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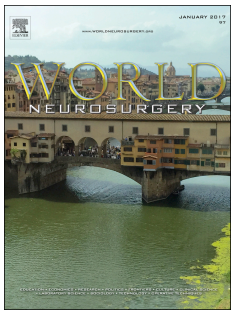

Leukoencephalopathy, intracranial calcifications, cysts and SNORD118 mutation (Labrune Syndrome) with obstructive hydrocephalus

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PII: S1878-8750(19)30407-3

DOI: https://doi.org/10.1016/j.wneu.2019.02.008

Reference: WNEU 11469

To appear in: *World Neurosurgery*

Received Date: 23 October 2018

Revised Date: 8 February 2019

Accepted Date: 9 February 2019

Please cite this article as: Shtaya A, Elmslie F, Crow Y, Hettige S, Leukoencephalopathy, intracranial calcifications, cysts and SNORD118 mutation (Labrune Syndrome) with obstructive hydrocephalus, *World Neurosurgery* (2019), doi: https://doi.org/10.1016/j.wneu.2019.02.008.

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Leukoencephalopathy, intracranial calcifications, cysts and SNORD118 mutation (Labrune Syndrome) with obstructive hydrocephalus

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Title character count: 116.

Number of references: 3.

Number of figures: 1.

Word count paper: 322.

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Labrune syndrome is characterised by brain calcifications, leukodystrophy, and formation of parenchymal and cerebellar cysts due to progressive diffuse cerebral microangiopathy[1]. This rare condition has been described in children and adults who may present with cognitive impairment, seizures, and a mixture of extrapyramidal, cerebellar and pyramidal signs[1]. Mutations in the gene *SNORD118*, encoding a small nucleolar RNA U8, were recently described in patients with leukoencephalopathy with brain calcifications and cysts (LCC; Labrune syndrome)[2,3], confirming the autosomal recessive basis of the condition. Patients may develop parenchymal and cerebellar cysts with mass effect and hydrocephalus that require neurosurgical intervention.

We describe a 12 year-old boy with a background history of motor developmental delay, who presented with a 3 month history of headache that worsened one week before admission, associated with nausea and vomiting and ataxia. His CT (Panels A-C) and MRI (Panels D-E) head scans demonstrated widespread calcifications in the cerebral hemispheres, left lentiform nucleus, thalami and dentate nuclei, bilateral cerebellar calcifications and a large left cerebellar cyst causing obstructive hydrocephalus (arrows show calcification, arrow head shows cyst). He underwent urgent endoscopic assisted aspiration of the cerebellar cyst with insertion of an ommaya reservoir. This resulted in reduction of the cyst size and resolution of the hydrocephalus (Panels F-G) and symptoms of raised intracranial pressure on follow up 12 months after surgery. Histopathology of a biopsy of the cyst wall and calcified regions showed non-specific changes with no evidence of malignancy Genetic testing revealed an n.72A>G and an n.*10G>T biallelic variants in *SNORD118* inherited from his carrier parents (See supplementary material for genetic

testing and electropherogram). Both variants have been previously described in other families affected by LCC.[2,3] To our knowledge, we report the first case of a child with LCC (Labrune syndrome) with infra and supra tentorial calcifications and cyst with obstructive hydrocephalus that required neurosurgical intervention. This rare and severe form of Labrune syndrome will represent a challenge to the neurosurgical community.

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Figure Legend

Panels A-C Axial CT images demonstrate the hydrocephalus, calcifications (arrows) and cyst (arrow head). Panels D-E Axial T2-weighted MRI demonstrating obstructive hydrocephalus, cysts (arrow head) and calcifications (arrows). Panels F-G Post-operative axial T2-weighted MRI showing resolution of the hydrocephalus and reduction in the size of the cerebellar cyst (arrow head).

