Fall 2014

Early-Onset Familial Alzheimer Disease

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Recommended Citation
Murphy, Laura J., "Early-Onset Familial Alzheimer Disease" (2014). MSN Student Scholarship. Paper 27.
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 people were affected 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 (Orphanev, 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, & Hsieh-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 severe cognitive decline. Specific symptoms can be identified such as forgetting recent events and/or personal history, becoming moody or withdrawn, increased 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, personal 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).

Implications for Nursing Care

In order to treat the cognition, behavior and functional abilities of EOFAD, both pharmacological and nonpharmacological interventions are needed. The top goals of treatment are focused on maintaining quality of life, ensuring a safe environment, maximizing function in daily activities. Support and education for the patient and family is imperative. Encourage the preparation of a 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):

- Donepezil
- Galantamine
- Rivastigmine
- Tacrine
- Memantine

Genetics of Alzheimer’s Disease

EOFAD is an inherited autosomal dominant disease. Scientists have discovered 3 rare deterministic genes that have been identified as a cause the disease:

- Apolipoprotein E (ApoE), chromosome 19, accounts for 50-60% of EOFAD
- Presenilin-1 (PSEN1), chromosome 14, accounts for 15-20% of EOFAD
- Presenilin-2 (PSEN2), chromosome 1, accounts for 3% of EOFAD (Wu et al., 2012).

There are four possible cases for early familial Alzheimer’s disease:

- One mutation has been identified in the family
- The patient was diagnosed with Alzheimer’s disease after age 65
- One mutation has been identified in the family
- The patient was diagnosed with Alzheimer’s disease before age 65

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 (PSEN1) on chromosome 14 accounts for 15% of EOFAD and presenilin-2 (PSEN2) on chromosome 1 accounts for 5% of EOFAD (Wu et al., 2012).

Each of these mutations contributes to the breakdown of APP. In people with EOFAD, these mutations lead to higher levels of two proteins that are a hallmark of the disease, in addition to cerebral cortical atrophy and intraneuronal neurofibrillary tangles (Bird, 2012). These amyloid plaques are a hallmark of the disease, in addition to cerebral cortical atrophy and intraneuronal neurofibrillary tangles (Bird, 2012). These amyloid plaques or cerebral cortical atrophy and intraneuronal neurofibrillary tangles cause damage to brain cells and synapses. The cerebral cortex and hippocampus shrink and the ventricles enlarge.

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 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.

Signs and Symptoms

- Amnesia
- Agitation
- Aggression
- Anxiety
- Appetite loss
- Apathy
- Confusion
- Depression
- Fatigue
- Hallucinations
- Memory loss
- Night terrors
- Sleep disorders
- Unusual behavior

Genetics of Alzheimer’s Disease

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References


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 had a small amount of 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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Cortical shrinkage (2014)