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Grey Matter Focal Subcortical Heterothopia-A Case Report

By Mihaela Lungu

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I. INTRODUCTION

The grey matter heterothopia (GMH) is a brain malformation caused by abnormal neuronal migration. This impairment of neuronal migration takes place during the third to fifth month of gestation.

In these cases, a subset of neurons fails to migrate from the vicinity of the ventricle into the developing cerebral cortex. So, they form nodules that line the ventricular surface: "normal neurons in abnormal locations". The neurons fail to climb to the end of their ladder correctly and are permanently situated in the wrong location.

Classically, GMH is a X-linked dominant disorder, more frequent in females. The patients present intelligence varying from normal to borderline mental retardation. Other related pathology include epilepsy, cardiovascular defects or coagulopathy. This disorder is usually associated with premature lethality in males, who have a high risk of aortic pathology.

Periventricular heterothopias is related to chromosome 5. Its incidence is unknown. It is caused by mutations in ARFGEF2 and FLNA genes. The FLNA gene provides instructions for producing the protein filaminin-A, which helps building the network of protein filaments-cytoskeleton, that gives structure to cells and allows them to change shape and move ()

In GMH, results are impaired. FLNA protein cannot perform this function, disrupting the normal migration of neurons, during the development of the brain. GMH is associated with mutations in the Filanin-A gene (FLN1, FLNA, ABP-280), ARFGEF2 gene and, in some individual cases, a chromosomal rearrangement in 5P151. Syndactily and mental retardation are also associated with N.H. (1)

GMH may also be caused by infection or trauma.

Macroscopically, GMH can be divided into:

1. Nodular heterothopias:
 - subependymal heterothopias- most common
 - subcortical heterothopias.
2. Diffuse heterothopias: band heterothopias (double cortex heterothopias, X-linked lissencephaly), laminar heterothopias, lissencephaly - types 1 and 2. (1)

a) Focal subcortical GMH (FSCGMH)

Clinical presentation: most commonly, patients present epileptic seizures such as partial seizures, progressing towards drug resistant epilepsy in the second decade of life. Additionally there are also some associated development delays or mental retardation. GMH are more frequent in patients with other congenital central nervous system anomalies, such as the agenesis of the corpus callosum, Chiari II malformation and pachygyria.

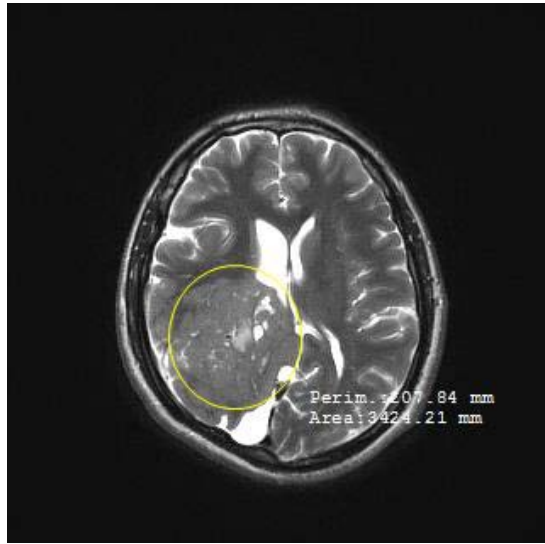
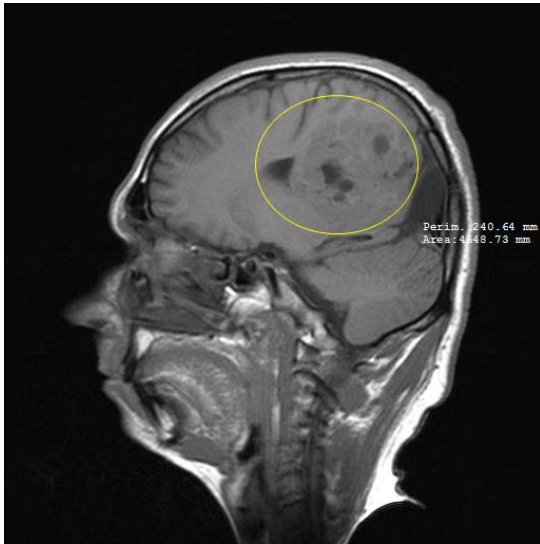
FSCGMH forms distinct nodules in the white matter, in a specific area. Patients present fixed neurologic deficits and develop partial epilepsy, between the ages of 6 and 10.

