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Laura J. Murphy
Otterbein University, laura.murphy@otterbein.edu

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Early-Onset Familial Alzheimer Disease
Laura J. Murphy, BSN, RN-BC
Otterbein University, Westerville, Ohio

Introduction

An Alzheimer’s diagnosis is an overwhelming and devastating experience 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 a force to be reckoned with. Although extremely rare, some exhibit symptoms as early as in their 30s.

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, & Hue-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 Specific symptoms can be identified such as forgetting recent events and/or own personal history, becoming moody or withdrawn, increasing difficulty with complex tasks.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, refluxes abnormal, swallowing impaired (Alzheimer’s Association, 2014a).

Implications for Nursing Care

In order to treat the cognition, behavior and functional abilities of EOFAD, speech pathology and non-pharmacological 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 living will and/or durable power of attorney for healthcare (Alzheimer’s Association, 2014c).

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 hippocampal 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.

References


FDA approved Alzheimer’s drugs


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