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Early-Onset Familial Alzheimer Disease

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Introduction

An Alzheimer's diagnosis is an overwhelming and devastating diagnosis for both patients and families. Now imagine if this diagnosis was made at age 55. Instead of looking forward to retirement and grandchildren, these people are preparing for a debilitating disease that will rob them of their memory, cognitive and functional abilities. Early-onset familial Alzheimer's disease (EOFAD) affects people before the age of 65. Although extremely rare, some exhibit symptoms as early as in their 30s.

One of the things most frustrating about this disease is so little is known about its cause. That and the fact there is no cure. There is research which demonstrates a connection between three gene mutations and the incidence of EOFAD. Knowledge of the potential cause behind their disease may help some patients and families better understand and cope with this disease.

EOFAD is rare. It is estimated only five percent of all Alzheimer's diagnoses can be attributed to early onset. In 2011, the Alzheimer's Association estimated 200,000 persons were afflicted with this disease in the United States (Barber, 2012). However, these numbers include both familial and sporadic forms. It is likely EOFAD accounts for less than one percent of all cases of Alzheimer's disease (Orphanet, 2009).

Pathophysiological Processes

❖ Signs and Symptoms

EOFAD is a progressive dementia that affects cognition, behavior and functional abilities. EOFAD progresses the same way as late-onset Alzheimer's disease however it affects patients at an earlier age, has definite family history, various non-cognitive neurological signs and symptoms, and is thought to have a more aggressive course and shorter survival time (Panegyres, & Huei-Yang, 2013)

The seven stages of Alzheimer's disease:

Stage 1: No impairment. No symptoms of dementia, normal function

Stage 2: Very mild cognitive decline. No symptoms of dementia detected but person may forget familiar words or location of everyday objects.

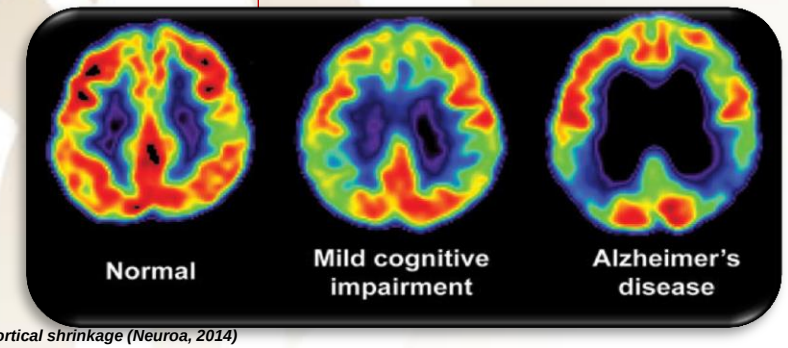
Stage 3: Mild cognitive decline. Memory or concentration problems may be detected. Problems may include trouble planning or organizing, greater difficulties performing tasks in social or work settings.

Stage 4: Moderate cognitive decline. Specific symptoms can be identified such as forgetting recent events and/or own personal history, becoming moody or withdrawn, increasing difficulty with complex tasks.

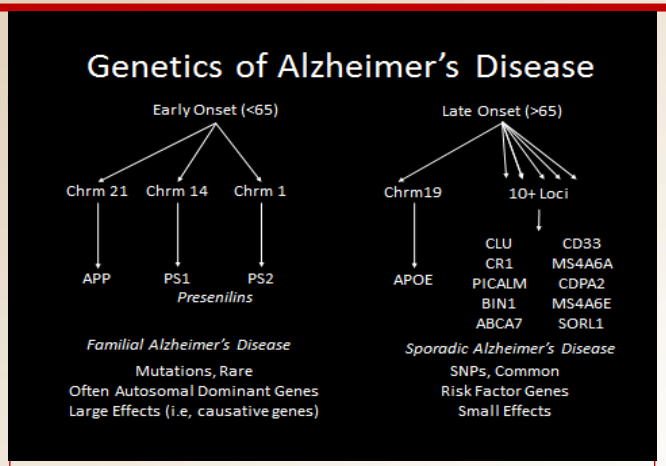
Stage 5: Moderately severe cognitive decline. Memory gaps are evident, assistance is needed with day to day activities. Confusion may exist about where they are or what day it is, trouble with mental arithmetic.

