Frontotemporal Degeneration: Misdiagnosed and Misunderstood
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Learning Objectives
• Learners will be able to define FTD and its subtypes
• Learners will understand the lived experience and be able to reference supports for those diagnosed and families
• Learner will be able to describe 3 symptoms unique to FTD and strategies for management

Your questions and engagement, welcome throughout!

My Families Experience with FTD
Deb Scharper
AFTD Ambassador
Dementia Overview

Dementia

“Dementia is the loss of cognitive functioning—thinking, remembering, and reasoning—and behavioral abilities to such an extent that it interferes with a person’s daily life and activities.” - NIH National Institute on Aging

- Dementia describes level of impairment in functioning
- Likelihood of developing dementia increases with age
- Not a specific disease

Types of Dementia

Alzheimer’s Disease
- Affects 60-80% of all dementias
- 5.8 million people in the U.S.
- Memory loss is the most common early symptom

Lewy Body Dementia
- Affects ~20% of all dementias
- 1.4 million people in the U.S.
- Typically involves motor symptoms in addition to cognitive and behavioral changes

Vascular Dementia
- Affects 10-15% of all dementias
- 40% of all dementias have a vascular component
- Caused by stroke(s) or other cerebrovascular damage

Frontotemporal Degeneration
- Affects ~50,000-60,000 people in the U.S.
- Most common forms involve changes in language or behavior
- Younger age of onset

Brain Imaging Comparisons

FTD Symptoms vs Alzheimer's Symptoms

Brief History of FTD

1892 Arnold Pick described first FTD patient
1911 Alzheimer described pathology: Pick bodies
Long period of little interest, then:
1993 Epidemiology, Clinical Criteria
1997 Abnormal tau protein: tau, FTDP-17, MAPT gene
2001 TDP-43 protein and progranulin (GRN) gene
2008 Expanded genetics and path: TARDBP gene, FUS
2011 C9orf72 gene – Link between ALS and FTD identified
2015 Multicenter funding for clinical trials in FTD
What is FTD?

FTD: A Cluster of Complex Disorders
also called: frontotemporal dementia, frontotemporal lobar degeneration or Pick's Disease

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<td>Semantic Variant</td>
<td>FTD + Motor Neuron Disease (ALS-FTD)</td>
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<td>Logopenic Variant</td>
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FTD Spectrum

FTD is a spectrum of diseases
**FTD and the Brain**

**Progressive Behavior/Personality Change**
- Behavioral Variant FTD (bvFTD)
- Frontotemporal dementia
- Most common form of FTD

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

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**FRONTAL LOBE**
- Prefrontal cortex

**Signs and Symptoms of Behavioral Variant FTD/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 and the Brain**

**Progressive Language Change**
- Primary Progressive Aphasia (PPA)

**Subtypes:**
- Nonfluent/Agrammatic Variant
- Semantic Variant
- Logopenic Variant

**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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**TEMPORAL LOBES**
- Processing sensory information
Signs and Symptoms of Primary Progressive Aphasia (PPA)

**Nonfluent/Agrammatic**
- Agrammatism – omitting words, errors in use of word endings, verb tense and pronouns
- Difficulty swallowing

**Semantic**
- Anomia – inability to recall names of objects
- Difficulty reading & writing words that don’t follow pronunciation or spelling rules

**Logopenic**
- Difficulty with finding right word when speaking
- Difficulty with repeating phrases

FTD and the Brain

Progressive Motor Function/ Movement Change
- Corticobasal Syndrome (CBS/CBD)
- Progressive Supranuclear Palsy (PSP)

- Voluntary movement
- Cognition
- Emotional processing

**Basal Ganglia & Brain Stem**

FTD Movement Disorders

**Corticobasal Syndrome**
- Movement Symptoms
  - Asymmetric parkinsonism
  - Dysmetric limb posturing
  - Hand may lift on its own (Alien limb phenomenon)
  - Jerking movements (embryonic)
  - Incoordinated movements (apraxia)
  - Gait and balance problems

- Cognitive Symptoms
  - Trouble with calculations
  - May not pay attention to one side of the body or world (neglect)
  - Word finding difficulty, shorter sentences
  - Apathy
  - Impulsivity

**Progressive Supranuclear Palsy**
- Movement Symptoms
  - Frequent falling
  - Changes in eye movements (blurred or restricted vertical eye movements)
  - Neck stiffness or posturing
  - Blank or surprised-looking stare
  - Slowness
  - Shuffling gait
  - Parkinsonism
  - Sometimes tremors but not always
  - Slurred speech, trouble swallowing
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.

Balancing a Dual Diagnosis

ALS
- Muscle weakness
- Muscle spasticity
- Fasciculations
- Twitching
- Muscle cramps

FTD
- Personality
- Language
- Behavior

Managing Symptoms
Person Centered Care

“Person-centered care is individualized compassion, empathy, respect and responsiveness to the needs, values, and expressed desires of each individual person”

- Person values guide clinical decisions
- Supports independence
- Promotes respect
- Honors choice

Lack of Insight (Anosognosia)

Profound lack of emotional concern about their disease and its impact on others

- The person “appears” fine
- Symptoms can be small in the beginning
- Caregivers notice “somethings off”
- Financial mismanagement
- Impulsive
- Poor safety awareness
- Narcissistic behavior

Modify the Environment

- Minimize Noise and Stimulation
- Reduce Clutter and Distractions
- Simplify Social Situations

Managing Expectations

• The behavior is not on purpose
• Is the behavior hurting anyone?
• Modify public outings to reduce the opportunity for inappropriate interactions

Diagnosis and Research

Journey to Diagnosis

FTD: TIME TO DIAGNOSIS
What to do if you see Warning Signs?

Getting a Diagnosis

- Health History
- Neurological Exam
- Brain Imaging
- Neuropsychological Testing
- Speech Evaluation

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

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/

Genetic Counselor

- Answer Questions
- Provide Disease Education
- Provide Emotional Support
- Provide Genetic Information

theaftd.org/ftd-genetics/genetic-counseling/
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.

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

Impact on Families
Overall, families dealing with FTD face an economic burden of around $120,000 each year — roughly twice the economic burden of Alzheimer’s.

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

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.

- 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

Building a Care Team

- Neurology
- Neuropsychology
- Social Work
- Elder Law
- Community/Spiritual Support
- Palliative Care
- Rehabilitative Services

Person w FTD & Family
Support
Support groups provide a resource, an outlet, and a place to collect and share your thoughts with others who understand.
www.theaftd.org

AFTD Resources

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
• Awareness
• Support
• Education
• Advocacy
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-support-groups

Partners in FTD Care

theaftd.org/for-health-professionals/partners-in-ftd-care

Stay Connected with AFTD Resources

Scan to sign up for AFTD’s newsletters

Contact AFTD’s HelpLine
  info@theaftd.org  866.507.7222
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!

Questions ???

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