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Case Report

# CT and MRI findings in leptomeningeal melanocytosis

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ABSTRACT

Leptomeningeal melanocytosis is a rare cause of seizure in the pediatric population. Shown here is a case of this disease in a 9-year-old male who presented with seizures and minor trauma. Imaging showed progression of leptomeningeal enhancement in the setting of increased seizure activity, and biopsy confirmed the diagnosis. The patient received immunotherapy but eventually succumbed to the disease. This case serves as an educational tool to improve awareness of melanocytic proliferation as a differential consideration for leptomeningeal enhancement.



## Introduction

Leptomeningeal melanocytosis is a rare proliferative disorder of melanocytes in the central nervous system. Most commonly encountered in the pediatric population, this unfortunate pathology is typically fatal when encountered. Most literature dedicated to the study of this entity has been limited to case reports given its rarity.

The case provided here serves as an example of this rare disorder and depicts imaging findings that can educate radiologists and ordering providers about leptomeningeal melanocytosis. The therapeutic approach utilized is also included.

## Case report

A 9-year-old male presented to the emergency department after experiencing a 5-minute tonic-clonic seizure. Approximately an hour prior to the seizure, he fell and struck his left forehead resulting in a small cut.

CT of the head demonstrated gyriform regions of high attention throughout the posterior right hemispheric sulci (Fig. 1). Given the history of trauma, this was attributed to subarachnoid hemorrhage. He was placed on levetiracetam for seizure control and discharged home.

In the month following original presentation, the patient began to experience progressive left-sided weakness. An MRI

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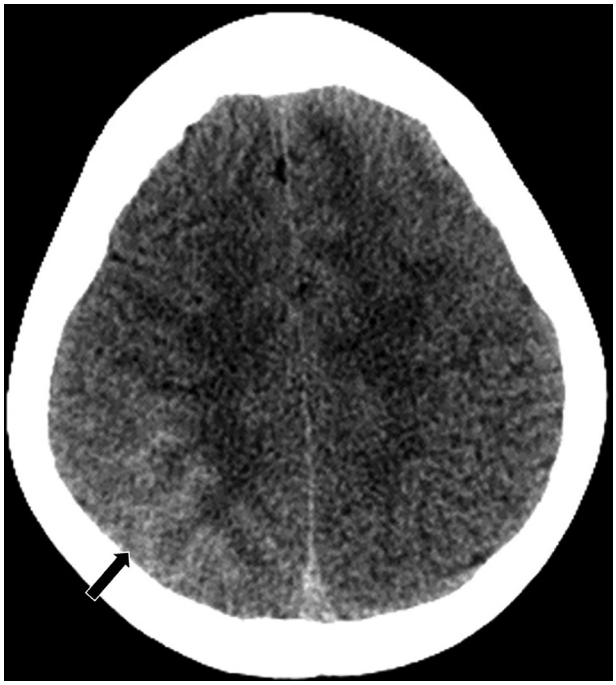

**Fig. 1 – Axial CT of the head demonstrates gyriform high attenuation material throughout the right posterior cortical sulci, greatest in the right parietal lobe (arrow). At the time of imaging, the patient was 9 years, 11 months old.**

