Case 18296

Eurorad

The subtle art of neuronal migration - Grey matter heterotopia

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Section: Neuroradiology
Area of Interest: Anatomy Neuroradiology brain Neuroradiology spine Case Type: Clinical Cases
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Patient: 35 years, male

Clinical History:

35-year-old gentleman referred by the GP with history of spasms around the left eye and twitching of the left side of the face and transient facial droops which lasted 2-3 minutes. He also complained of left arm reduced sensation with pins and needles at the fingertips.

Imaging Findings:

MRI head: Incidental note is made of a 10 x 9.6 x 7.7 mm nodule which projects into the trigone of the right lateral ventricle. It has signal characteristics which matches those of grey matter on all sequences. No restricted diffusion on DWI and ADC.

MRI head with contrast: No enhancement of this 10 mm subependymal lesion, thus establishing the diagnosis of unilateral focal nodular subependymal grey matter heterotopia.

Discussion:

Grey matter heterotopias (GMH) are a group of neurological disorders characterized by the ectopic position of neurons; thus they are also known as neuronal migration disorders. Disruption of the complex sequential processes, such as neuronal migration, cell proliferation, cortical organization, and formation of neuronal networks, yields cortical malformations including GMH; this is characterized by the ectopic position of neurons along the ventricular walls or in the deep white matter [1].

Macroscopically GMH can be classified into Nodular and diffuse heterotopia [2]. Nodular heterotopia encompasses Subcortical and Subependymal, whereas diffuse types are called band heterotopias.

Although most cases are sporadic, some are X-linked recessive (Xq28) [1]. Abnormal production of Filamin-1 is most likely to be the result of X-linked recessive disease. Affected females have relatively mild cognitive deficits and tend to develop epilepsy later, whereas affected males are spontaneously aborted, often due to cardiovascular defects. Those that survive have epilepsy and developmental delay.

Imaging was needed in this patient due to the clinical presentation of the patient. Given the above-mentioned presentation, an infarct was initially queried, however, this was ruled out by MRI head (DWI). GMH can be subtle and is easily missed on head imaging—therefore, it is paramount that the clinical presentation is correlated to the Image findings.

Incidental findings on non-contrast MRI, prompted contrast-enhanced MRI for the following reasons:

1. To establish the final diagnosis.
2. To rule out other potential differentials by showing non-enhancement.
3. To look for any associated anomalies such as sub-ependymal giant cell astrocytoma, sub-ependymal tuber of the tuberous sclerosis and normal germinal matrix [4].

Final diagnosis was made on contrast-enhanced MRI which ruled out other possible differentials.

Outcome

The patient was offered Neurostimulation after a Multi-disciplinary team meeting. Other therapeutic options include Carbamazepine Dietary therapy, Neurostimulation devices such as vagus nerve stimulation, responsive neurostimulation, and deep brain stimulation and Surgical resection of the affected area which may provide relief from seizures [3].

Teaching points

- GMH is found in 15% of patients [5] with cortical developmental malformation; the diagnosis is easily missed on MRI scans as the focal subtype of heterotopias can be very subtle & resemble the normal grey matter. Therefore, thorough analysis is paramount.
- 2% of the patients with grey matter heterotopia present as epilepsy therefore it is important to have GMH as a differential in a patient presenting with seizures.

Written informed patient consent for publication has been obtained.

Differential Diagnosis List: Subependymal grey matter heterotopia (unilateral focal), Subependymal giant cell astrocytoma, Subepencymal tuber of tuberous sclerosis, Occlusive infarct, Taylor dysplasia

