

A microscopic image of neurons, likely from a mouse model of Frontotemporal Dementia (FTD). The neurons are stained with a blue dye (likely DAPI) to highlight the nuclei and a yellow/gold dye to highlight cytoplasmic inclusions, which are characteristic of neurofibrillary pathology. The neurons are interconnected by a network of fine, white, fibrous structures, possibly representing the cytoskeleton or extracellular matrix.

UCSF Weill Institute for Neurosciences

Memory and Aging Center

# Advances in FTD: Implications for the family

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Genetic Counselor

AFTD Education Conference, May 3, 2019



# What does this mean for my family?



Gather  
information  
about family

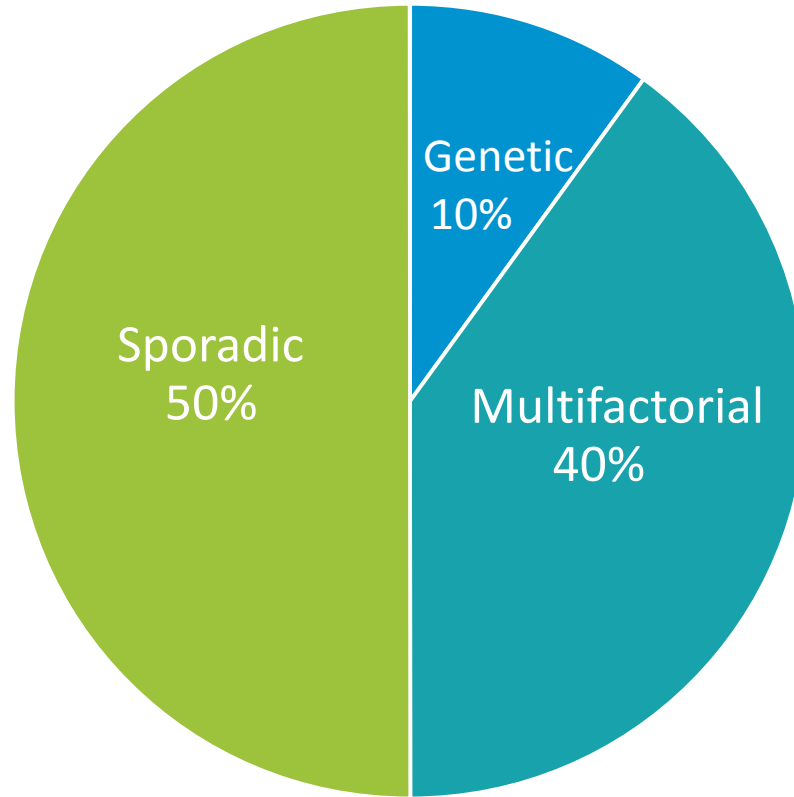
Discuss genetic  
testing with  
relative(s)

Contact a  
genetics  
professional

Consider  
participating in  
research



# Frontotemporal dementia by the numbers



## Genetic

## Multifactorial

## Sporadic

Threshold for  
FTD symptoms →



Number of genes in one family	1	Few to Many	Many
Degree of gene impact	Large	Moderate	Small

G = gene , E = environment



3 genes are responsible for genetic FTD\*

*MAPT* (“tau gene”)

*GRN* (“progranulin gene”)

*C9ORF72* (“C9 or FTD-ALS gene”)

