

## **PreFrontAls**

Searching for therapeutic interventions in Frontotemporal dementia with C9ORF72 repeat expansions in the presymptomatic stage

Frontotemporal dementias (FTD) constitute the second-most common types of dementia before the age of 65. These types of dementia are characterized by loss of neuronal cells in the frontal and temporal lobes of the brain, related to progressive behavioural changes, cognitive problems and language difficulties. Amyotrophic lateral sclerosis (ALS) accompanies FTD in 20% of the patients leading to progressive motor deficits and disability.

There are known genetic factors that play an important role in FTD, with mutations in three different genes: MAPT, GRN, and C9ORF72. The disease-causing mutation in the C9ORF72 gene is the most frequent mutation responsible for both FTD and ALS. This genetic defect has just recently been discovered and the disease processes in this type of FTD and ALS still have to be elucidated.

In this project we will examine the disease process in a group of individuals who carry the genetic mutation but who show no disease symptoms yet. This way we can examine sensitive changes (biomarkers) on MRI, in blood and cerebral spinal fluid samples, that could predict disease onset and progression. We can than evaluate these biomarkers as important outcome measure for disease-modifying treatments.

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<sup>\*</sup> Contributions from participating JPND Member Countries are currently being finalised for this project