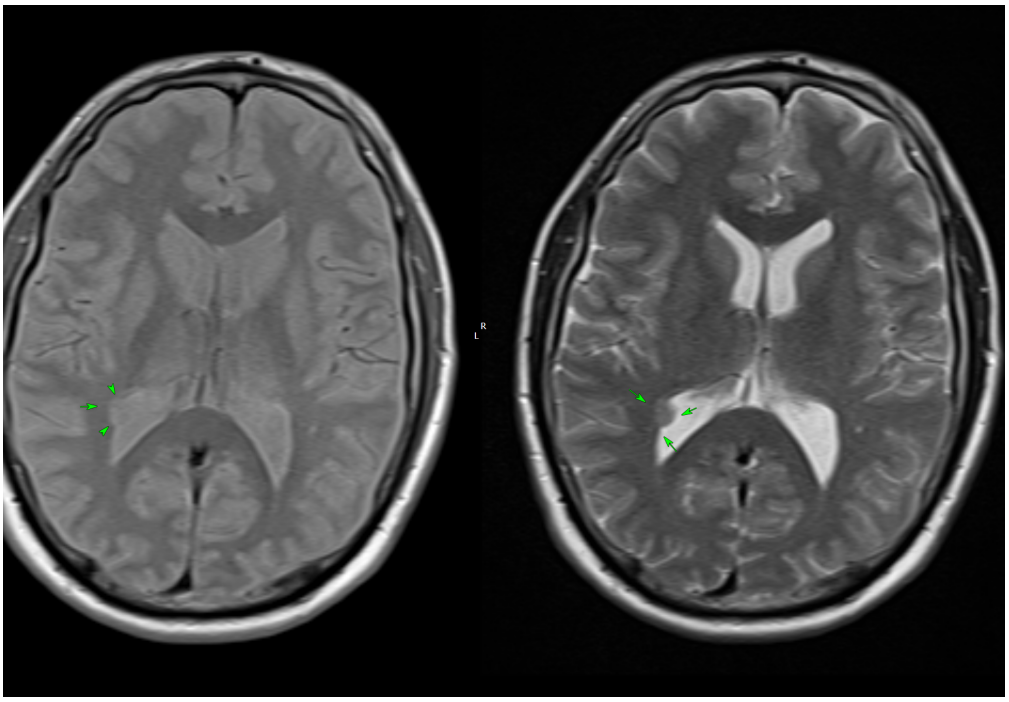
Final Diagnosis: Subependymal grey matter heterotopia (unilateral focal)

References:

Watrin F, Manent JB, Cardoso C, Represa A (2014) Causes and consequences of gray matter heterotopia. CNS Neurosci Ther 21(2):112-22 (PMID: 25180909)
Vriend I, Oegema R (2021) Genetic causes underlying grey matter heterotopia. Eur J Paediatr Neurol 35:82-92 (PMID: 34666232)
Alwaki A (2021) Periverntricular Nodular Heterotopias: Epilepsy.com
El-Feky M (2023) Subependymal grey matter heterotopia. Case study, Radiopaedia.org. https://doi.org/10.53347/rID-75935