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\* Most genetic families

Sporadic FTD



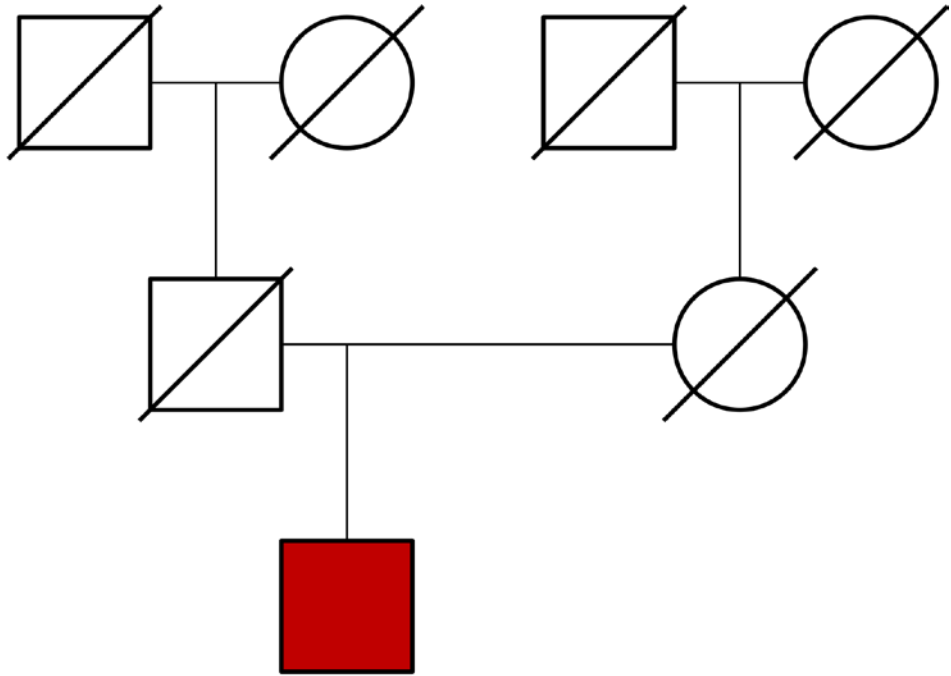
Multifactorial FTD



Genes involved in sporadic and multifactorial FTD are not entirely understood



# Sporadic FTD



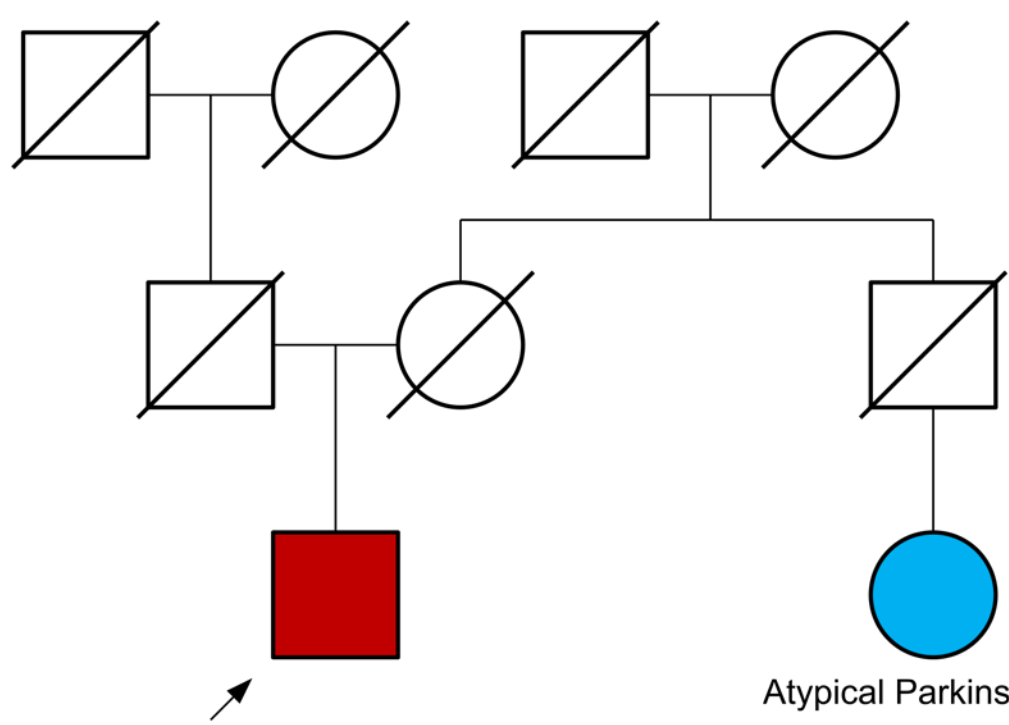
~~MAT~~

~~GM~~

~~C9orf72~~

Frontotemporal dementia

# Multifactorial FTD



Frontotemporal dementia