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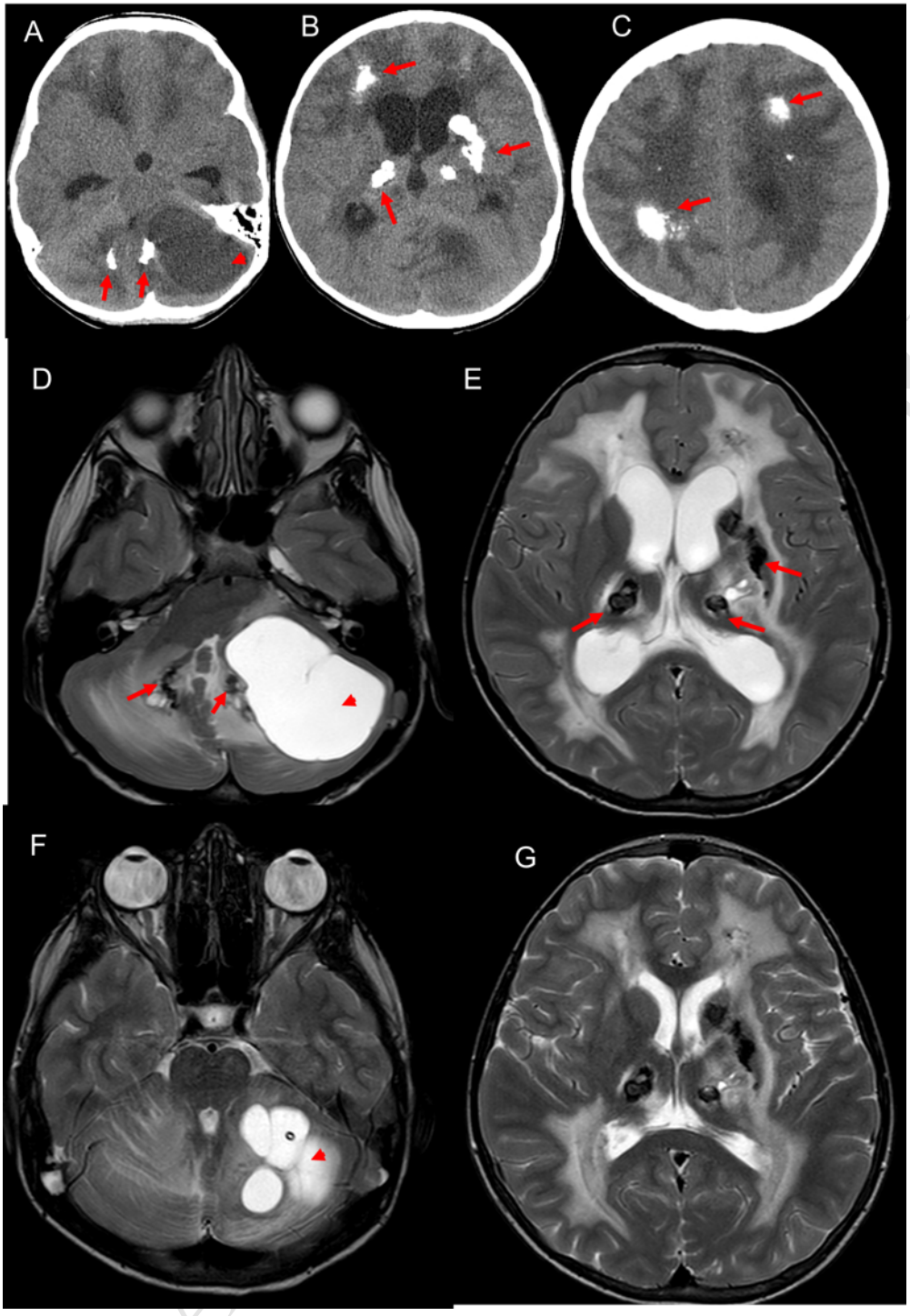
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Abbreviation:

Magnetic resonance imaging: MRI

T2 weighted: T2W

Computed tomography: CT

Leukoencephalopathy, intracranial calcifications and cysts: LCC

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