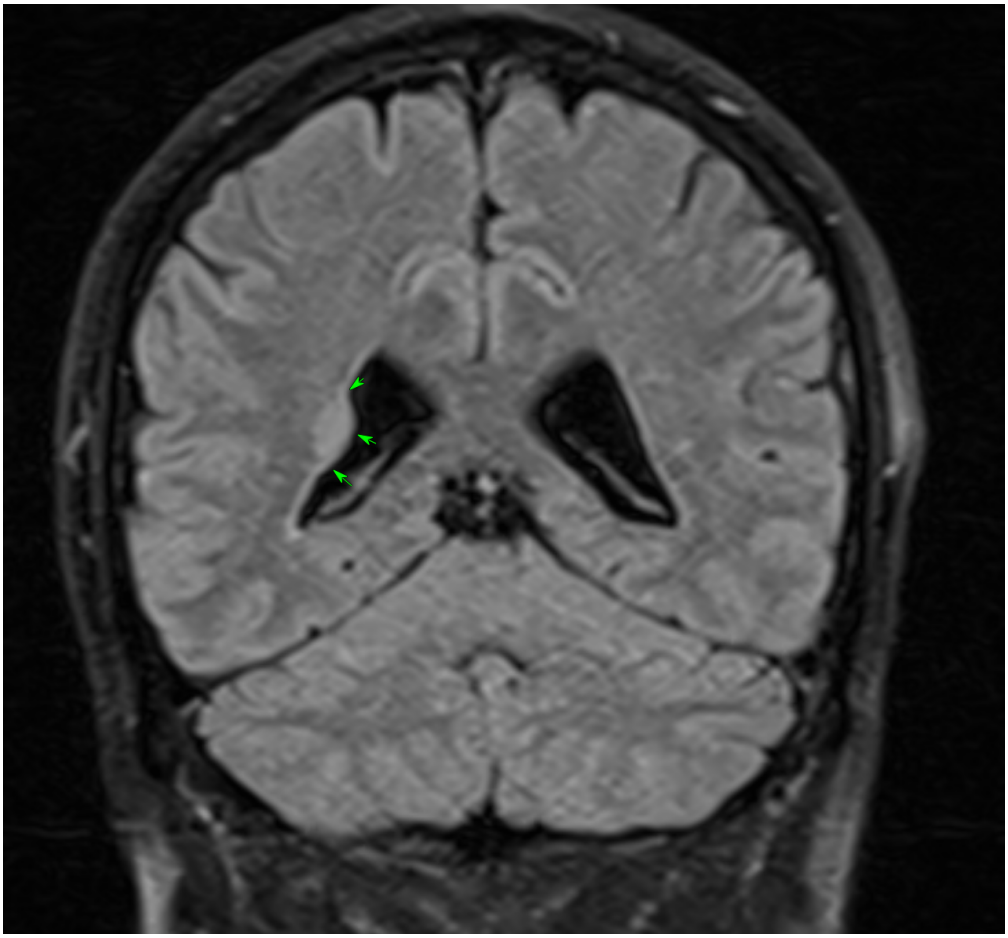
Figure 1

a



Description: Non-contrast MRI brain axial PD **Origin:** © Department of Radiology, Basildon and Thurrock University Hospital, United Kingdom, 2023

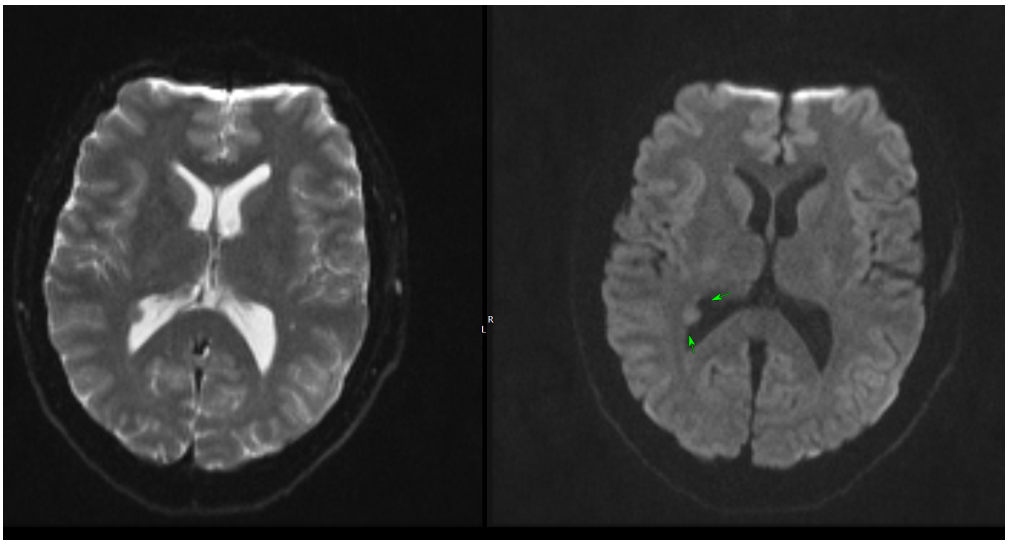
b



Description: T2 and coronal T2 FLAIR: 10x7mm well-defined focal nodular lesion in subependymal location, signals similar to grey matter on all sequences **Origin:** © Department of Radiology, Basildon and Thurrock University Hospital, United Kingdom, 2023