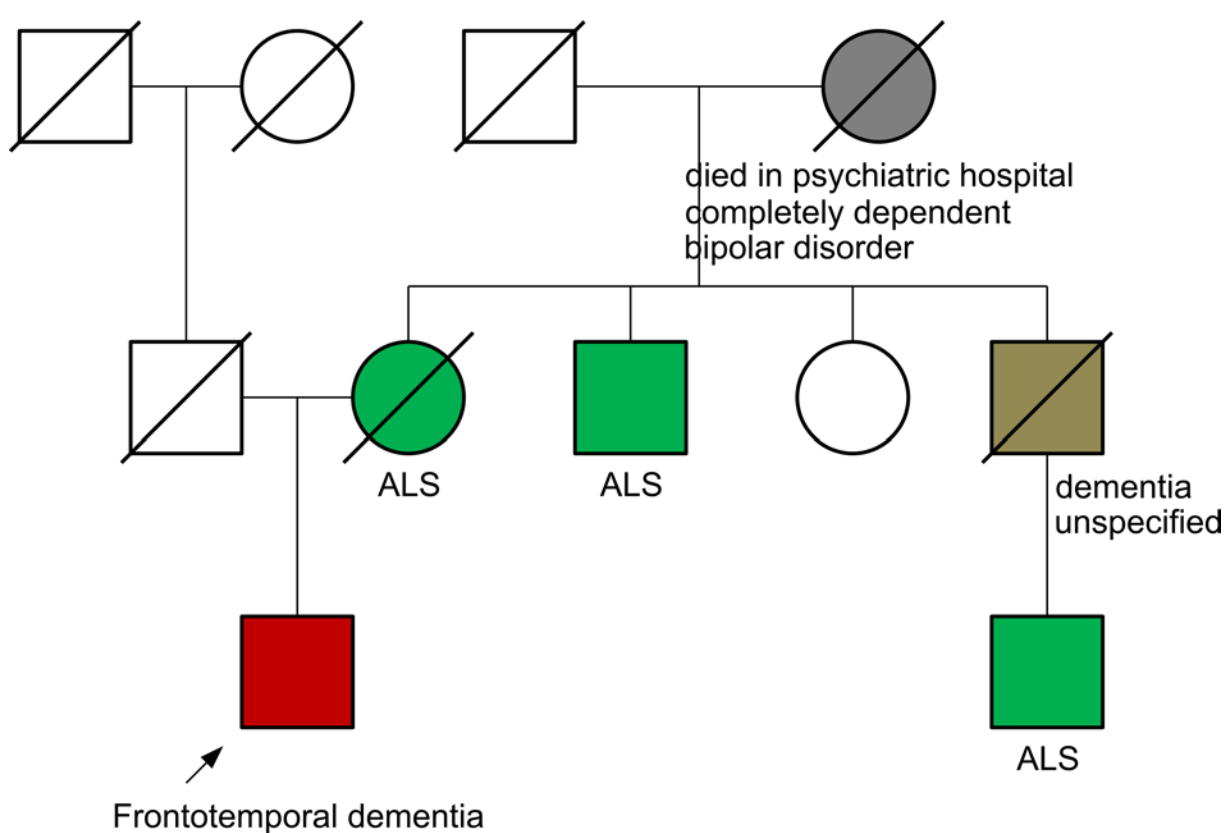
Atypical Parkinsonism

~~MAT~~

~~GRN~~

~~C9orf72~~

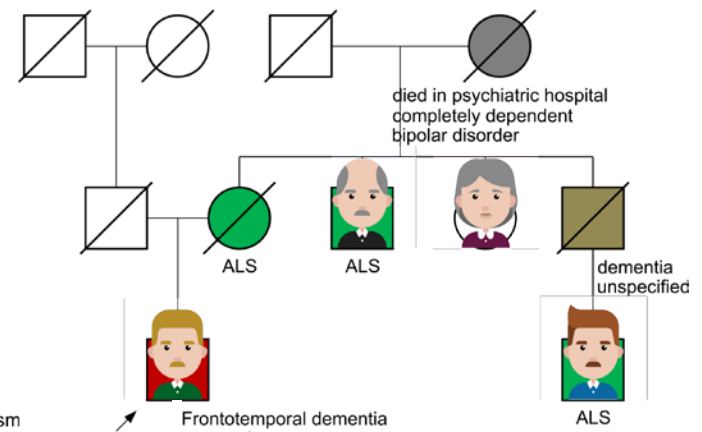
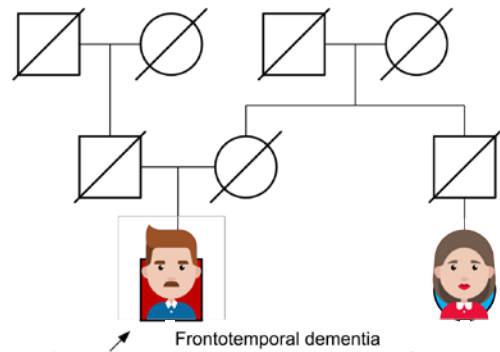
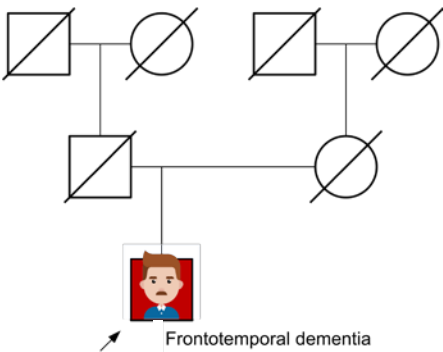
# Genetic FTD



~~MAPT~~

~~APP~~

*C9ORF72*  
✓



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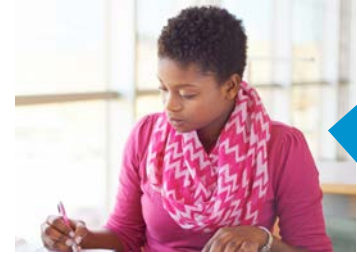


Gather  
information  
about family

