Project title: Genetic and epigenetic modulAtors in Rare neurodegenerative diseases with DEmentia: a National study on autosomal dominant Alzheimer disease and genetic frontotemporal degeneration with dementIA (GARDENIA)

Project ID: PNRR-MR1-2022-12375654

CUP: C83C22001300001

Description: Current evidence suggests that the age at symptom onset is highly variable across the genes implicated in AD and FTD. Although mutation type can account for a large part of the observed variance in onset, substantial variation remains within many AD/FTD families and mutation types, suggesting the existence of genetic or environmental modifiers. Our working hypothesis is that, in genetic AD and FTD, common alterations of specific pathways may modulate, under distinct phenotypes and/or causing gene, the disease onset/progression; and that this paradigm could by applied also to other rare forms of dementia characterized by a wide age at onset range. Herein, we aim to create a national core collection of WES and WGM data on monogenic rare forms of AD/FTD and to expand knowledge on genetic/epigenetic modulators of age at onset. The final goal is to draw strategies for the development of innovative therapeutic approaches aimed at slowing disease progression in rare forms of dementia.

Financing Programme and Mission:
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IRCCS Role Coordinator/ Partner

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State of the project: Ongoing

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Website: https://www.fatebenefratelli.it/