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Early-Onset Familial Alzheimer Disease

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An Alzheimer’s disease 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) is the diagnosis of people under the age of 65. Although extremely rare, some exhibit symptoms as early as in their 30s. Although extremely rare, some exhibit symptoms as early as in their 30s. It is likely persons were afflicted with this disease but not diagnosed. The Alzheimer's Association estimated 200,000 (5% of all Alzheimer's disease) over the age of 65. 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: Milder 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 Specific symptoms can be identified such as forgetting recent events or one’s own personal history, becoming moody or withdrawn, increasing difficulty with complex tasks.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 one is or what day it is, with 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 wanter and become less active.
Stage 7: Very severe cognitive decline. Ability to respond to environment is lost. Need maximum assistance with personal care, requires abnormal, swallowing impaired (Alzheimer’s Association, 2014a). Genetic mutation differences in EOFAD and late onset Alzheimer’s disease (Alzheimer’s Association, 2014b).  There are five FDA approved drugs that treat the symptoms of Alzheimer’s disease (Alzheimer’s Association, 2014c).

Underlying Pathophysiology

EOFAD is an inherited autosomal dominant disease. Scientists have discovered 3 rare deterministic genes that have been identified as a cause of the disease: amyloid precursor protein (APP), presenilin-1 (PS1) and presenilin-2 (PS2). 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 impair communication between neurons. These amyloid plaques are a hallmark of the disease, in addition to cerebral atrophy and intraneuronal neurofibrillary tangles (Bird, 2012). These intraneuronal tangles cause damage to brain cells and synapses. The cerebral cortex and hippocampus shrink and the ventricles enlarge.

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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). 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, 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).

References

Genetics of Alzheimer’s Disease
Early-Onset Familial Alzheimer Disease
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genetics-of-alzheimer’s-disease-familial-early-onset-and-sporadic-late-onset.org

Significance of Pathophysiology

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).

Underlying Pathophysiology
EOFAD is an inherited autosomal dominant disease. Scientists have discovered 3 rare deterministic genes that have been identified as a cause of 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 impair communication between neurons. These amyloid plaques are a hallmark of the disease, in addition to cerebral atrophy and intraneuronal neurofibrillary tangles (Bird, 2012). These intraneuronal tangles cause damage to brain cells and synapses. The cerebral cortex and hippocampus shrink and the ventricles enlarge.

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FDA approved Alzheimer’s drugs

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Introduction
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Pathophysiological Processes
Signs and Symptoms
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Genetics of Alzheimer’s Disease
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, 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).

Implications for Nursing Care
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