Talk to your relatives



Record information



Ask questions

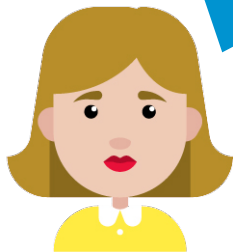
- Who is living or deceased?
- Current ages or ages at death?
- Who has dementia?
- Is there a medical diagnosis?
- Age at first symptom?
- Which symptoms at the beginning? Now?
- Genetic testing? Autopsy?



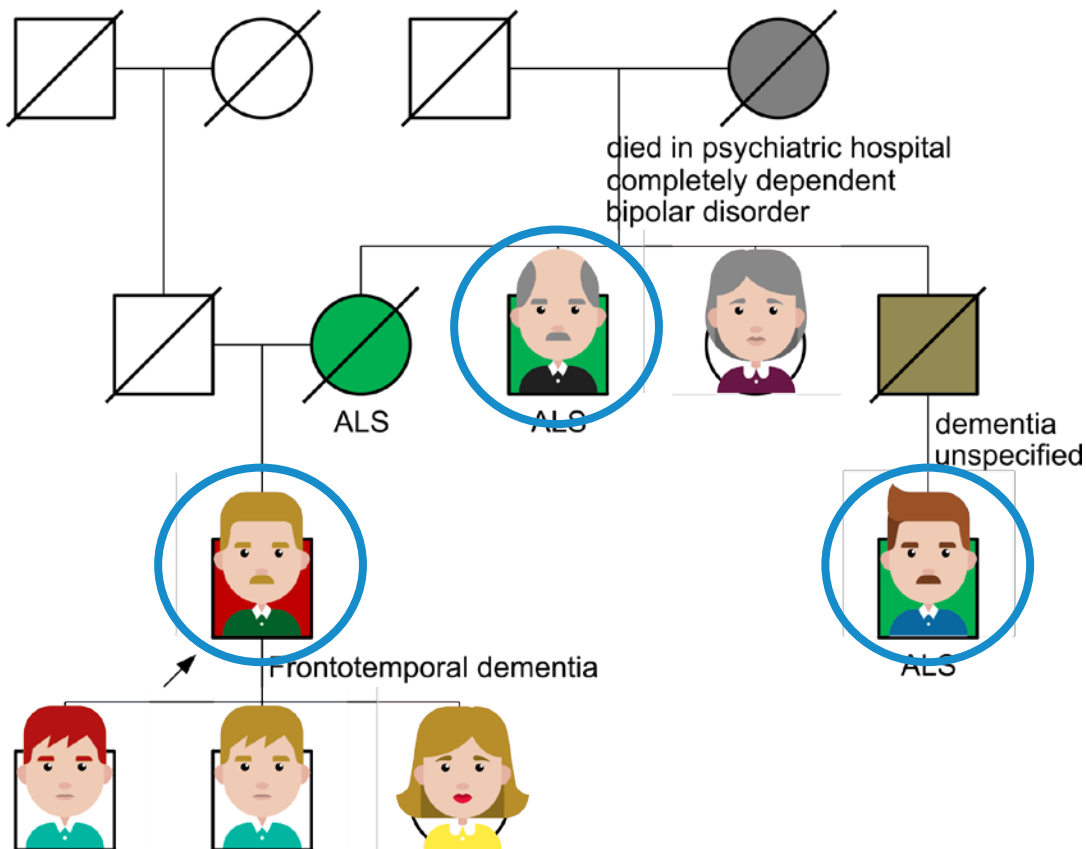
Discuss genetic testing with relative(s)

