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A genetically-proven case of Huntington's disease in the Philippines

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Introduction: Huntington's disease (HD) is a rare, neurodegenerative disorder characterized by chorea,behavioral manifestations, and dementia.Its cause has been linked to abnormal expansion in a length of a CAG triplet repeat sequence in a gene on chromosome 4p, now called the Huntington gene (HD). The disease occurs in all racial groups but is most common in people of northern European origin.Studies revealed an incidence of 0.38 per 100,000 per year with a lower incidence in the Asian studies. The worldwide service-based prevalence of HD was 2.71 per 100,000 and overall prevalence conducted in Asia showed 0.40 per 100,000. A literature review revealed no previous reports of genetically-proven case of Huntington's disease in the Philippines.

Case description: A 30-year old Filipino male from Jolo, Sulu consulted to the hospital with the complaint of involuntary, brief, irregular, jerky movements of extremities. History started when the patient was 21 years old. There was gradual progression of symptoms which initially started as jerky, repetitive, purposeless movements of the head and shouldersuntil there was involvement of trunk and all extremities. He developed behavioral changes in the form of frequent outbursts of anger, loss of temper and irritability, and manifested with cognitive problems that he had to stop schooling. His father had the same manifestations when he was still alive. He has 2 paternal uncles and an aunt who also presented to the hospital with the same symptoms. On physical examination, he has random facial grimaces with intermittent protrusion of the tongue and irregular shoulder jerks with athethoid movement of the distal extremities. He has generalized random movement of the different parts of the body including truncal musculatures and has involuntary, brief, irregular, jerky movements that flow from proximal to distal extremities. The patient has parakinesias and motor impersistence. Cranial CT scan was done which showed atrophy of bilateral caudate nucleus. Genetic testing was done to our patient. Blood specimen was sent to Mayo Clinic and PCR based assay was utilized to detect CAG repeat expansions in exon 1 of the HTT gene. The CAG repeat of the patient was 53 revealing full penetrance.

Discussion: HD is rare among Asians and this is the first genetically-proven case in the Philippines. Since relatively uncommon, Huntington's disease can be devastating for patients and their families. This case illustrates the potential benefit of utilizing genetic testing and counseling to the other family members, and conducting further study on the ancestral place of the patient.

Frontal lobe involved in conceptual explicit and implicit memory

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Background: According to the multiply memory of system, memory can be classified into explicit memory and implicit memory; According to the processing theory of memory: memory is divided into conceptual and perceptual processing. And component process framework indicated that different brain areas may process with specific memory components. But, at present only a tiny part of the neuropsychological studies of frontal components involved in memory.

Method: In this study, 25 patients with frontal lobe damage and 25 matched with the age, education of healthy control groups, two groups of patients were taken general cognitive function test, as well as explicit memory and implicit memory test.

Result: the frontal lobe injured group not only exists damage in background neuropsychology tasks, but the conceptual explicit memory and implicit memory are obvious damage, the performance of perceptual explicit memory and implicit memory in frontal lobe damage group has no obvious different with healthy controls.

Conclusion: The results indicated that frontal lobe characteristic to participate in the conceptual memory processing, and the present study take a neuropsychological verification to the component process model of memory.

Keywords: Frontal lobe; implicit memory; explicit memory; component process model; neuropsychology test.

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