



Frontotemporal Degeneration (FTD)

Misdiagnosed and Misunderstood

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What is Dementia?

Dementia is a general term for brain degeneration that interferes with day-to-day life. Symptoms can include:

- Loss of cognitive functioning—thinking, problem solving and reasoning
- Loss of memory: short or long-term
- Inappropriate or socially unacceptable behavior
- Change in personality
- Difficulty with physical movement

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Today's Objectives

Learners will be able to:

- 1 Describe how FTD differs from other dementias
- 2 Define the FTD subtypes as well as common symptoms
- 3 List supports and services to assist families

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The Dementia Umbrella



Dementia is a general medical term for changes that can include problems with thinking, language, memory, behavior and more.

Some of the most common causes of dementia are:

- Alzheimer's Disease
- Lewy Body Dementia
- Frontotemporal Degeneration (FTD)
- Vascular Dementia
- Mixed Dementias

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My Story Is Personal, But I'm Not Alone



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Dementia estimates and hallmarks

Alzheimer's Disease <ul style="list-style-type: none"> • 5.8 million people in the U.S. • Memory loss is the most common early symptom • Amyloid plaques and tangles 	Vascular Dementia <ul style="list-style-type: none"> • ~10-15% of all dementias are vascular only • ~40% of all dementias have a vascular component • Caused by stroke(s) or other cerebrovascular damage
Lewy Body Dementia <ul style="list-style-type: none"> • 1.4 million people in the U.S. • Typically involves motor symptoms in addition to cognitive and behavioral changes • Lewy Body protein deposits on nerve cells 	Frontotemporal Degeneration <ul style="list-style-type: none"> • ~60,000 people in the U.S. but commonly misdiagnosed • Most common forms involve changes in language or behavior • Younger age of onset • TAU, TDP-43 and FUS

Mixed dementia ~50%

Alzheimer's Association 2020; Arslanalp et al. 2020; Knopman, G. 2019; Schneider, J. et al. 2020

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FTD Overview and Specific FTD Disorders

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What is FTD?

- A type of dementia caused by a family of disorders
- Leading cause of dementia in those younger than 60
- Personality, speech, and movement are common symptoms
- Takes an average of 3.6 years to achieve diagnosis
- Large caregiver burden due to age of onset and symptomology

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Brain Imaging Comparisons

Image from: Heiss, Wolf-Dieter & Rosenberg, Gary & Thiel, Alexander & Berlot, Rok & Heuck, Jacques. (2016). Neuroimaging in vascular cognitive impairment: A state-of-the-art review. BMC Medicine. 14. 10.1186/s12916-016-0725-0.

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What Happens Where?

Frontal Lobes

Prefrontal cortex

- Reasoning, decision making
- Control of behavior
- Executive functions (Planning, organizing)
- Problem solving
- Attention, concentration
- Emotional control
- Safety awareness
- Initiating action
- Physical movement

Temporal Lobes

Processing sensory information

Right Lobe

- Inhibition of speech
- Visual memory, pictures, shapes and faces, art

Left Lobe

- Verbal memory
- Understanding words and names
- Sorting new information

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Brief History of FTD

- 1892 Arnold Pick described first FTD patient
- 1911 Alzheimer described pathology: Pick bodies

Long period of little interest, then:

- 1993 + Clinical Criteria for Behavioral Variant FTD
- 1997 Abnormal tau protein: tau, FTDP-17, MAPT gene
- 2006 TDP-43 protein and progranulin (GRN) gene
- 2008 – 2009 Expanded genetics and path: TARDBP gene, FUS
- 2011 C9orf72 gene – Link between ALS and FTD identified
- 2015 Multicenter funding for clinical trials in FTD

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FTD: A Cluster of Complex Disorders

also called: frontotemporal dementia, frontotemporal lobar degeneration or Pick's Disease

<p>Progressive Behavior/Personality Change</p> <p>Behavioral Variant FTD (bvFTD)</p>	<p>Progressive Language Change</p> <p>Primary Progressive Aphasia (PPA)</p> <p>Subtypes:</p> <ul style="list-style-type: none"> Nonfluent/Agrammatic Variant Semantic Variant Logopenic Variant 	<p>Progressive Motor Function/Movement Change</p> <p>Corticobasal Syndrome (CBS/CBD)</p> <p>Progressive Supranuclear Palsy (PSP)</p> <p>FTD + Motor Neuron Disease (ALS-FTD)</p>
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Common Symptoms of bvFTD

