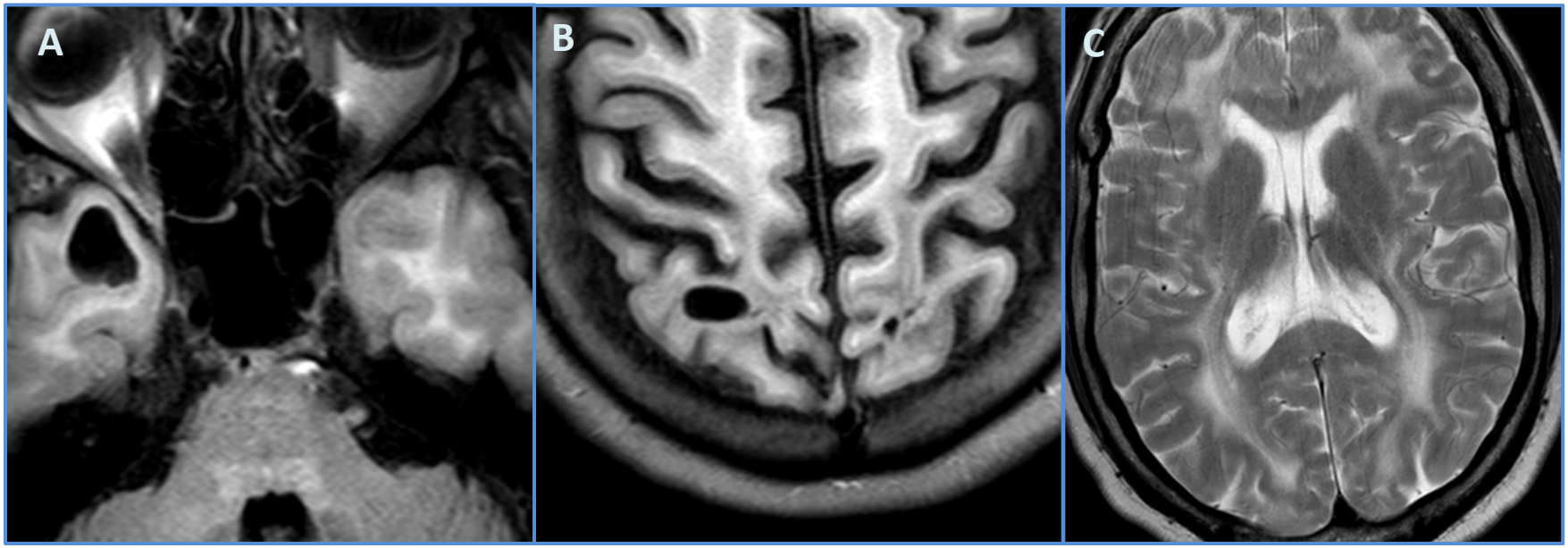
A 42 year old man with schizophrenia presented with generalised tonic-clonic seizures. On enquiry there was a history of macrocephaly soon after birth, and slowly progressive clumsiness and seizures between age 6 and 16 years, for which no diagnosis had been made. He was otherwise well.



A. Nitkunan PhD [1], S. Saipriya MD [2], A. D. MacKinnon MD [2], H. R. Cock FRCP MD [1,3]

1 Department of Neurology, Atkinson Morley Regional Neuroscience Centre, St. George's Hospital, London, UK

2 Department of Neuroradiology, Atkinson Morley Regional Neuroscience Centre, St. George's Hospital, London, UK

3 Institute of Medical & Biomedical Education, St Georges University of London, UK

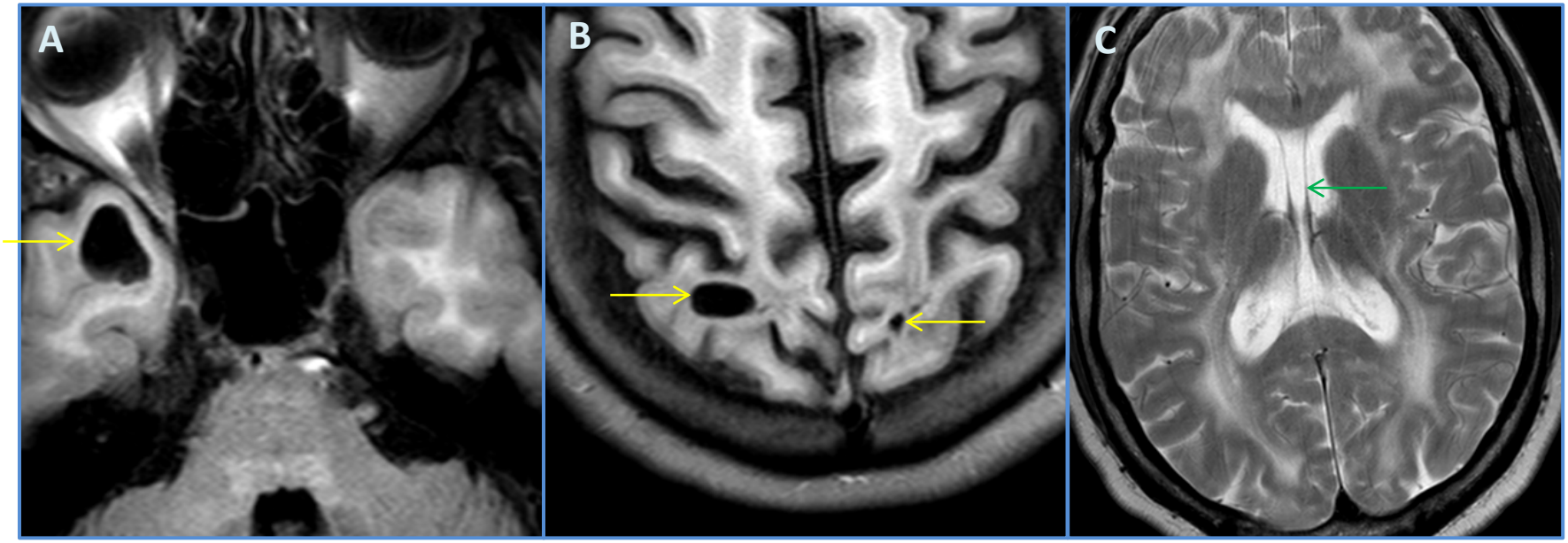


Figure 1 Axial FLAIR images (A) through the anterior temporal poles and (B) superior frontoparietal convexities showing subcortical cysts (yellow arrows). Axial T2 (C): the patient also has a cavum septum pellucidum et vergae (green arrow). Note also the extensive confluent white matter hyperintensity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

Megalencephalic leukoencephalopathy with subcortical cysts (MLC) 1

Background

- Rare autosomal recessive dysmyelination disorder with two 2 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

1. van der Knaap, Marjo S., Ilja Boor, and Raúl Estévez. "Megalencephalic leukoencephalopathy with subcortical cysts: chronic white matter oedema due to a defect in brain ion and water homoeostasis." *The Lancet Neurology*11.11 (2012): 973-985.

2. Yi-Fang Tu, Cheng-Yu Chen, Chao-Ching Huang, and Chang-Shin Lee. Vacuolating Megalencephalic Leukoencephalopathy with Mild Clinical Course Validated by Diffusion Tensor Imaging and MR Spectroscopy AJNR Am J Neuroradiol 2004 25: 1041-1045.

3. van der Knaap, Marjo S., et al. "Pattern of White Matter Abnormalities at MR Imaging: Use of Polymerase Chain Reaction Testing of Guthrie Cards to Link Pattern with Congenital Cytomegalovirus Infection 1." *Radiology* 230.2 (2004): 529-536.