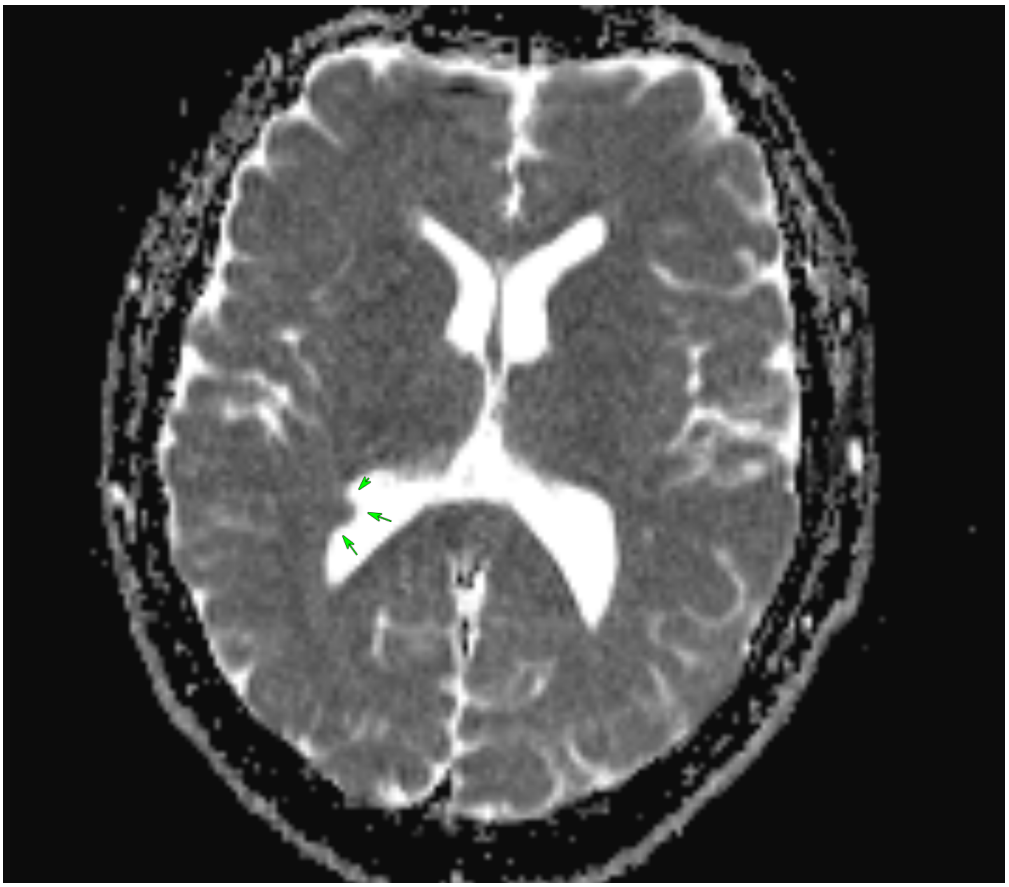
Figure 2

a



Description: DWI sequence **Origin:** © Department of Radiology, Basildon and Thurrock University Hospital, United Kingdom, 2023

