Newborn Screening, Inborn Errors of Metabolism

AB121. Late infantile neuronal ceroid lipofuscinosis in a Filipino child presenting with epilepsy and progressive neurodegeneration

Mary Anne D. Chiong[1,2], Benilda Sanchez-Gan[1,2]

[1]Department of Pediatrics, Philippine General Hospital, University of the Philippines Manila, Manila, Philippines; [2]Institute of Human Genetics, National Institutes of Health, Manila, Philippines

Background: Neuronal ceroid lipofuscinosis is a group of disorders characterized by neurodegeneration and intracellular built up of auto-fluorescent lipopigment (ceroid lipofuscin). They are classified by age of onset into infantile, late infantile, juvenile, and adult forms. Among these, the classic late infantile type is caused by mutations in the tripeptidyl peptidase 1 (*TPP1*) gene and is characterized by age of onset between 2–4 years, seizures, early progressive cognitive impairment, and visual loss.

Methods: Clinical and biochemical analysis was performed.

Results: The present patient was a 4-year-old girl who presented at 2 years and 10 months old with seizures followed by ataxia, regression of skills, and eventual visual decline. TPP1 enzyme activity was below normal for age.

Conclusions: This report may help increase the awareness of neuronal ceroid lipofuscinosis among physicians on the cluster of characteristic symptoms of this disorder which will facilitate early diagnosis and prompt institution of appropriate management.

Keywords: Late infantile neuronal ceroid lipofuscinosis; neuronal ceroid lipofuscinosis type 2; tripeptidyl peptidase 1

doi: 10.21037/atm.2017.s121

Cite this abstract as: Chiong MA, Sanchez-Gan B. Late infantile neuronal ceroid lipofuscinosis in a Filipino child presenting with epilepsy and progressive neurodegeneration. Ann Transl Med 2017;5(Suppl 2):AB121. doi: 10.21037/atm.2017.s121