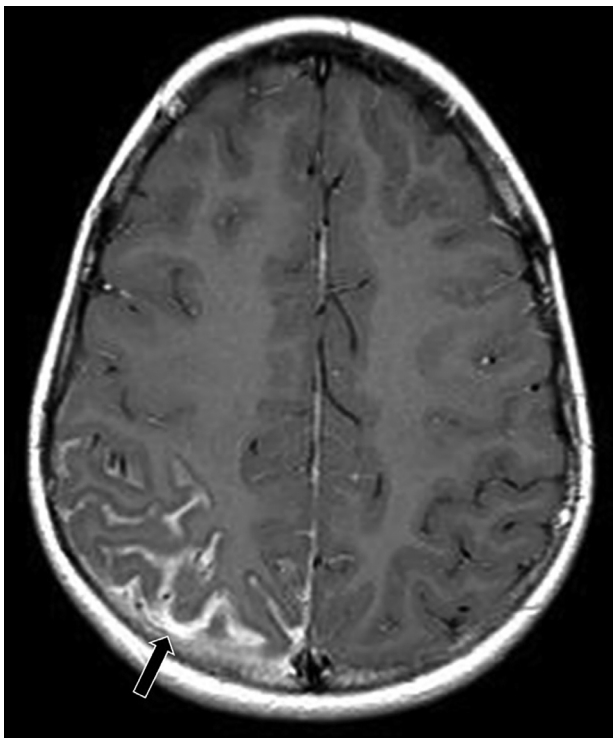

**Fig. 2 – Axial T1-weighted image of the brain at patient age 11 years, 0 months following intravenous gadolinium administration reveals leptomeningeal enhancement (arrow) through the abnormal area identified on CT.**

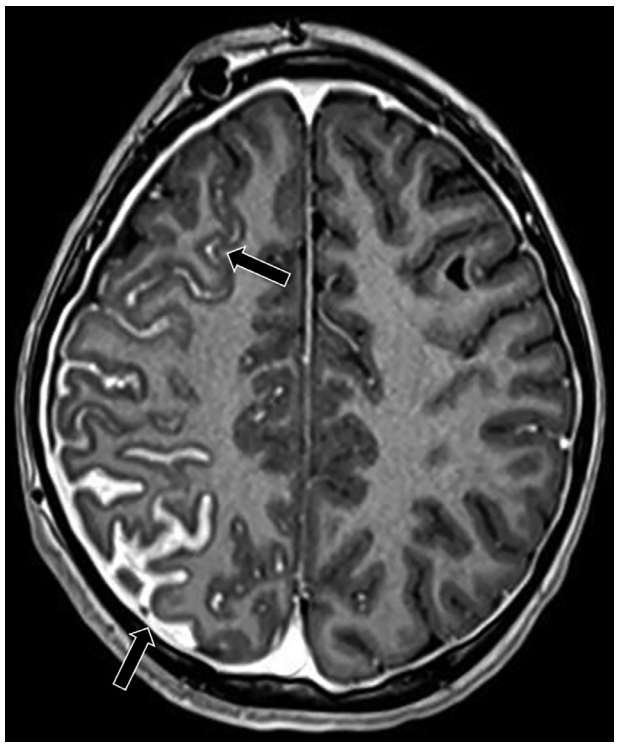

**Fig. 3 – Axial T1-weighted image of the brain following intravenous gadolinium administration at patient age 11 years, 5 months demonstrates disease progression with interval advancement of leptomeningeal enhancement (arrows).**

with and without contrast was ordered, and leptomeningeal enhancement was demonstrated throughout the right posterior cortical sulci corresponding to the high attenuation regions on the previous CT (Fig. 2). At this point, a presumptive diagnosis of Sturge-Weber disease was assigned, and he was followed by the neurology service.

The patient underwent an additional MRI of the brain with and without contrast 6 months following the original presentation, and the extent of leptomeningeal enhancement showed progression (Fig. 3). An open biopsy was next sought. Upon craniotomy, the affected surface of the brain exhibited a dark pigment. Biopsy confirmed the diagnosis of leptomeningeal melanocytosis (Fig. 4). Interestingly, the patient's mother had a history of melanoma diagnosed during pregnancy, however, the tumor cells recovered from the patient were shown to have an XY karyotype by fluorescent in situ hybridization on formalin-fixed paraffin-embedded neoplastic tissue. This means that vertical metastasis/maternal chimerism is unlikely. The patient's melanoma underwent Foundation One sequencing which demonstrated an activating mutation in NRAS.

