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Genetic risks of Alzheimer's disease – A Case Study

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***Abstract*— This case study involves collecting information from the affected individuals and the risk that the next generation will possibly have.**

Keywords—***APO e4 gene, amyloid plaques, vascular dementia***

I. INTRODUCTION

Alzheimer's disease (AD) or simply Alzheimer's is a type of dementia, more specifically a chronic neurodegenerative disease, which causes problems with memory, thinking and behavior. The cause of Alzheimer's disease is poorly understood [ref 5]. About 30% of the risk is believed to be genetic with many genes usually involved. Other risk factors include a history of head injuries, depression, or hypertension. The disease process is associated with plaques and tangles in the brain. The reason for these plaques is due to the errors in amyloid precursor protein (APP) processing. It was seen that APP located on chromosome 21, together with the people with trisomy 21 (Down Syndrome) who have an extra chromosome copy almost universally exhibit at least the earliest symptoms of AD by 40 years of age. Also, a specific isoform of apolipoprotein, APOE4, is a major genetic risk factor for AD. While apolipoproteins enhance the breakdown of beta amyloid, some isoforms are not very effective at this task (such as APOE4), leading to excess amyloid buildup in the brain. Symptoms usually develop slowly and get worse over time, becoming severe enough to interfere with daily tasks.

II. METHODOLOGY

In this particular case study, the information of three different patients has been collected who were suffering from alzheimer's. Not only the genetic factors but also the environmental factors play a key role leading to this condition. The three patients include a male aged 86 and two females aged 82 and 63.

Case 1:

The patient is a male. The age of the patient at the time of onset of this condition was 84. The patient's blood group was O+. His past history/medical history showed that he was diabetic and had already undergone a bi-pass surgery once. His family history included an elder brother already affected with alzheimer's and younger sister also is known to be affected. The patient was seen to be in the final stage of alzheimer's – vascular dementia. Age factor is one of the reasons for causing this condition as it is seen that all the siblings are seen to be affected with dementia after they cross an age of 82.

The patient was seen to be administered with various drugs such as neurobion, janumet/glycomet, risperidone etc. Basically, most of these are seen to be given to the patient to treat type II diabetes and some cardiovascular problems faced by the patient.

A component present in the medicine janumet, metformin, significantly increases the generation of both intracellular and extracellular ß – amyloid peptides, which is mediated by transcriptional up-regulation of ß-secretase, resulting in the elevated protein level (>40 peptides) and increased enzymatic activity (leading to the production of insoluble inclusion bodies, that lead to impairment of nerve function coordination resulting in memory loss). This is one potentially harmful consequence of using this widely prescribed antidiabetic drug (in elderly diabetic patients).

Case 2:

The second patient is a female. The age of the patient at the time of onset of this condition was 82. The patient's blood group was B+. The patient was seen to be in the second stage of alzheimer's – mild cognitive impairment. This person neither had any health issues nor any past medical issues but was still diagnosed with Alzheimer's at 82. This might be due to the environmental factors such as age and lifestyle. Symptoms possessed are similar to any other Alzheimer's patient, such as memory loss, difficulty in communication with others, absent mindedness, etc.

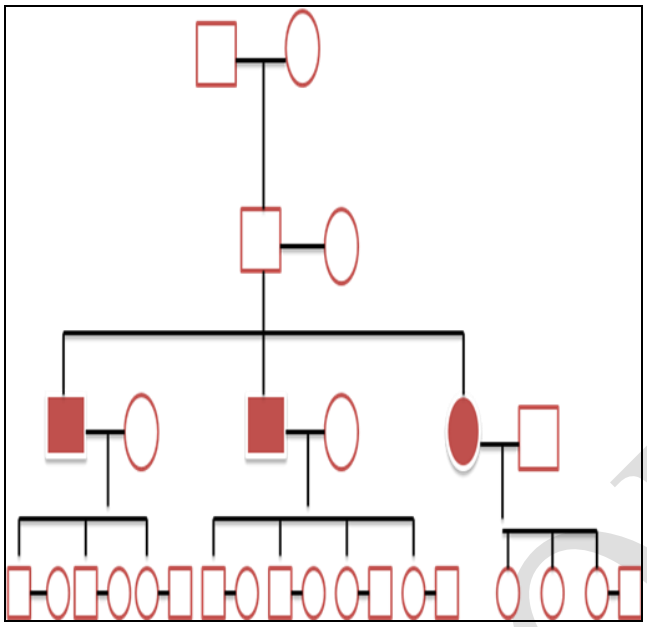
Case 3:

The third patient in this study is also a female. The age of the patient at the time of onset of this condition was 63. The patient's blood group was B+. The medical history indicates high blood pressure, diabetes, heart attack and paralysis. The patient was seen to be in the second stage of alzheimer's – mild cognitive impairment. The patient has diabetes (type II), survived heart attacks (two) and suffers paralysis. Regular injections of insulin are required by the patient (atleast 3 times a day). After paralytic attack, she was diagnosed with Alzheimer's. As mentioned earlier, people with diabetes and heart diseases are evidently more