WAIT

I'll get tested first.



Healthy adult child



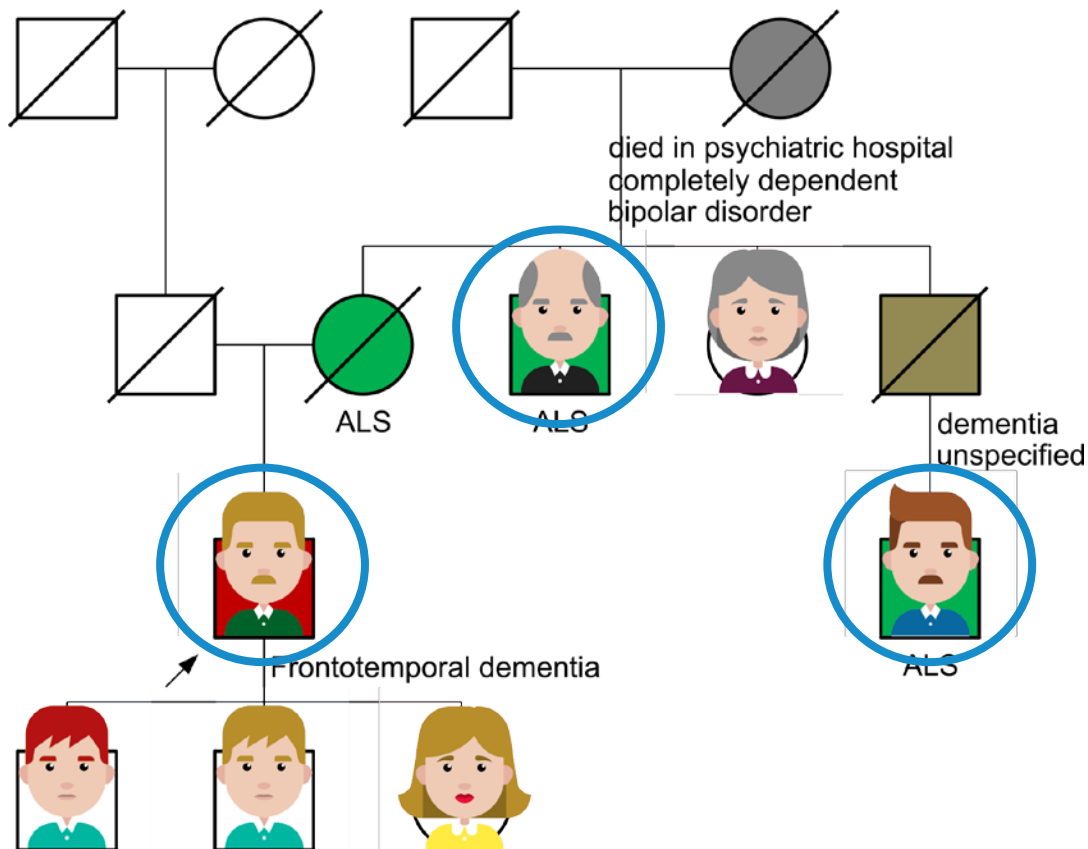
Discuss genetic testing with relative(s)



