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# An Unusual Case of Megalencephalic Leukoencephalopathy Together with Giant Subcortical Cysts

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**ABSTRACT**

**Introduction:** Megalencephalic leukoencephalopathy (MLC) is an uncommon neurodegenerative disorder affecting children. Common neurological findings include mental retardation, movement disorders related to cerebellar or extra-pyramidal dysfunction, macrocephaly, and epileptic seizures.

**Case Report:** A 4-year-old male presented with sudden-onset generalized tonic–clonic seizure to the emergency department at our university hospital. After the patient regained consciousness, abnormal neurological signs including macrocephaly, dysarthria, ataxia, and spasticity were detected. There were convulsive episodes that were diagnosed as epilepsy in his past medical history. Macrocephaly had first been noticed when he was 7 months old. In his follow-up examination, brain magnetic resonance imaging (MRI) revealed primary giant subcortical cysts in the bilateral temporal and frontal regions in his coronal T1-weighted image. Written informed consent was taken from the patient's parents.

**Conclusion**: Emergency physicians and pediatricians should maintain a high index of suspicion for MLC when treating children with macrocephaly and epileptic seizures in the emergency department or outpatient clinics. In particular, they should be considered in the differential diagnosis of subcortical cysts on performing brain MRI. Moreover, the early and accurate diagnosis of MLC will lead to genetic advisory assistance to the families of patients.

**Keywords:** Epileptic seizure, giant subcortical cysts, megalencephalic leukoencephalopathy

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## Introduction

Megalencephalic leukoencephalopathy (MLC) is an uncommon neurodegenerative disorder characterized by infantile-onset megalencephaly and cerebral leukoencephalopathy, particularly subcortical cysts in the anterior temporal area of the brain. Clinical manifestations of the disorder greatly vary (1). We present an uncommon case of MLC associated with giant subcortical cysts in this report.

## Case Report

A 4-year-old male was admitted with an abrupt attack of a generalized tonic–clonic seizure to the emergency department at our university hospital. Rectal diazepam was given as the first-line antiepileptic drug, and the patient was responsive to intravenous levetiracetam (loading dose of 20 mg/kg). After the patient regained consciousness, a neurological examination was performed. Abnormal neurological signs of the patient including macrocephaly, dysarthria, ataxia, and spasticity were detected.

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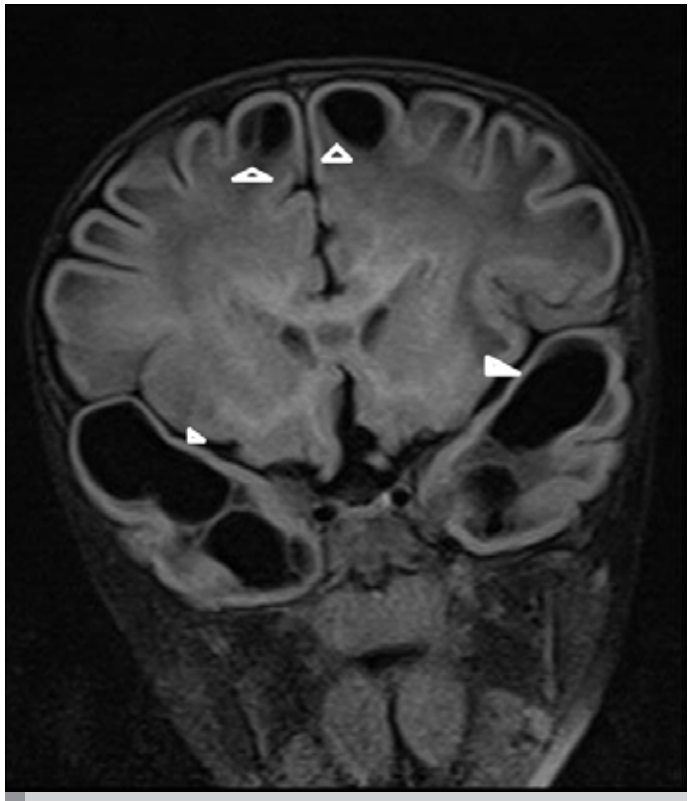

FIGURE 1. Giant subcortical cysts on performing brain magnetic resonance imaging

Further, optic nerve atrophy was diagnosed during an examination of the eyes. There were convulsive episodes diagnosed as epilepsy in his past medical history. However, he was not under regular treatment. Megalencephaly had first been noticed when he was 7 months old; he started walking and uttering single words when he was 18 months old. There was no family history of macrocephaly or neurological diseases.

In the patient's laboratory test results, hemogram parameters, serum sodium levels, liver function test results, and other serum electrolyte levels were normal. During his follow-up, brain magnetic resonance imaging (MRI) demonstrated primary giant subcortical cysts in the bilateral temporal and frontal areas in the coronal T1-weighted image (Figure 1). In addition, there was no abnormality in the metabolic test results. The clinical outcome was favorable after therapy. His seizures were controlled with antiepileptic medications, and patient is being followed up by the Department of Pediatric Neurology. Written informed consent was taken from the patient's parents.

## Discussion

Megalencephalic leukoencephalopathy (MLC) is an uncommon neurodegenerative disorder affecting children. Common neurological findings include movement disorders such as cerebellar or extrapyramidal dysfunction, speech movement, macrocephaly, and epileptic seizures (1, 2). Brain MRI findings are diagnostic. In most cases, brain MRI shows "swollen white matter" and diffuse symmetrical white matter changes in the cerebral hemispheres. Subcortical cysts are always present in the anterior temporal area and are also constantly observed in the frontoparietal region (3). The differential diagnosis of MLC contains disorders with increased volumes of white matter and macrocephaly such as Canavan disease, Alexander disease, L-2-hydroxyglutaric aciduria, and merosin-deficient congenital muscular dystrophy (4, 5).

As in the present case, brain MRI findings are typical and consistent with those of MLC. Moreover, brain MRI findings showed diffuse white matter changes with giant temporal cysts (Figure 1).

The disorder does not have any specific treatment. Studies have demonstrated that a mutation in the KIAA0027 gene termed as megalencephalic leukoencephalopathy with subcortical cysts 1 is responsible for MLC. Pathogenic mutations are considered to have a wide spectrum as marked clinical differences have been demonstrated among members of families known to have the mutation and in between families (6, 7). Genetic advisory assistance to families of these patients seems to be the most reasonable solution today (8).

Our patient had no family history of macrocephaly or neurological disorders despite the first-degree consanguinity of his parents. A genetic workup has been planned in our patient, and his family has been informed. In our patient, MLC associated with an epileptic seizure was diagnosed on the basis of clinical and radiological findings. Parenteral anticonvulsant therapy and intravenous fluid support were given in the emergency department. After diagnosis, treatment was initiated with oral levetiracetam (20 mg mg/kg/day) at our outpatient clinic. The clinical outcome was favorable after therapy.

## Conclusion

Emergency physicians and pediatricians should maintain a high index of suspicion for MLC when treating children with macrocephaly and epileptic seizures in the emergency department or outpatient clinics. In particular, they should be considered in the differential diagnosis of subcortical cysts on performing brain MRI. Moreover, the early and accurate diagnosis of MLC will lead to genetic advisory assistance to the families of patients.

**Informed Consent:** Written informed consent was obtained from the parents of the patient who participated in this study.

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