Management of the patient presenting GMH is based on symptomatic treatment, including antiepileptic drugs and surgical therapy for cardiovascular defects (most often persistence of Botallo's duct and aortic valvulopathy.)

II. CASE REPORT

This article aims to present a case of a 40 year old male who was admitted to our clinic for recurrent epileptic seizures, associated with cognitive impairment and a spastic left palsy. The seizures started since when he was 20 years old, with focal sensory and motor seizures. The frequency of these episodes at the beginning is unknown.

Subsequently, the frequency of the seizures escalated to 6 episodes per week. Various antiepileptic drugs were employed, consisting in levetiracetamum, carbamazepinum, valproic acid, phenitoinum, oxcarbazepinum. With this schedule, the patient still had 2-3 seizures per week.



a) *Clinical and paraclinical examinations*

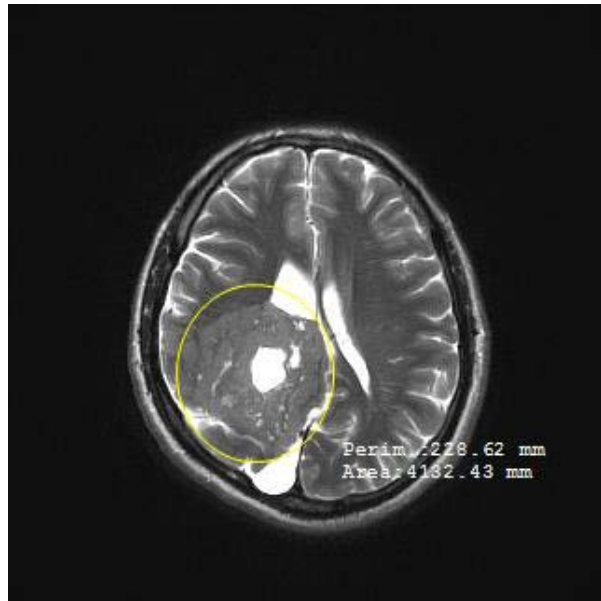
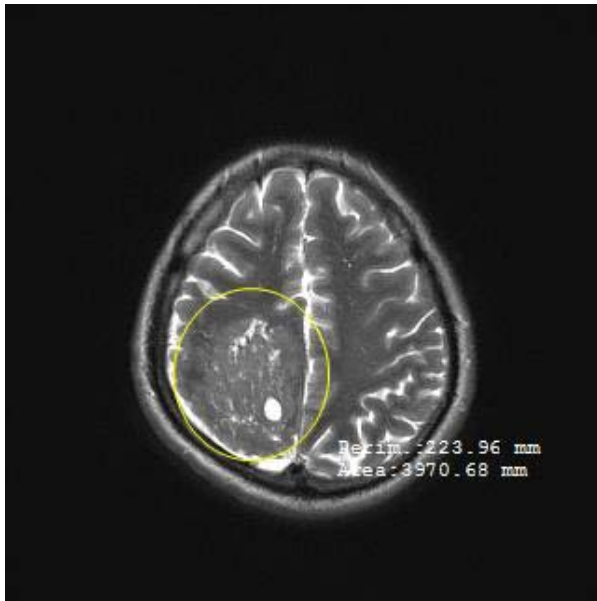
The personal and heredo-colateral history were unremarkable. The neurological examination shows bradylalia, bradipsychia, spastic left palsy - MRC of 3/5 and the pshychological examination shows a mild cognitive impairment.

