



## Fully funded PhD project

# Stability mapping of proteins involved in conformational diseases by nanopore technology

### Thesis project

Conformational diseases are protein misfolding pathologies often resulting in cellular degeneration. The incidence of a majority of human diseases is closely linked to population ageing and since the mean age in developed countries is expected to increase, these illnesses will further burden their communities. On the other hand, some of these conformational diseases are rare debilitating and can be fatal to young adults. All of these diseases result in reduced life expectancy and quality of life. ***Our proposal establishes a fundamental interdisciplinary research approach of protein stability in Human diseases. Wilson's disease is one of those pathologies that is manifested by an alteration of the ATP7B copper transporter and an accumulation of copper in the organs and especially in liver and brain. Atypical forms of the pathology are still difficult to manage. Indeed, for some patients, the disease has a late onset of the symptoms with serious medical consequences. Anticipating and predicting the impact of one or several combined mutations on the ATP7B conformation and on its function, will allow identifying among patients, those with a pejorative late outcome. Using nanopore technology, we aim to define the relationship between stability of mutant proteins and Wilson disease severity.***

**Key words:** Protein folding and stability, conformational diseases, single molecule, nanopores

**Directors:** Juan Pelta ([juan.pelta@univ-evry.fr](mailto:juan.pelta@univ-evry.fr)) and Philippe Manivet ([philippe.manivet@aphp.fr](mailto:philippe.manivet@aphp.fr))

**Co-supervisors:** Manuela Pastoriza-Gallego, Benjamin Cressiot

**Localization:** Paris area.

### References

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