Prion protein dysfunction in inherited metabolic myopathies

Our project aims at exploring a novel hypothesis to account for muscular deficits in patients suffering from metabolic myopathies. We propose that the cellular prion protein, which is described to play a role in muscle, may be involved in the pathophysiology of these rare diseases. By combining cellular and molecular approaches and by taking advantage of muscle cells from patients, our objective is to assess how this protein is affected in patients, to reach a better knowledge of the role exerted by this protein in muscle, and to define how an alteration of this protein could disrupt muscle function.