## The UK GENetic Frontotemporal dementia Initiative (UK GENFI)

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The GENetic Frontotemporal dementia Initiative (GENFI) is an international multicentre cohort study investigating genetic forms of frontotemporal dementia (FTD), particularly those who have mutations in the progranulin (GRN), microtubule-associated protein tau (MAPT) and chromosome 9 open reading frame 72 (C9orf72) genes. However we also study rarer FTD-causing genes including TBK1, TARDBP, SQSTM1 and VCP.

The cohort is made up of both people who have developed symptoms and also those who are at risk of carrying one of the FTD mutations (i.e. siblings and children of symptomatic carriers). Studying presymptomatic individuals destined to develop the disease later in life allows us a window into the earliest changes in the disease process.



The UK GENFI study currently encompasses four active

The GENFI study collects biomarker data including:

- Clinical assessments
- Cognitive and neuropsychiatric data
- DNA, RNA, serum, and plasma
- CSF
- MRI including T1, T2, DTI, rsfMRI and ASL lacksquare

The key aims of the study include developing novel biomarkers of disease onset and progression.

sites although is also linked to cognitive and genetic clinics in Liverpool, Birmingham, Sheffield, Newcastle, Bristol, Southampton, and Exeter.

As well as contributing to the wider international GENFI study there are a number of UK-specific projects underway:

- GENFI-Cog iPad battery: we have developed a computerized cognitive battery of tests that can be performed at people's homes to assess the earliest cognitive changes in genetic FTD. This will be performed 3 monthly over 1 year.
- Eyetracking battery: we have developed a short eyetracking battery tapping into executive function and social cognition







## Figure 3: Current UK GENFI Recruitment





## Figure 2: UK GENFI sites

Allied to the UK GENFI study we also run a national support group for FTD family members – this meets once a year in London.



We are now preparing for the next phase of GENFI: we are working with a number of pharma companies to plan trials of novel therapies including antisense oligonucleotides using the UK GENFI platform.