Discovery Through Collaboration

10 Years Remarkable Progress in Familial Frontotemporal Dementia

Ten years ago, in October 1996, Dr. Norman Foster organized an international conference in Ann Arbor, Michigan on Neurological diseases genetically linked to chromosome 17. He decided to hold the conference after caring for a member of a family with dementia linked to chromosome 17, which he helped report on in 1994. The conference brought together a team of researchers from all around the world that had identified and studied other families with these traits.

At the time, several disorders were linked to chromosome 17, including frontotemporal dementia (FTD), parkinsonism, and other neurological and behavorial disturbances. However, it was unclear whether all of these chromosome 17-linked diseases were the same or if they had different causes.

One major finding from the conference was the conclusion that all families who had been studied appeared to have similar illnesses that were forms of FTD.

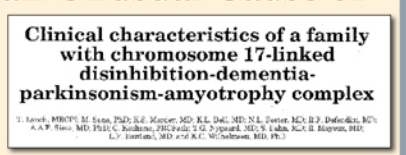
Two years later, investigators who attended the Ann Arbor conference collaborated with other researchers to identify mutations in the tau gene. This mutation was the cause of FTD in many, but not all of these families.

The search for a similar gene mutation in other families continued fruitlessly for the next 8 years, until this year. 10 years after the original conference, mutations in the progranulin gene were found to explain all the remaining cases. Remarkably, two genes located close together, but apparently otherwise unrelated, both cause FTD. Ten years of research and collaboration have finally paid off. Now many researchers are using this information to help develop treatments that can help patients with this disease.

1994

Reporting an Unusual Cause of Dementia:

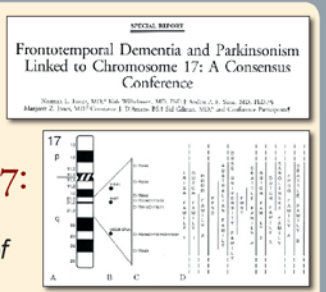
Clinical Study of an Extended Family with Chromosome 17-linked Dementia (Lynch et al., Neurology)


Clinical characteristics of a family with chromosome 17-linked disinhibition-dementia-parkinsonism-amyotrophy complex

1996

Conference on Familial FTD Linked to Chromosome 17:

A Consensus Panel (Foster et al., Annals of Neurology)

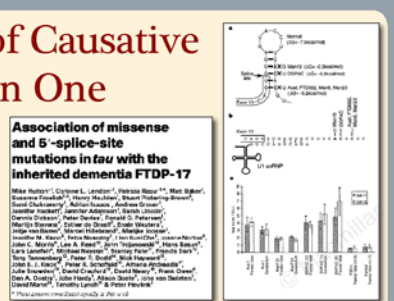

SPECIAL REPORT

Frontotemporal Dementia and Parkinsonism Linked to Chromosome 17: A Consensus Conference

1998

Discovery of Causative Mutations in One Gene:

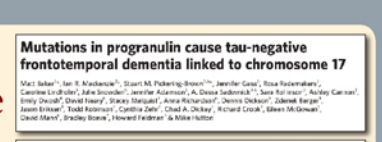
Mutation in tau gene found (Hutton et al., Nature)

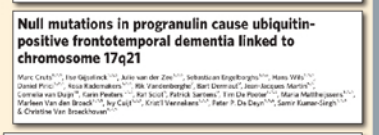

Association of missense and 5′-splice-site mutations in *tau* with the inherited dementia FTDP-17

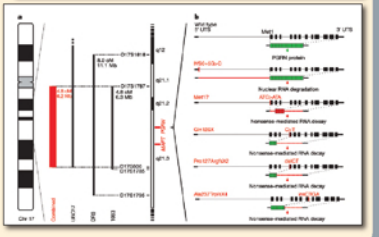
2006

Discovery of Causative Mutations in a Second Gene!!!

A second mutation found in progranulin gene (Cruts et al., Nature; Baker et al., Nature)


Mutations in progranulin cause tau-negative frontotemporal dementia linked to chromosome 17


Null mutations in progranulin cause ubiquitin-positive frontotemporal dementia linked to chromosome 17q21



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