JPND research
EU Joint Programme – Neurodegenerative Disease Research

ModelPolyQ

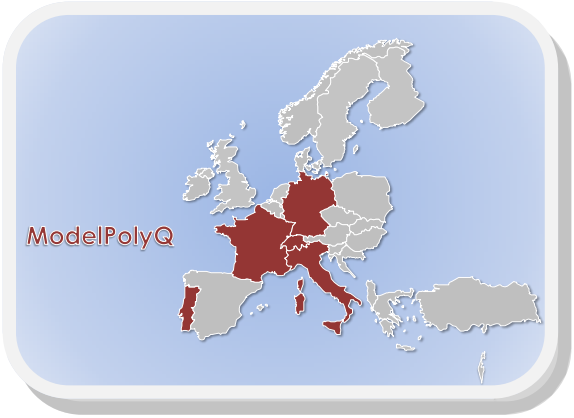
Advanced models of polyglutamine disorders (Huntington's disease, SCA3 and SCA7)

Polyglutamine (polyQ) diseases are a group of nine neurodegenerative diseases caused by the over-repetition of the CAG codon. This over-repetition translates into polyQ tracts within specific proteins for each disorder. Despite important progresses in the knowledge of the pathological mechanisms involved, effective therapies are still lacking.

Advances in this field depend on innovative, predictive models of disease for which there is an urgent need for both mechanistic and preclinical studies. In this project we focus on three polyQ disorders: Huntington's disease and spinocerebellar ataxias type 3/Machado-Joseph disease and 7.

We will generate novel, improved disease models; thoroughly characterize and compare both these and previously generated models; and standardize reproducible methodologies to investigate mechanisms that present commonalities between polyQ diseases. We concentrate on the leading models, namely genetically-modified rodent models and induced pluripotent stem cells (iPSC). The first have been and are instrumental to the progression of the field, while the latter offer the promise of enabling major advances.

We expect that this project will make important contributions to the field of polyQ diseases by providing the models and methodologies to enable significant advances in the understanding of the mechanisms of these diseases and to provide the tools for pre-clinical identification and validation of effective new therapies for polyQ disorders.



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