After discussion of potential therapies including palliative craniospinal irradiation, MEK inhibitor therapy or immune-therapy, the patient was prescribed the combination of ipilumumab and nivolumab, anti-PD-1/immune modulators, as upfront therapy as this combination has the overall best reported 2-year survival to date [1–4]. The dominant tumor nodule in the right parieto-occipital region was treated

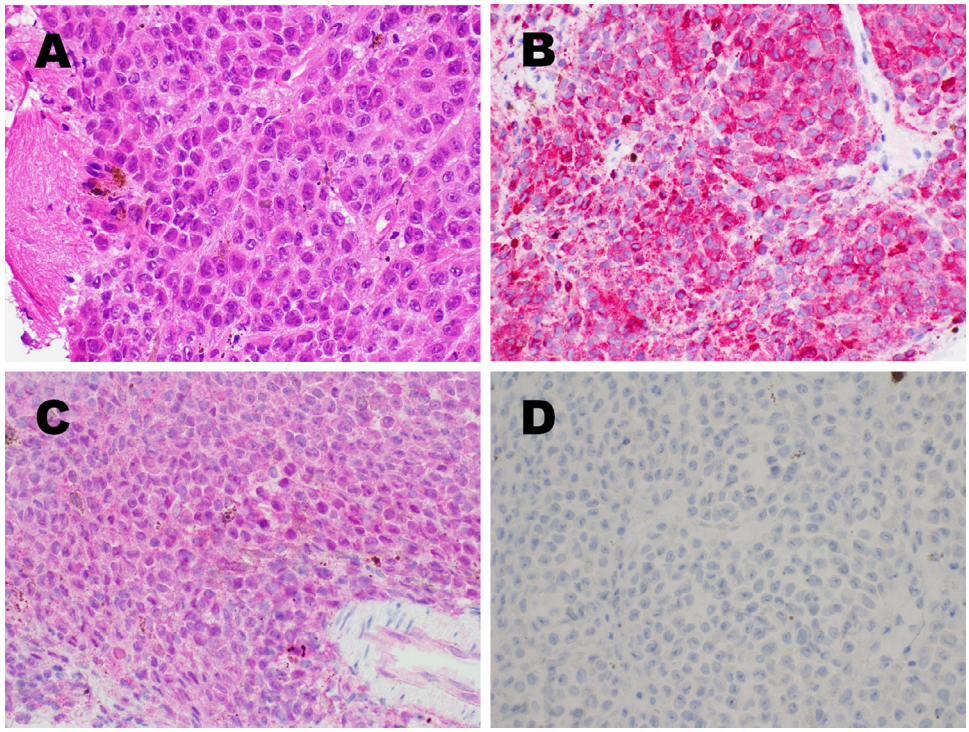


**Fig. 4 – Histologic photomicrographs acquired at 40X. (A) Hematoxylin/Eosin. (B) Positive Mart1/Melan-A. (C) S100 positivity. (D) SALL-4 negativity.**

concurrently with Cyberknife stereotactic radiosurgery to 18 Gy in 1 fraction. Repeat imaging after 2 months on therapy had demonstrated slight decrease in the radiated nodule, but progression of the leptomeningeal tumor as noted above. Unfortunately, the patient suffered florid progression of both leptomeningeal tumor and the primary tumor nodule 3 months into therapy and expired 1 year following original presentation.

## Discussion

Primary melanocytic tumors of the central nervous system are rare and arise from normally occurring melanocytes of neural crest cell origin found in the leptomeninges [5] and were first described in 1859 by Virchow [6]. This group of tumors may present as diffuse melanocytic proliferation such as with melanocytosis or meningeal melanomatosis, or as discrete circumscribed masses such as with melanocytoma or melanoma [7]. Leptomeningeal melanocytosis consists of diffuse melanocytic proliferation exhibiting histologically benign features without atypia, mitosis, necrosis, or invasion of brain parenchyma [5, 8]. It primarily affects the pediatric population. It is often also seen with giant congenital pigmented nevi in neurocutaneous melanosis [8]. It can occur at any age but is most common in patients under 10 years of age, as was the patient described here at the time of presentation.

Patients may present with seizures, vomiting, intracranial hypertension, and neurologic deficits. The seizures described in this patient were typical for this disease. Despite their histologically benign appearance, leptomeningeal melanocytosis is associated with a poor prognosis. There is no definitive treatment, and palliative management includes tumor debulking and ventricular shunt placement to relieve hydrocephalus [9].

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