

## **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. 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 consulted to the hospital with the complaint of involuntary movements of extremities. There was gradual progression of symptoms which initially started as jerky, repetitive, purposeless movements of the head and shoulders until there was involvement of trunk and all extremities. He developed behavioral changes and cognitive problems. His father, 2 paternal uncles and aunt also presented with the same symptoms. He has random facial grimaces with intermittent protrusion of the tongue and irregular shoulder jerks with athetoid movement of the distal extremities. He has involuntary, brief, irregular, jerky movements that flow from proximal to distal extremities. Cranial CT scan showed atrophy of bilateral caudate nucleus. Genetic testing showed CAG repeat of 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.