JMD
https://doi.org/10.14802/jmd.21120 / J Mov Disord 2021 Dec 24 [Epub ahead of print]
pISSN 2005-940X / eISSN 2093-4939

CASE REPORT

Labrune's Syndrome Presenting With Stereotypy-Like Movements and Psychosis: A Case Report and Review

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ABSTRACT

Labrune's syndrome, or leukoencephalopathy with brain calcifications and cysts (LCC), is a rare genetic syndrome with variable neurological presentations. Psychiatric manifestations and involuntary movements are uncommonly reported. We report the case of a 19-year-old female, initially diagnosed with Fahr's syndrome, who presented to us with acute psychosis, abnormal behavior and involuntary movements. Her brain computed tomography showed extensive bilateral intracranial calcifications without cysts. Genetic testing detected two compound heterozygous variants, NR_033294.1 n.*9C>T and n.24C>T, in the *SNORD118* gene, confirming the diagnosis of LCC. We discuss the expanding phenotypic spectrum of LCC and provide a literature review on the current diagnosis and management of this rare syndrome.

Keywords Fahr's syndrome; Intracranial calcification; Labrune's syndrome; Malaysia; *SNORD118*.

Leukoencephalopathy with brain calcifications and cysts (LCC) is a rare inherited microangiopathic disorder of the brain first reported in 1996.[1] To this date there are less than 100 cases reported worldwide and the clinical spectrum for this condition is still expanding. There is no established diagnostic criteria for LCC thus, patients are usually diagnosed late. Clinicians rely heavily on serial brain imaging and clinical progression to establish this diagnosis. Here we report the first case of LCC in Malaysia with atypical features and provide a review of this rare condition.

CASE REPORT

A 19-year-old female previously diagnosed with Fahr's syndrome and epilepsy, first presented to Universiti Kebangsaan Malaysia Medical Centre in June 2018 with a three-day history of abnormal behavior associated with emotional lability, paranoia, irrelevant speech and a poor sleep pattern. There was gait instability and involuntary movements of her hands, mainly on the right. On examination, she was of short stature, had inappropriate affect, smiled and cried occasionally and did not obey commands. She had irrelevant speech with slurring of words. Neurological examination revealed no deficits or signs of meningism. There were choreoathetoid movements of her upper limbs (right > left), which appeared stereotyped and mimicked a traditional Malay dance movement (Supplementary Video 1 in the online-only Data Supplement). Brain computed tomog-

Received: August 16, 2021 **Revised:** September 25, 2021 **Accepted:** October 19, 2021
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