Stage 6: Severe cognitive decline. Extensive assistance is needed with daily activities, memory worsens, personality changes may occur. trouble remembering names of spouse or caregiver, changes in sleep patterns, frequent trouble with bowel and bladder control, may wander and become lost.

Stage 7: Very severe cognitive decline. Ability to respond to environment is lost. Need maximum assistance with personal care, reflexes abnormal, swallowing impaired (Alzheimer's Association, 2014a)



Cortical shrinkage (Neuroa, 2014)



Gene mutation differences in EOFAD and late-onset Alzheimer's (Neuroa, 2014)

❖ Underlying Pathophysiology

EOFAD is an inherited autosomal dominant disease. Scientists have discovered 3 rare deterministic genes that have been identified as a cause the disease: amyloid precursor protein (APP) on chromosome 21 accounts for 10-15% of EOFAD, presenilin-1 (PS1) on chromosome 14 accounts for 75-80% of EOFAD, and presenilin-2 (PS2) on chromosome 1 accounts for <5% of EOFAD (Wu et al., 2012). Each of these mutations contributes to the breakdown of APP. The result of this breakdown process is the formation of harmful beta-amyloid protein fragments that are the main components of plaques (National Institute on Aging, 2014). The plaques build up and interrupt communication between neurons. These amyloid plaques are a hallmark of the disease, in addition to cerebral cortical atrophy and intraneuronal neurofibrillary tangles (Bird, 2012). These intraneuronal tangles cause damage to brain cells and synapses. The cerebral cortex and hippocampus shrinks and the ventricles enlarge.

People who inherit an early onset Alzheimer's mutation have a nearly 100% chance of developing the disease. Each child of a parent with an early-onset mutation has a 50/50 chance of inheriting the disease. EOFAD is extremely rare, an estimated 1% or less of Alzheimer's cases are attributed these genes (Orphanet, 2009).

❖ Significance of Pathophysiology

Genetic testing and counseling can be offered to the rare families that have the known genetic mutation for EOFAD. However, since there are currently no treatments to prevent, cure or even slow the process of Alzheimer's, this testing would have little to no effect on medical treatment decisions (Alzheimer's Association, 2014b). It could, however, help families make decisions about financial matters, reproduction and career planning (Bird, 2012). Although not common, if the disease causing mutation has been identified in the family, prenatal testing can be done by DNA analysis of the fetal cells.

Implications for Nursing Care

In order to treat the cognition, behavior and functional abilities of EOFAD, both pharmacologic and nonpharmacologic interventions are needed. The top goals of treatment are focused on maintaining quality of life, ensuring a safe environment, maximize function in daily activities. Support and education for the patient and family is imperative. Encourage the preparation of a living will and/or durable power of attorney for health care (Alzheimer's Association, 2014c).

There are five FDA approved drugs that treat the symptoms of Alzheimer's disease (Alzheimer's Association, 2014c):

Generic	Brand	Approved for
donepezil	Aricept	all stages
galantamine	Razadyne	mild to moderate
memantine	Namenda	moderate to severe
rivastigmine	Exelon	all stages
tacrine	Cognex	mild to moderate

FDA approved Alzheimer's drugs

Conclusion

Alzheimer's disease is a dreadful and insidious disease to have regardless of age but it is especially hard when it affects individuals in the prime of their life. This disease robs people of their lives and leaves the remnants for the families and loved ones. Often times EOFAD is misdiagnosed, not recognized and inadequately managed. Dementia in the young is overshadowed by the aging population (Armari, Jarmolowicz & Panegyres, 2013). Because EOFAD is so exceptionally rare, it has not received much attention. Unlike other diseases, it lacks the visibility, funding and advocacy groups. Additionally, it has not been included in any clinical studies or drug trials (Strobel, 2014).

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