Apathy	Disinhibition	Deficits in Executive Function Skills
Loss of Empathy	Changes in eating habits or diets	Compulsive Ritualistic Behavior

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FTD-ALS

The discovery in 2011 that the *C9orf72* gene mutation can cause both FTD and amyotrophic lateral sclerosis (ALS) has transformed a long-held belief that ALS is 'purely' a movement disorder and that FTD is 'purely' a cognitive or behavioral form of dementia.



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Signs and Symptoms of Primary Progressive Aphasia (PPA)

Nonfluent/Agrammatic	Semantic	Logopenic
<ul style="list-style-type: none"> • Agrammatism – omitting words, errors in use of word endings, verb tense and pronouns • Difficulty swallowing 	<ul style="list-style-type: none"> • Anomia – inability to recall names of objects • Difficulty reading & writing words that don't follow pronunciation or spelling rules 	<ul style="list-style-type: none"> • Difficulty with finding the right word when speaking • Difficulty with repeating phrases

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Impact on Families

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FTD Movement Disorders

Corticobasal Syndrome

- *Asymmetric parkinsonism*, changes often begin on one side of the body
- Hand may lift on its own (alien limb phenomena)
- Jerking movements
- Uncoordinated movements
- Gait and balance problems

Progressive Supranuclear Palsy

- *Changes in eye movements (slowed or restricted vertical eye movements)*
- Blank or surprised-looking stare
- *Frequent falling*
- Neck stiffness or posturing
- Slowness or shuffling gait
- Parkinsonism
- Sometimes tremors but not always
- Slurred speech, trouble swallowing

Cognitive Symptoms possible with both disorders

- *Trouble with calculations*
- *Word finding difficulty, shorter sentences*
- *Apathy*
- *Impulsivity*

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Economic & Social Costs

\$99k
↓
\$50k

Twelve months before an FTD diagnosis, most families reported a household income in the \$75,000–\$99,000 range. Twelve months after diagnosis, income fell as much as 50 percent.

FTD vs ALZ

Overall, families dealing with FTD face an economic burden of around **\$120,000** each year — roughly twice the economic burden of Alzheimer's.

37%

of FTD caregivers said they stopped working post-diagnosis.

58%

of respondents said that FTD caused their loved ones to make poor financial decisions.

*Published in the scientific journal *Neurology*, 11/14/17

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Kids & Teens

- Young onset diagnosis very often means that kids or teens may be home
- Caregivers need to balance the needs of the children with the person living with FTD
- Children become part of the care team

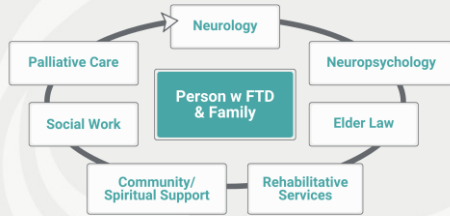


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Diagnosis, Treatment and Research

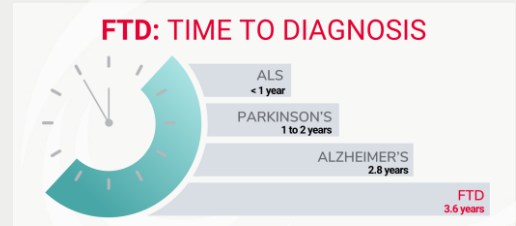
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Helping Families Build a Care Team



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Journey to Diagnosis



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Support

Support groups provide a resource, an outlet, and a place to collect and share your thoughts with others who understand

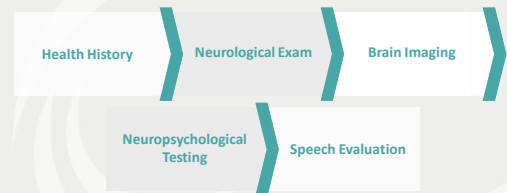
www.theaftd.org



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What to do if you see Warning Signs?