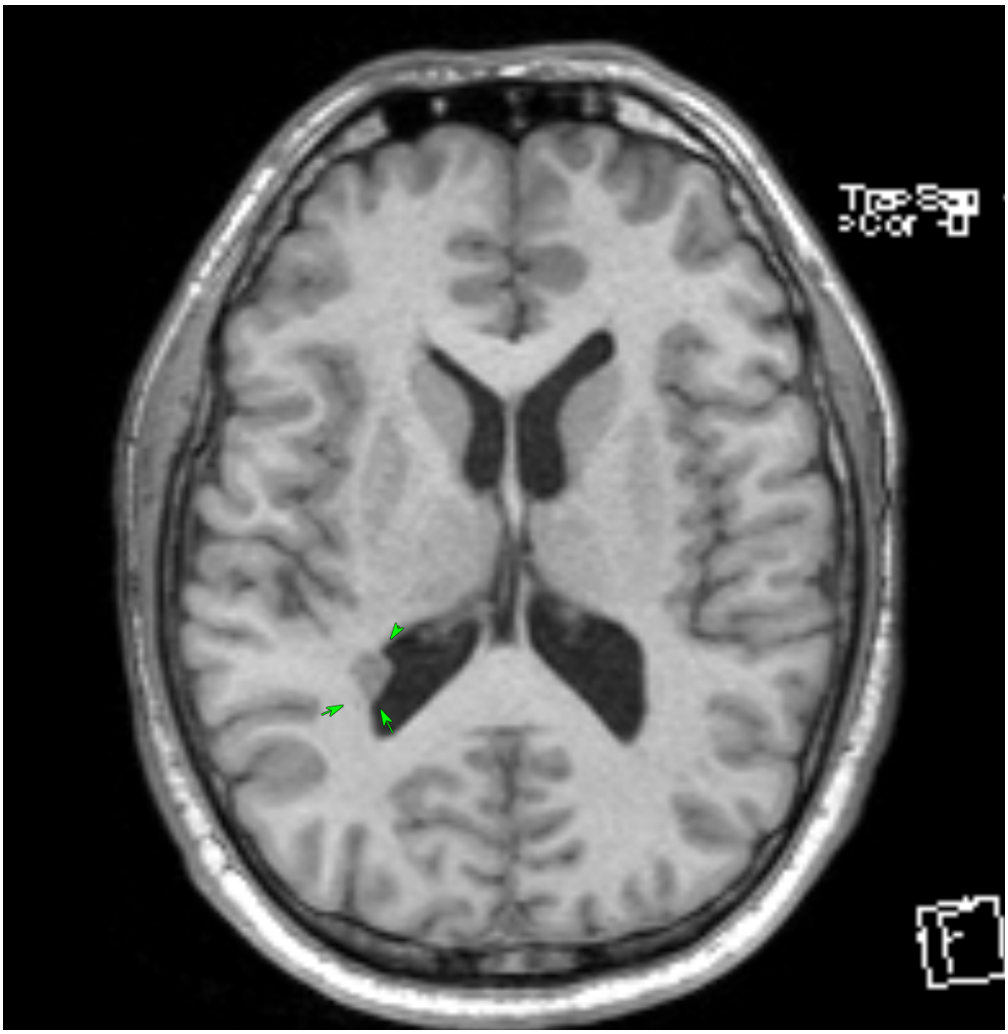
b



Description: Corresponding ADC sequence. No restricted diffusion could be seen on the DWI/ADC sequences is corresponding ADC **Origin:** © Department of Radiology, Basildon and Thurrock University Hospital, United Kingdom, 2023

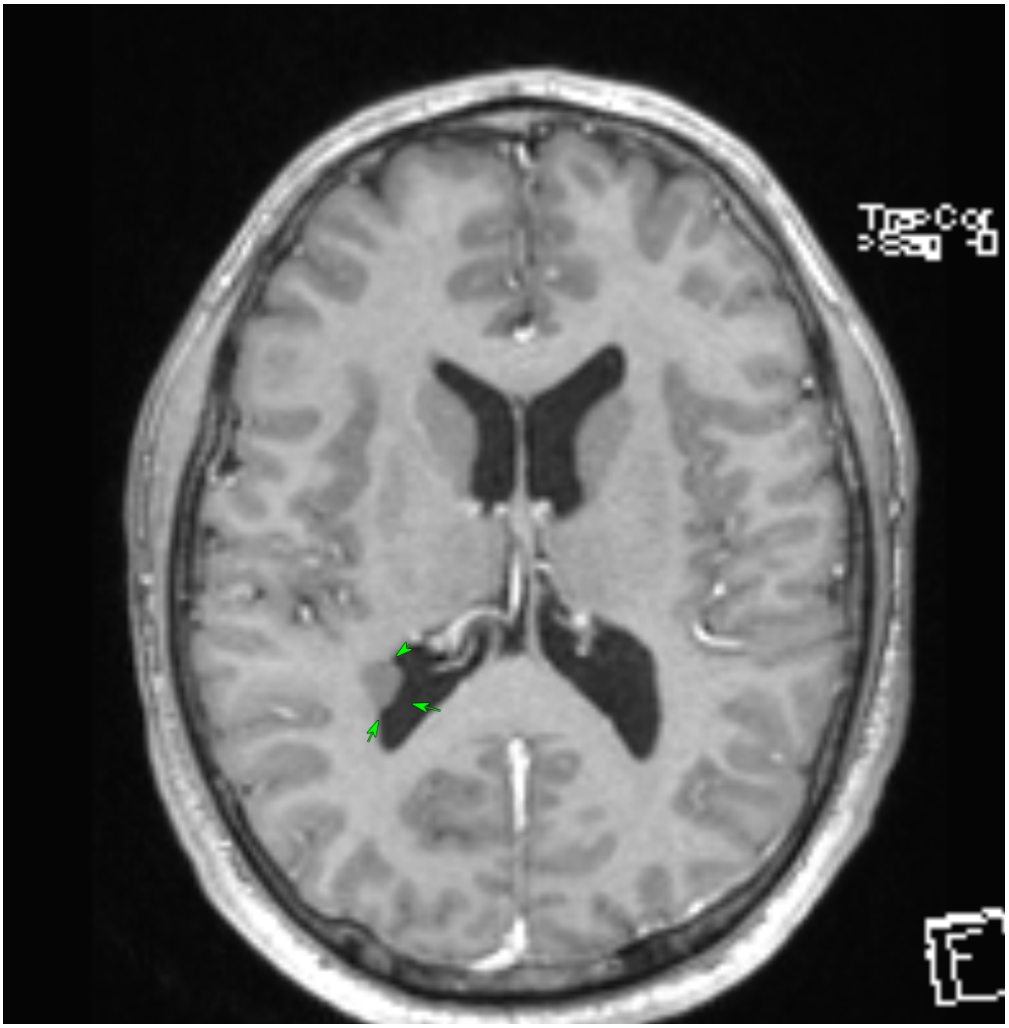
Figure 3

a



Description: Pre-contrast T1 **Origin:** © Department of Radiology, Basildon and Thurrock University Hospital, United Kingdom, 2023

b



Description: Post-contrast T1. Axial pre & post-contrast T1 sequence: No enhancement seen within it