Brain & Development 38 (2016) 302–309

BRAIN & DEVELOPMENT
Official Journal of the Japanese Society of Child Neurology

www.elsevier.com/locate/braindev

Original article

Three cases of right frontal megalencephaly: Clinical characteristics and long-term outcome

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Received 1 August 2015; received in revised form 8 September 2015; accepted 11 September 2015

Abstract

Aim: To delineate the clinical and neuroimaging characteristics of localized megalencephaly involving the right frontal lobe.
Method: Data from three patients aged 14–16 years at the last follow-up were retrospectively reviewed.
Results: All the patients were normal on neurological examination with no signs of hemiparesis. Enlargement of the right frontal lobe with increased volume of subcortical and deep white matter, as well as thickening of the ipsilateral genu of the corpus callosum was common. The onset of epilepsy was 4–7 years of age, with seizure types of massive myoclonus in two and generalized tonic-clonic in two, which could be eventually controlled by antiepileptics. Interictal electroencephalography showed frontal alpha-like activity in one, and abundant spike–wave complexes resulting in diffuse continuous spike–wave activity during sleep in two patients even after suppression of clinical seizures. Psychomotor development appeared unaffected or slightly delayed before the onset of epilepsy, but became mildly disturbed during follow-up period of 7–11 years.
Conclusion: Certain patients with right frontal megalencephaly can present with a milder epileptic and intellectual phenotype among those with localized megalencephaly and holohemispheric hemimegalencephaly, whose characteristic as epileptic encephalopathy was assumed from this study.

Keywords: Hemimegalencephaly; Frontal lobe; Localized megalencephaly

1. Introduction

Hemimegalencephaly (HME) is a rare type of congenital brain malformation, characterized by the enlargement of either cerebral hemisphere. HME is accompanied by dysplasia of the cerebral cortex with structures of polymicrogyria, agyria/pachygyria, and white matter heterotopia, which results in intractable epilepsy and profound disabilities [1,2]. For such cases, early surgical intervention, including hemispherectomy, is warranted to achieve a better outcome in the control of epilepsy and to promote psychomotor development [2].

Localized megalencephaly is now recognized as an enlargement limited to a small part of one cerebral hemisphere [3,4]. The occipital-lobe-predominant type,

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http://dx.doi.org/10.1016/j.braindev.2015.09.005

or posterior quadrantic dysplasia, is the most common type of localized megalencephaly. Although psychomotor developmental delays appear less severe in the posteriorly localized megalencephaly than the HME with holohemispheric involvement, complication of infantile spasms and/or daily, intractable seizures are common, and surgical resection of the megalencephalic region is often necessary for the control of epilepsy [3]. The occurrence of frontally localized megalencephaly is rare, and there is very little clinical information available on this condition [3]. Here we report three cases of the latter group for further recognition and characterization.

2. Method

Three patients (age 14–16 years, all males) with findings of localized, frontal megalencephaly on magnetic resonance imaging (MRI) (Fig. 1) were recruited from three medical institutes specialized in epilepsy management, and the clinical information was retrospectively reviewed. The study protocol was approved by the institutional review boards. Familial and perinatal histories and initial psychomotor development were unremarkable in all the patients. There was no consanguinity among parents of any of the patients. No evidence of macrocephaly, hemihypertrophy, or neurocutaneous syndromes was found in any patient. The age at diagnosis of focal megalencephaly ranged from 3 to 7 years, and in each case, the diagnosis occurred when the patient was receiving medical attention for the treatment of their epilepsy. Clinical details during follow-up period of 7–11 years are summarized in the following sections (see also Table 1). Genetic examinations such as G-band chromosomal analysis were not conducted for any patient.

For the quantitative assessment of the enlargement of the unilateral frontal lobe, an axial T1-weighted image was chosen from each patient, wherein the anterior horn of the lateral ventricles was the largest. In addition, the right and left frontal lobes as well as the frontal white

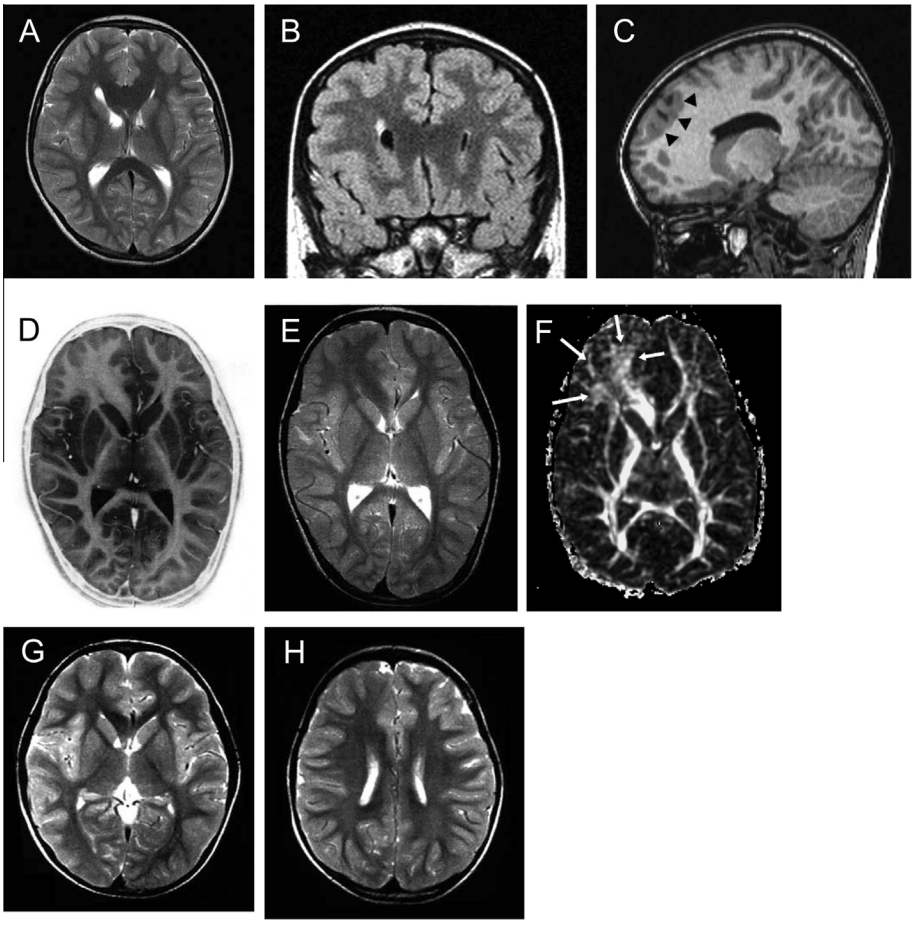

Fig. 1. Magnetic resonance imaging of patients with unilateral frontal lobe megalencephaly (A, E, G, H: T2-weighted images, B: fluid-attenuation inversion recovery image, C: T1-weighted image, D: T1 inversion recovery image, F: fraction anisotropy map) (A–C: patient 1, D–F: patient 2, G and H: patient 3) Right frontal lobes appear larger than left frontal lobes in these patients. Blurring gray-white matter boundary (D), cortical thickening with polymicrogyric contour (arrowheads in C), and increased volume of ipsilateral frontal white matter and genu of the corpus callosum (A, D, E, G), are noted. In F, increased signal intensity of the callosal genu, and enlarged frontal white matter (arrows) are noted in the right hemisphere.