm.inmg.fr/schaeffer/

Internship supervisors : *name + position + email*
Arnaud JACQUIER, AHU (arnaud.jacquier@univ-lyon1.fr)

Project title : Investigating the implication of a new gene in Hereditary Sensory and Autonomic Neuropathies Using Induced Pluripotent Stem Cell (iPSC)-Derived Models

Project summary : *approx 10 lines*

Our laboratory specializes in the study of hereditary neuromuscular disorders, such as hereditary neuropathies (Charcot-Marie-Tooth disease, Spinal Muscular Atrophy), muscular dystrophies (Duchenne Muscular Dystrophy), and neuromuscular junction disorders (Congenital Myasthenic Syndromes). We aim to better understand the cellular and molecular mechanisms underlying these diseases, with a particular focus on the use of relevant human models derived from induced pluripotent stem cells (iPSCs).

This internship will focus on the functional characterization of a genetic mutation from a new gene involved in a hereditary sensory and autonomic neuropathy. The student will work with iPSC lines differentiated into sensory neurons (nociceptors) in order to reproduce the pathological phenotypes observed in patients. The aim is to demonstrate the pathogenicity of the mutation and to elucidate the associated pathophysiological mechanisms.

During this internship, you will be learned: Culture of human iPSCs and their derivatives, cellular differentiation techniques (sensory neurons), cell biology analyses (immunofluorescence, confocal imaging, viability assays), biochemistry (Western blot, protein analysis), molecular biology (RT-qPCR, transfection, sequencing).

3-5 recent publications :

- **Jacquier A**, Risson V, Simonet T, Roussange F, Lacoste N, Ribault S, Carras J, Theuriet J, Girard E, Grosjean I, Le Goff L, Kröger S, Meltoranta J, Bauché S, Sternberg D, Fournier E, Kostera-Pruszczyk A, O'Connor E, Eymard B, Lochmüller H, Martinat C, Schaeffer L. « Severe congenital myasthenic syndromes caused by agrin mutations affecting secretion by motoneurons ». **Acta Neuropathologica 2022**

- **Jacquier A**, Theuriet J, Fontaine F, Mosbach V, Lacoste N, Ribault S, Risson V, Carras J, Coudert L, Simonet T, Latour P, Stojkovic T, Piard J, Cosson A, Lesca G, Bouhour F, Allouche S, Puccio H, Pégat A, Schaeffer L. « Homozygous COQ7 mutation: a new cause of potentially treatable distal hereditary motor neuropathy ». **Brain, 2022**
- Musawi S, Donnio LM, Zhao Z, Magnani C, Rassinoux P, Binda O, Huang J, **Jacquier A**, Coudert L, Lomonte P, Martinat C, Schaeffer L, Mottet D, Côté J, Mari PO, Giglia-Mari G. « Nucleolar reorganization after cellular stress is orchestrated by SMN shuttling between nuclear compartments." **Nat Commun. 2023**
- Halegua T, Risson V, Carras J, Rouyer M, Coudert L, **Jacquier A***, Schaeffer L*, Ohlmann T and Mangeot P. "Delivery of precise Prime editing in human precursors cells using advanced pseudoviral NanoScribes particles." **Nat Commun. 2025**