*People with symptoms should test first*



*Testing is still helpful, even when healthy adult children don't test*





Contact a  
genetics  
professional

[www.rarediseasesnetwork.org/cms/artfl](http://www.rarediseasesnetwork.org/cms/artfl)



Longitudinal Evaluation of  
Familial Frontotemporal  
Dementia Subjects

LEFFTDS



[www.nsgc.org](http://www.nsgc.org)

National Society of Genetic Counselors  
Celebrating 40 Years  
1979 - 2019

ABOUT US EVENTS PUBLICATIONS NEWSROOM EDUCATION CAREERS BECOME A MEMBER

NEWS & INSIGHTS  
POLICY & PUBLICATIONS  
ABOUT GENETIC COUNSELORS  
MEMBER CENTER  
FIND A GENETIC COUNSELOR

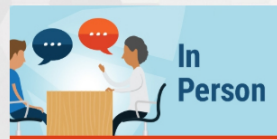
## FIND A GENETIC COUNSELOR

The Find a Genetic Counselor directory offers access to over 3,300 genetic counselors (US and Canada).

Check with your insurance company to verify coverage of genetic counseling, testing and authorized providers. For more information, visit [aboutgeneticcounselors.com](http://aboutgeneticcounselors.com).

To start your search, tell us **how you would prefer to meet with a genetic counselor**.

- Searches of the "In Person" directory will show genetic counselors who meet patients in a designated location and in person. Searches can be limited to a given location.
- Searches of the "By Phone" directory will show genetic counselors who meet with patients via phone, video conferencing and other virtual methods.



In  
Person



By  
Phone



Consider  
participating in  
research



**A·R·T·F·L**

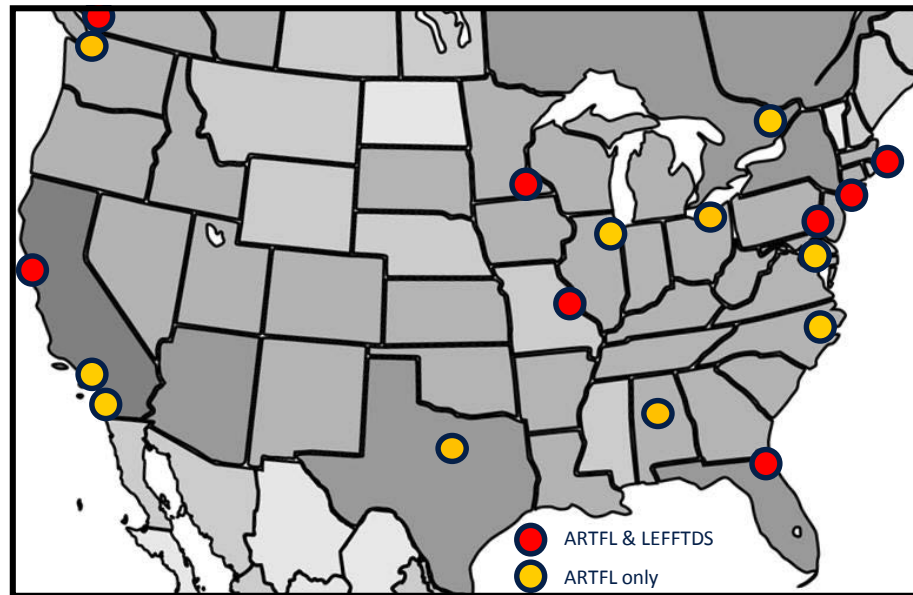
Advancing Research & Treatment for  
Frontotemporal Lobar Degeneration

Longitudinal Evaluation of  
Familial FrontoTemporal  
Dementia Subjects

LEFFTDS



[www.rarediseasesnetwork.org/cms/artfl](http://www.rarediseasesnetwork.org/cms/artfl)  
[www.clinicaltrials.gov](http://www.clinicaltrials.gov)  
[www.ftdregistry.org](http://www.ftdregistry.org)



# Thanks!



UCSF Memory and Aging Center