b) *Interpretation of the cerebral MRI*

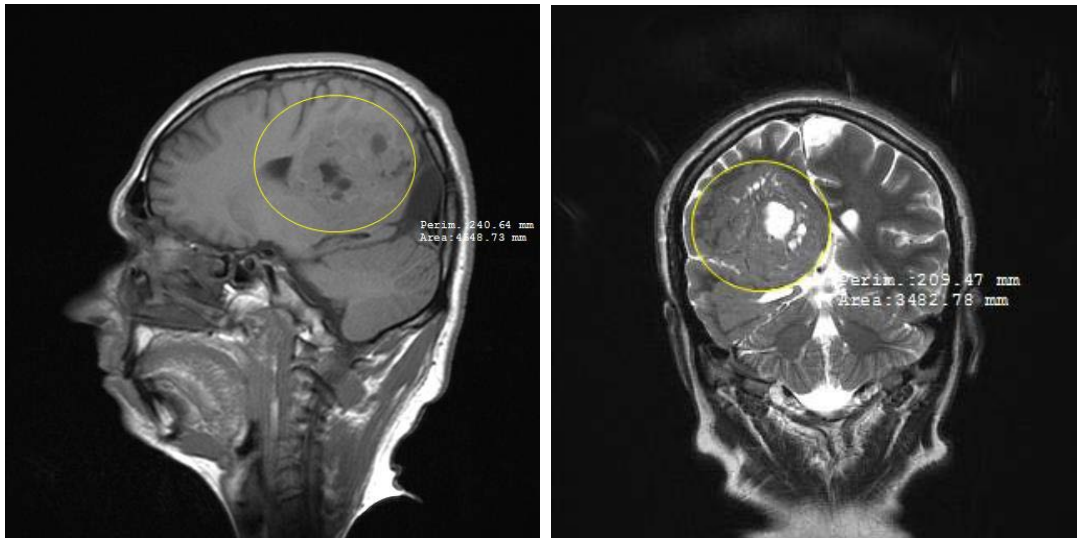
The brain MRI using Gadolinium shows a conglomerate of nodular grey matter, with bands of

white matter, alternately disposed, located in the right parietal lobes, spanning from the ventricular walls to the cortex surface. The nearby parietal and temporal cortexes show an underdeveloped gyrus and inadequate thickness.

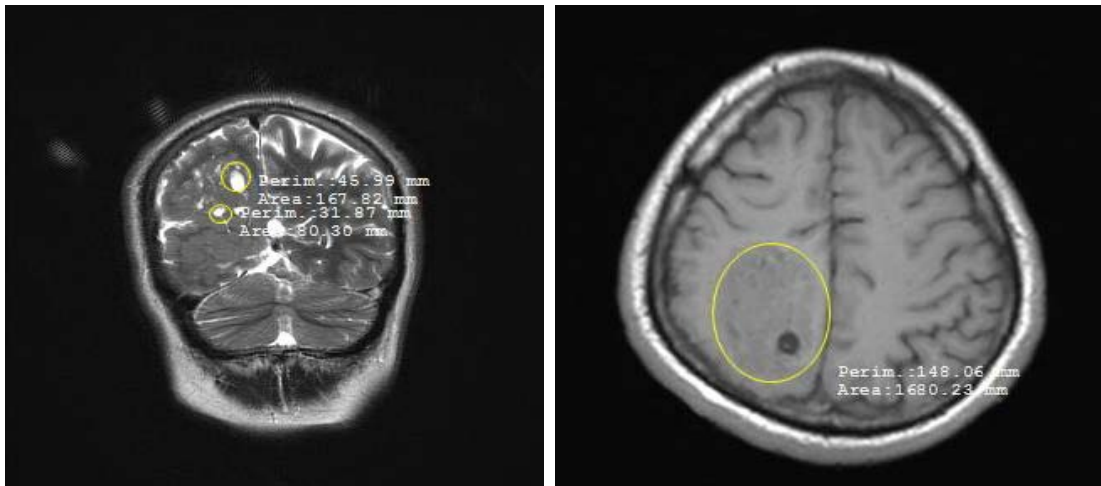
The corpus callosum has posterior agenesis and the lateral ventricle is imprinted.



No signal modifications are present within the cerebral parenchyma. Ventricular system appears normal in size. Arterial vascular paths from the base of the skull present normal MRI flow.



Lateral venous sinuses are normal in appearance.



There are no more modifications of the brain tissue. The ventricular system is normal. The arteries and dural venous sinuses appear normal.

Conclusion: Giant right sided temporo-parietal pseudotumoral heterothopia.

Treatment and evolution:

The current scheme contains the following drugs: Levetiracetamum (2000 mg daily) and Carbamazepinum (800 mg daily).

The patient still has partial motor-sensorial seizures, with a frequency of 2-3 episodes per week, mainly during the day.

The psychological examination shows a mild cognitive impairment.

The mental abilities have remained at the same level since 2014, when he was first diagnosed.

III. CONCLUSIONS

The case presented in the article shows the fact that the GMH has special clinical aspects- seizures, cognitive impairment, focal neurological palsy, but

characteristic aspects of the cerebral MRI, which reveals the cortical malformations.

The diagnosis was established after a long period of time, only due to MRI examination.

The patient is still monitored in order to determine the possible changes of treatment.

The case is isolated in the family.

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