### Accepted Manuscript

First case of genetically confirmed CLN3 disease in Chinese with cDNA sequencing revealing pathogenicity of a novel splice site variant

Nike Kwai-Cheung Lau, Chor Kwan Ching, Hencher Han Chih Lee, Wai Kwong Mario Chak, N.G. Kwan Shing, Neil A. Hanchard, Chloe Miu Mak

PII: S0009-8981(18)30387-5

DOI: doi:10.1016/j.cca.2018.07.040

Reference: CCA 15326

To appear in: Clinica Chimica Acta

Received date: 5 June 2018
Revised date: 20 July 2018
Accepted date: 23 July 2018



Please cite this article as: Nike Kwai-Cheung Lau, Chor Kwan Ching, Hencher Han Chih Lee, Wai Kwong Mario Chak, N.G. Kwan Shing, Neil A. Hanchard, Chloe Miu Mak, First case of genetically confirmed CLN3 disease in Chinese with cDNA sequencing revealing pathogenicity of a novel splice site variant. Cca (2018), doi:10.1016/j.cca.2018.07.040

This is a PDF file of an unedited manuscript that has been accepted for publication. As a service to our customers we are providing this early version of the manuscript. The manuscript will undergo copyediting, typesetting, and review of the resulting proof before it is published in its final form. Please note that during the production process errors may be discovered which could affect the content, and all legal disclaimers that apply to the journal pertain.

## ACCEPTED MANUSCRIPT

First case of genetically confirmed CLN3 disease in Chinese with cDNA sequencing revealing pathogenicity of a novel splice site variant.

Dr Nike Kwai-Cheung LAU<sup>1</sup> Ikc416@ha.org.hk, Dr Chor Kwan CHING<sup>1</sup> chingck2@ha.org.hk, Dr Hencher Han Chih LEE<sup>1</sup> Ihc305@ha.org.hk, Dr Wai Kwong Mario CHAK<sup>2</sup> chakwk@ha.org.hk, Dr Kwan Shing NG<sup>3</sup> nks422@ha.org.hk, Dr Neil A. HANCHARD<sup>4</sup> hanchard@bcm.edu and Dr Chloe Miu MAK<sup>1</sup> makm@ha.org.hk

#### \*Corresponding author:

Dr Chloe Miu MAK

Tel: (852) 2990 1882

Email: makm@ha.org.hk

Postal address: Chemical Pathology Laboratory, Department of Pathology, Princess Margaret Hospital, 2-10 Princess Margaret Hospital Road, Lai Chi Kok, Kowloon, Hong Kong SAR

<sup>&</sup>lt;sup>1</sup>Kowloon West Cluster Laboratory Genetic Service, Chemical Pathology Laboratory, Department of Pathology, Princess Margaret Hospital, Hong Kong

<sup>&</sup>lt;sup>2</sup>Department of Paediatrics and Adolescent Medicine, Tuen Mun Hospital, Hong Kong

<sup>&</sup>lt;sup>3</sup>Department of Pathology, Tuen Mun Hospital, Hong Kong

<sup>&</sup>lt;sup>4</sup>Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, USA

#### Download English Version:

# https://daneshyari.com/en/article/8309425

Download Persian Version:

https://daneshyari.com/article/8309425

<u>Daneshyari.com</u>