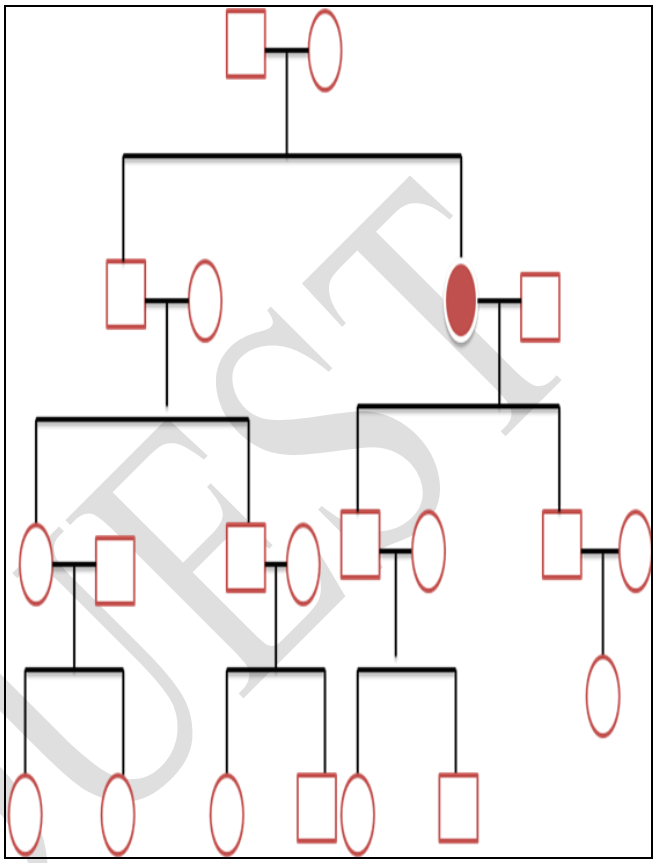
prone to the risk of getting Alzheimer's at a low age. Her medication was similar to the person in case 1, in addition drugs also included some antidepressants.

III. FIGURES

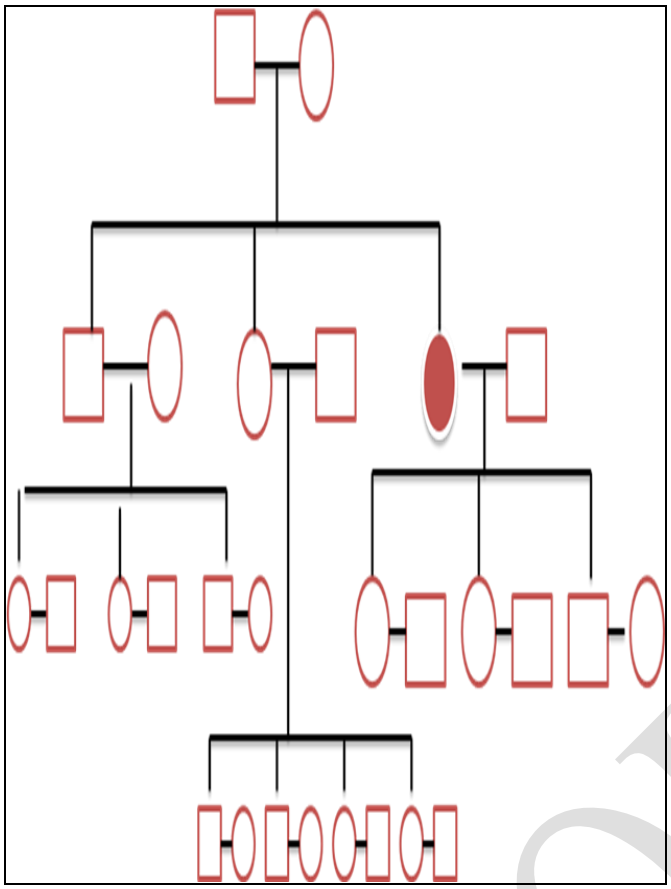
Case 1:



Case 2:



Case 3:



Results

We studied three cases and found that the risk of getting Alzheimer's increased as the age increased. In case study1, as all the siblings in generation III are affected, there is a high risk that the next generation will be getting affected with it. Whereas in the other case studies, environmental factors indicate the presence of the disease.

Discussion

A recent study published in JAMA psychiatry by Anette Ribe and colleagues showed a relationship between schizophrenia and onset of dementia (ref. 1). According to their study, by the age of 80, people with schizophrenia had a greater risk not only for developing dementia but also for developing it at a younger age (ref. 1). In fact 22.4% of individuals with schizophrenia who went on to develop dementia were diagnosed with it before the age of 65. Many also had clinical features seen in persons with Alzheimer's disease.

Brain cell connections and the cells themselves degenerate and die, eventually destroying memory and other important mental functions. Memory loss and confusion are the main symptoms. Our study has given a hint that genetic predisposition would cause the early onset of the disease.

Conclusion

A conclusion can be thus made that no cure exists as such for Alzheimer's, but medication and management strategies may temporarily improve symptoms of such patients.

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