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Amanda Pack

Otterbein University, amanda.pack@otterbein.edu

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Early Onset Alzheimer’s Disease in Individuals with Down Syndrome: Explored Theories of Pathophysiology

Amanda Pack, BSN, RN
Otterbein University, Westerville, Ohio

Introduction

This presentation is based on a focus on some theories related to the pathophysiology of early-onset Alzheimer’s Disease (AD) in individuals with Down Syndrome (DS). Down Syndrome, a genetic condition where an individual has a partial or full extra chromosome 21, is the most common genetic cause of intellectual disability in the United States (National Down Syndrome Society, 2012). Average life expectancy for individuals with DS is typically lower than that of the general population. The prevalence of early-onset AD is characterized by having AD by the age of 65. The purpose of this presentation is to aid practitioners in having a better understanding of the disease process in order to provide appropriate screening and diagnosis for this growing population.

Signs and Symptoms

Alzheimer’s (2014) describes the early presentation of AD as difficulty in remembering newly learned information, with advances stages potentially presenting with decreased appetite, decreased interest in being sociable, conversations, or ball, Holland, Watson and Huppert (2010) also explore the decreased enthusiasm for usual activities irritability, uncooperativeness or aggression.

Pathophysiological Phenomena

Underlying Pathophysiology

AD is not completely understood, including in individuals with DS. Moreover there are some locations of amyloid plaques and neurofibrillary tangles, and there are several additional contributing factors that are associated with AD beyond that. It is known that individual with DS have a partial or full triplication of chromosome 21, as well as amyloid plaques and NFTs by the age of 40. The following are examples of suggested theories that could be explored in DS and early-onset AD beyond that. In addition to the amyloid plaques and NFTs, there are several other contributing factors must be explored.

1. Head et al. (2011) state that the amyloid plaques contain the amyloid Beta peptide that is derived from a longer precursor protein, amyloid precursor protein (APP), which is located on chromosome 21. Overexpression of the APP is suggested as the cause of amyloid plaques in individuals with DS.

2. Sermon et al. (2010) state that neuropathological changes that occur in AD include abundant amyloid plaques, neurofibrillary tangles along with neurit thief disturbances of the neocortical hyperphosphorylated tau that are accompanied by significant brain atrophy. Changes include loss of neuronal, neuritic, and synaptic elements. They also argue the neuroanatomical changes begin to accumulate in an early age and may persist in late stages of AD. Watson and Huppert (2011) state that “preclinical” stage of AD corresponds to the stage of cognitive change at an early age. A baseline assessment should be performed on the DS individual by the age of 35 with an annual assessment starting at age 40. Early signs of dementia and AD often go unnoticed in individuals with DS due to their atypical presentation of behavioral changes instead of memory impairment.

3. The further study of individuals with DS can hold the key to answer questions of the pathology of early-onset AD and AD in general. Further studies could also explore the potential effects of selective serotonin-norepinephrine inhibitors.

4. Every individual with DS will have AD altogether diagnostic criteria of amyloid plaques and NFTs by the age of 40. The triplication of chromosome 23 is that the brain has a greater prevalence of genetic alterations that contribute to this. It makes this evident that chromosome 23 is a primary factor that contributes to the development of AD. Further focused studies on the role that chromosome 21 plays in individuals with early onset AD can potentially be used to develop targeted drugs to stop slow down the process.

Conclusion

Every individual with DS will have AD altogether diagnostic criteria of amyloid plaques and NFTs by the age of 40. The triplication of chromosome 23 is that the brain has a greater prevalence of genetic alterations that contribute to this. This makes it evident that chromosome 23 is a primary factor that contributes to the development of AD. Further focused studies on the role that chromosome 21 plays in individuals with early onset AD can potentially be used to develop targeted drugs to stop slow down the process.

Brain Scan Comparison of Individuals with DS and AD, Ghose, 2011.

Implications for Nursing Care

Since it is known that all individuals with DS will have neuropathological features of AD, it is pertinent that health care professionals monitor for signs of cognitive change at an early age. A baseline assessment should be performed on the DS individual by the age of 35 with an annual assessment starting at age 40. Early signs of dementia and AD often go unnoticed in individuals with DS due to their atypical presentation of behavioral changes instead of memory impairment. Behavioral changes can also be attributed in the DS population as part of their developmental disability. Whitman, McBrien, and Bower (2010) developed a standardized screening checklist to help health care professionals know when to make a referral for a dementia assessment for individuals with DS and other developmental disabilities.

References


http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3156200/


