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LEUKOENCEPHALOPATHY WITH CALCIFICATIONS AND CYSTS (LCC): 5 PERSONAL CASES AND LITERATURE REVIEW

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Introduction

LCC is a rare autosomal recessive cerebral angiomatous-like **microangiopathy** caused by SNORD118 mutations and characterized by diffuse and **asymmetric white-matter lesions** associated with **multiple calcifications** and **cysts**.

The entire clinical spectrum of LCC is not yet fully determined. We herein report **5 personal genetically confirmed LCC cases** (*figure 1*) and review the literature to collect clinical and imaging data from all previous case reports.

Results

We analysed data from 92 patients. Mean age at first clinical manifestations was 16.1 y-o (from 1 month to 71 y-o). Evidence of consanguinity was obvious in only 4% of cases.

Epileptic seizures revealed LCC is 36% of cases. **Progressive neurological deficits** were reported as the first neurological symptom in 26% of patients. **Developmental delay** and **intracranial hypertension** (ICH) were inaugural signs in 14% of patients respectively. One ischemic and one haemorrhagic **stroke** revealed LCC in 2 different adults (2%). Rarely, LCC was incidentally discovered (2%).

We observed during the clinical course of the disease slow progression of focal neurological deficits in 79% of cases, cognitive impairment in 45% of cases, epilepsy in 38% of cases and ICH in 30% of cases. Two patients (2%) presented haemorrhagic strokes.

To treat cystic expansion, surgical treatments were frequently needed (28 cases).

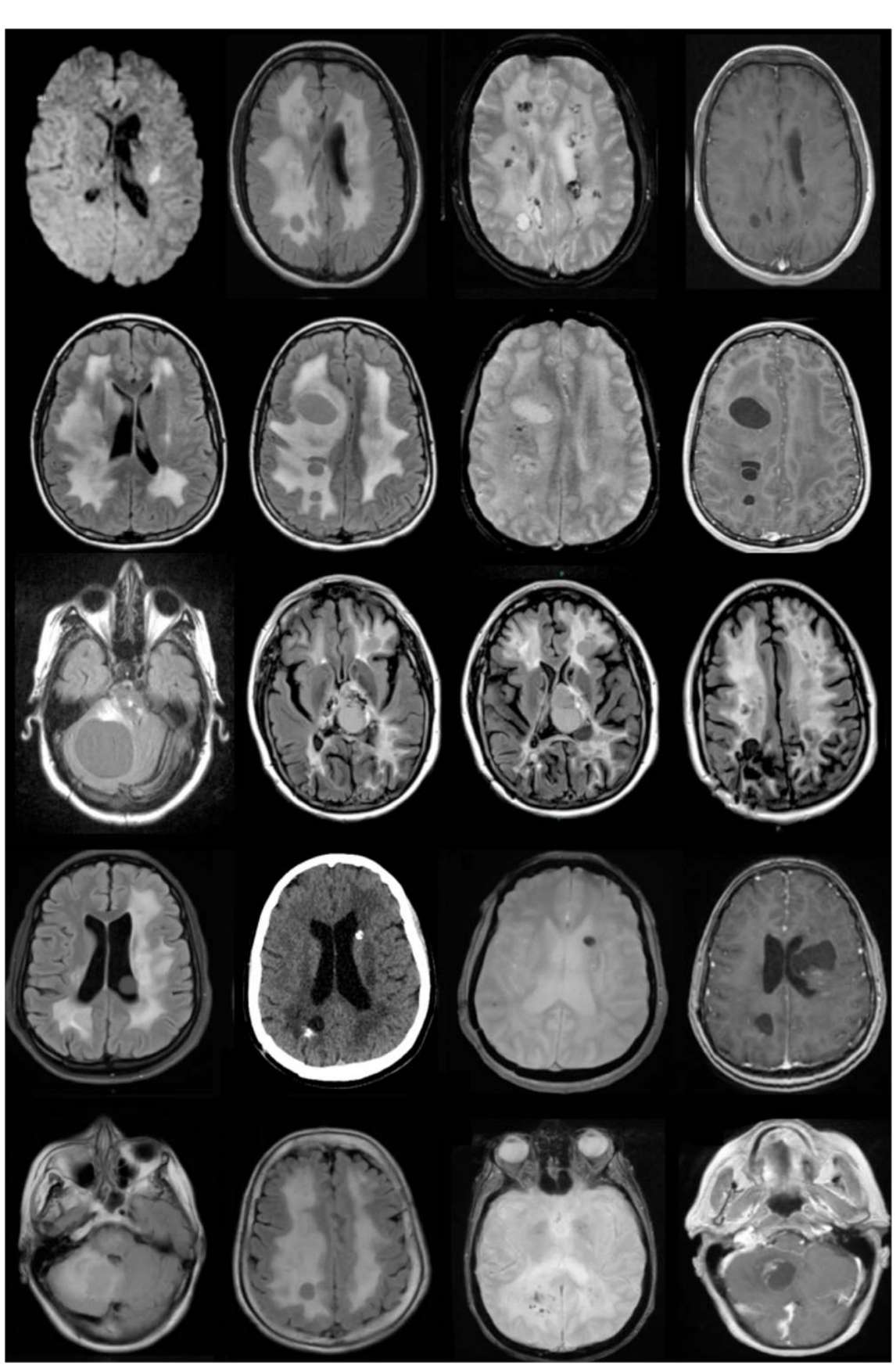

Figure 1 – Neuroradiological aspects of our 5 patients
1e lane: Sudden R-hemiparesis revealing ischemic stroke at 30 y-o.; 2d lane: ICH in a 28 y-o woman; 3d lane: Generalized seizure at 11 y-o; 4th lane : Epileptic seizure in a 18 y-o woman; 5th lane : Progressive cerebellar ataxia in a 60 y-o woman.

Discussion and Conclusion

Age at onset, clinical presentation and course of LCC is heterogenous. These important variabilities may be related to genetic variant with different phenotypic consequences.

Most of neurological signs (epileptic seizures, progressive neurological deficit and ICH) seem to be related to cyst expansion. Yet, the exact origin of cognitive impairment remains unclear; recurrent cysts and/or the accumulation of white matter lesions might be involved. The occurrence of ischemic and haemorrhagic events may imply that, as other hereditary small vessel diseases as CADASIL, CARASIL, HERNS, cerebroretinal angiopathy with COL4A1 or familial angiopathy amyloid, LCC might predispose to strokes.

LCC should be suggested in cases of white matter lesions associated with cerebral cysts and calcifications, regardless of patient's age and even in the absence of a familial history of neurological disorder.