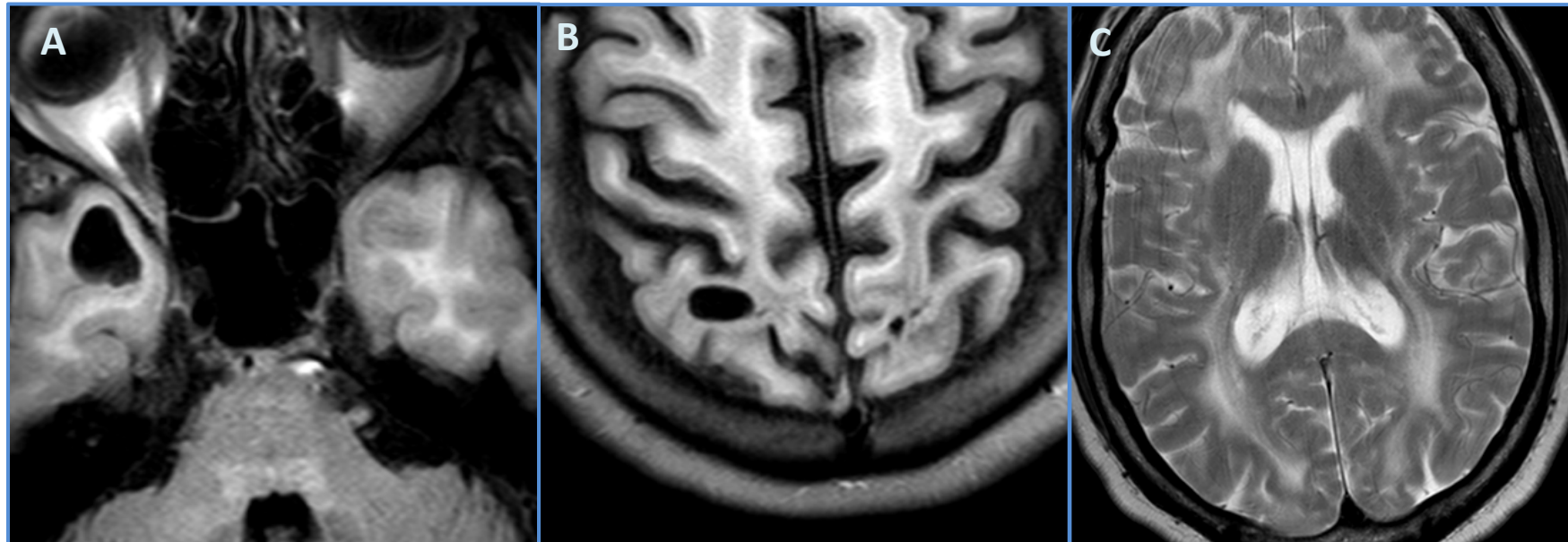


A 42 year old man with schizophrenia presented with generalised tonic-clonic seizures. On enquiry there was a history of macrocephaly soon after birth, and slowly progressive clumsiness and seizures between age 6 and 16 years, for which no diagnosis had been made. He was otherwise well.



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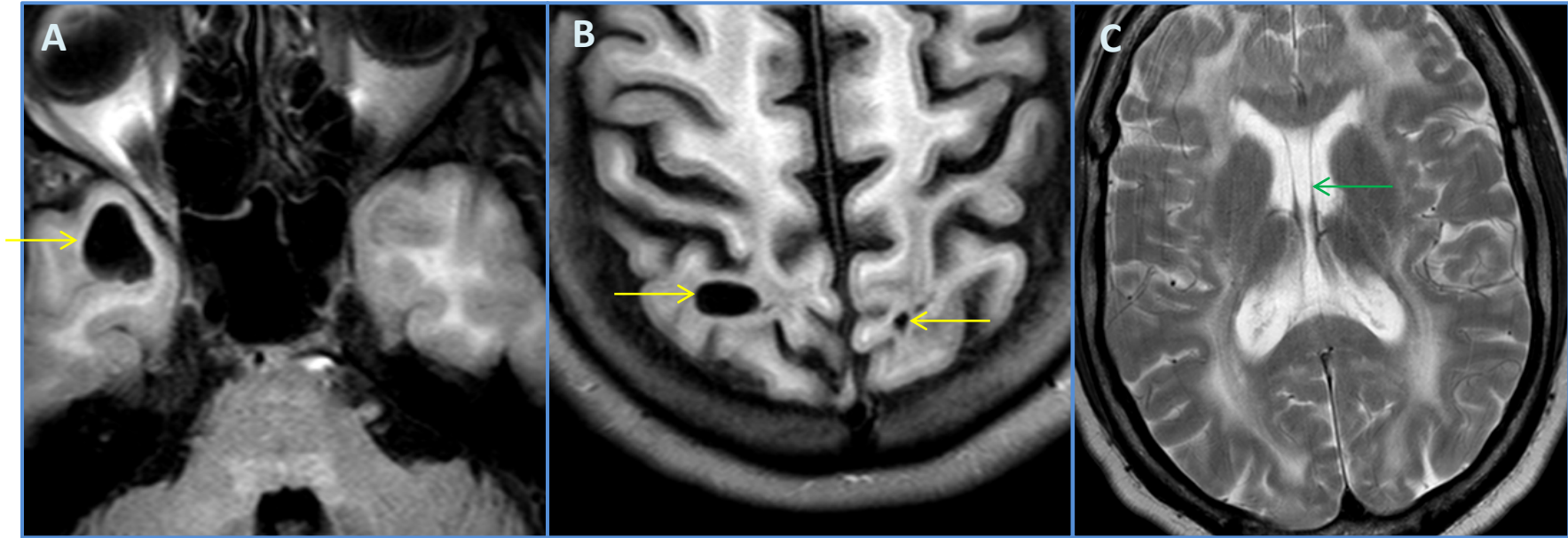


Figure 1 Axial FLAIR images (A) through the anterior temporal poles and (B) superior frontoparietal convexities showing subcortical cysts (yellow arrows). Axial T2 (C): the patient also has a cavum septum pellucidum et vergae (green arrow). Note also the extensive confluent white matter hyperintensity in all 3 images.

# Megalencephalic leukoencephalopathy with subcortical cysts (MLC) 1

## Background

- Rare autosomal recessive dysmyelination disorder with two genetic mutations attributed to it- MLC1 and HEPACAM (1).

## Presentation and diagnosis

- The diagnosis was made provisionally based on clinical and radiological findings, and confirmed by genetic testing, which in our patient revealed an MLC1 mutation.
- Typically, patients present with a variable degree of macrocephaly within the first year, followed by marked motor developmental delay seen in early childhood. Slow cognitive decline also ensues after a variable period of time and early onset seizures that respond to therapy is often a feature (2).
- This diagnosis is therefore almost always made in a paediatric age group.

## Imaging features

- Subcortical white matter cysts are invariably present at the anterior temporal poles and frequently seen at the superior fronto-parietal convexities. There is diffuse cerebral white matter T2 hyperintensity but with relative sparing of the internal capsules and corpus callosum. There is no correlation between severity of MR findings and clinical course of the patients (3).
- Cavum septum pellucidum and cavum vergae are reported in most of the patients with MLC 1.

## Differentials

- Other differential diagnoses to consider include congenital CMV infection, where the related encephalopathy is clinically static, and vanishing white matter disease, where the clinical deterioration is much more severe and typically leads to death in childhood (3).

Treatment is supportive and aimed at symptomatic reduction with seizure control and motor dysfunction.

## References

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