Getting a Diagnosis:



<https://www.theaftd.org/what-is-ftd/research-and-medical-centers/>

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FTD Treatments & Therapeutic Interventions

There is hope with current research and research participation

- *Currently: No disease altering treatment to stop or slow the disease*
- Active research is looking for treatments and effective therapies:
 - Learn more: www.theaftd.org/research-clinical-trials/featured-studies/
- There are effective treatments that can manage symptoms:
 - Off-label use of medications
 - Supportive therapies: Speech Therapy, Physical Therapy, Occupational Therapy, Support Groups, Palliative care, Dementia Friendly spaces

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FTD Research Opportunities

Scientists are closer than ever to new therapies and diagnostic tools for FTD and we all have a role to play in helping the research succeed.



- Many FTD clinical studies and trials are underway, seeking participants.
- Studies include experimental treatments to target FTD *symptoms* as well as potential *disease-modifying* treatments for people with sporadic and familial FTD.
- Studies also include *naturalistic/observational* studies to map the course of FTD.
- Some studies do not require the participant to know their genetic status.

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Is FTD Inherited?

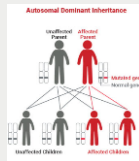
Familial FTD - Approximately 40% of people with FTD have a family history of dementia, a major psychiatric condition, or progressive changes in movement.

Genetic FTD - A subset of familial FTD cases are caused by a variant in a single gene inherited from a parent

Three genes cause the majority of genetic FTD:

- C9orf72 (FTD-ALS)
- GRN
- MAPT

Sporadic - For most people with FTD no one else in the family has FTD and relatives of someone diagnosed do not have an increased risk.



AFTD strongly recommends considering genetic counseling as a first step to answer questions about genetic status. For more info: www.theaftd.org/ftd-genetics/what-causes-ftd/

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Where to Learn More



Sign up for the FTD Disorders Registry: ftdregistry.org

- Co-founded by two non-profits, the Registry is a resource for persons diagnosed, families, and caregivers to participate in research.
- Healthcare providers and others may sign up to receive communications.
- The Registry provides one location to learn about research participation opportunities and share their stories to inform research design.
- Participants' personal information is NEVER shared.

Sign up for AFTD newsletters to stay informed about research opportunities and progress: theaftd.org

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Genetic Counselor

- Answer Questions
- Provide Disease Education Information
- Provide Emotional Support
- Provide Guidance
- Provide tools and support in discussing risk and or status to family



AFTD Resources

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AFTD – From Hope to Action

AFTD envisions a world with compassionate care, effective support, and a future free of FTD.

Our mission is to improve the quality of life of people affected by FTD and drive research to a cure. We work every day to advance:

- Research
- Education
- Awareness
- Advocacy
- Support



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How to Get Involved



Share FTD information and increase awareness



Raise funds in support of AFTD's mission



Join AFTD's Volunteer Network
theaftd.org/get-involved/volunteers-network



Become an advocate for change!

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Resources from AFTD


HelpLine info@theaftd.org
866-507-7222

Websites www.theaftd.org
www.aftdkidsandteens.org

Publications *Help & Hope*
Partners in FTD Care
The Doctor Thinks It's FTD. Now What?
What About the Kids?
Understanding the Genetics of FTD
Walking with Grief: Loss and the FTD Journey

Grants Respite, Travel, Quality of Life

Support National and regional on-line and local in-person groups
theaftd.org/living-with-ftd/aftd-support-groups



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Stay Connected with AFTD Resources

Scan to sign up for AFTD's newsletters



Contact AFTD's HelpLine
info@theaftd.org
866.507.7222

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AFTD Resources for Medical and Care Professionals

- Partners in Care quarterly publication focusing on a specific issue or topic
 - [Partners in FTD Care](http://Partners.in.FTD.Care) | [AFTD \(theaftd.org\)](http://AFTD.theaftd.org)
- Clinical presentations of symptoms:
 - [Clinical Presentations](http://Clinical.Presentations) | [AFTD \(theaftd.org\)](http://AFTD.theaftd.org)
- Webinars and Annual Conference
- AFTD Helpline: 1-866-507-7222 or info@theaftd.org

For additional info, go to [For Health Professionals](http://For.Health.Professionals) | [AFTD \(theaftd.org\)](http://AFTD.theaftd.org) or contact the AFTD Helpline.



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Questions?

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