SAQsTeam

CASE 6

A 73 year old woman was brought to neurological evaluation by her brother because of a 3 year history of memory impairment. She had completed high school and worked in a clerical position until her retirement in 1985. She had lived alone and maintained her own home and financial affairs since the death of her husband in 1980. The brother had begun to notice gradually worsening memory impairment and difficulty finding words, but the patient became

angry at the suggestion that she may have a progressive impairment. Others had noted decline in housekeeping and financial affairs, but she had no complaints.

1)What is the most likely diagnosis?

Alzheimer's disease.

2)What is the type of this disease?

Neurodegenerative disease

3)What are some investigations the doctor could do?

Although pathologic examination of brain tissue remains necessary for the definitive diagnosis of Alzheimer disease,

-clinical examination

-Radiological methods

allow accurate diagnosis in 80% to 90% of cases.

4)What is the cause of Alzheimer's disease?

Caused by abnormal accumulation of insoluble "protein aggregates" (Beta amyloid) that is resistant to normal degradation. and is cytotoxic and causes neuronal dysfunction and eventually neuronal death.

5)What is the pattern of incidence of this disease?

- Most cases are sporadic
- 5%-10% are familial

6)What are the major microscopic abnormalities in this disease?

- Neuritic plaques.
- Neurofibrillary tangles.
- Amyloid angiopathy (less specific.)

7)Mention 2 proteins that accumulate in a brain of Alzheimer patients?

1- Amyloid beta protein.

2- TAU protein.

8)What are the genetic factors that lead to Alzheimer disease?

- Mutation of Amyloid precursor protein (APP) gene on chromosome 21
- Mutation of presenilin-1(PS1)gene on chromosome 14
- Mutation of presenilin-2(PS2)gene on chromosome 1

all these three mutation above lead to early onset of familial disease.

- Mutation of Apolipoprotein E (ApoE) gene on chromosome 19 increases the risk and lower the age of onset of the disease.
- Mutation of SORL1 gene associated with late onset of Alzheimer disease.

9)Mention one syndrome associated with this disease?

Down syndrome (trisomy 21.)

10)What is the principal clinical manifestation of this disease?

Dementia.

11)What are the macroscopic features of brain of Alzheimer patient (gross pathology in autopsy) ?

- Cortical atrophy.
- Widening of the cerebral sulci .(in temporal, frontal and parietal lobes)

- Compensatory ventricular enlargement (hydrocephalus ex vacuo).

12)What is the first brain region usually affected in this disease?

Entorhinal cortex(EC).

13)What are the stains/lab routines used in studying the histopathology of this disease?

Silver staining or immunohistochemistry.

14)What are the risk factors of alzheimer disease?

- Age:Increased age is the greatest known risk factor for Alzheimer's.
- Family history: Those who have a parent, brother, sister or child with Alzheimer's are more likely to develop the disease.
- Genetics:
- APOE-e4 may be a factor in 20-25% of Alzheimer's cases.

15)What is the possible management for alZheimer's?

1-Treating cognitive symptoms.
2-Managing behavioral and psychological symptoms.
3-Monitoring Alzheimer's disease.
4-Regulating neurotransmitter activity (enhancing cholinergic function improve (symptoms).
5-Polyphenols such as flavonoids reduce proinflammatory responses.

16)What are your management goals with Alzheimer's patients?

1-Maintain quality of life.
2-Maximize function in daily activities.
3-Enhance cognition, mood and behavior.
4-Keep him in a safe environment.
5-Promote social engagement, as appropriate.

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