

Acute Promyelocytic Leukemia With Intracranial Myeloid Sarcoma: A Rare Pediatric Case And Diagnostic Challenge

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Abstract

A rare case of isolated intracranial myeloid sarcoma (MS) with PML: RAR α fusion was identified. Histopathological examination of surgically resected brain tissue demonstrated a malignant neoplasm with limited lineage specificity. DNA-based next-generation sequencing detected the PML:RAR α fusion, which enabled definitive diagnosis of isolated intracranial MS with this molecular alteration. Surgical resection followed by systemic therapy guided by acute promyelocytic leukemia (APL) protocols achieved durable remission in this patient. A rare case of isolated intracranial myeloid sarcoma (MS) with PML:RAR α fusion was identified. Histopathological examination of surgically resected brain tissue demonstrated a malignant neoplasm with limited lineage specificity. DNA-based next-generation sequencing detected the PML:RAR α fusion, which enabled definitive diagnosis of isolated intracranial MS with this molecular alteration. Surgical resection followed by systemic therapy guided by acute promyelocytic leukemia (APL) protocols achieved durable remission in this patient.

Introduction

Myeloid sarcoma (MS) is an extramedullary malignant neoplasm composed of myeloid blasts or immature myeloid cells and may arise at virtually any anatomical site [1]. Isolated MS is defined by the absence of prior or concurrent leukemia and by negative bone marrow findings at diagnosis, with an estimated annual incidence of approximately 0.7 cases per million children [2]. Accurate diagnosis of isolated MS remains challenging and depends on confirmation of myeloid lineage in tumor cells [2]. A rare case of isolated intracranial MS in a 15-year-old female with PML:RAR α fusion is presented, in which remission was achieved following treatment guided by the Chinese Children's Cancer Group Acute Promyelocytic Leukemia 2017 (CCCG-APL-2017) protocol.

Case Report

A rare case of isolated intracranial MS with PML:RAR α fusion was identified in a 15-year-old female who presented with a 2-week

history of persistent headache and swelling of the right cheek. Magnetic resonance imaging revealed a 4.7 × 4.6 × 4.8 cm mass in the right temporal region (**Figure 1**). Neuronavigation-guided craniotomy was performed to achieve surgical resection. Histopathological evaluation demonstrated a small, round-cell malignant neoplasm with areas of necrosis. Immunohistochemical (IHC) staining of core biopsy material showed that immature myeloid cells were positive for myeloperoxidase and Ki-67 (15%) and negative for CD34, CD30, CD68, CD138, CD20, and CD163. DNA-based targeted next-generation sequencing of the resected tissue confirmed the presence of PML:RAR α fusion with a positive rate of 44.57%. Fluorescence in situ hybridization demonstrated 82% positivity for the same fusion (**Figure 2**). Peripheral blood counts, coagulation parameters, bone marrow examination, and cerebrospinal fluid (CSF) analysis were unremarkable, establishing the diagnosis of isolated intracranial MS with PML:RAR α fusion.

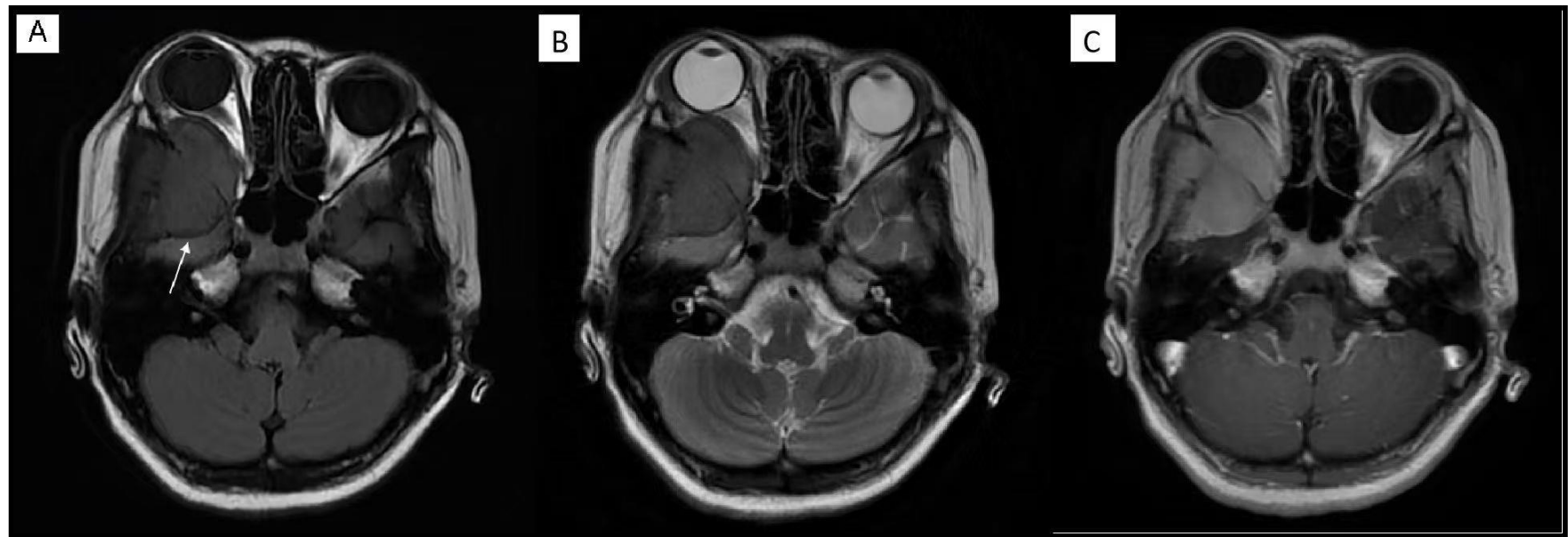


Figure 1: Magnetic Resonance Imaging (MRI) Of The Brain.

A: T1-weighted MRI showing an isointense lesion (mass) in the right middle cranial fossa, involving adjacent meninges and temporal bone (white arrow).

B: T2-weighted MRI showing low signal intensity of the lesion.

C: Contrast-enhanced MRI showing marked enhancement of the lesion (mass).

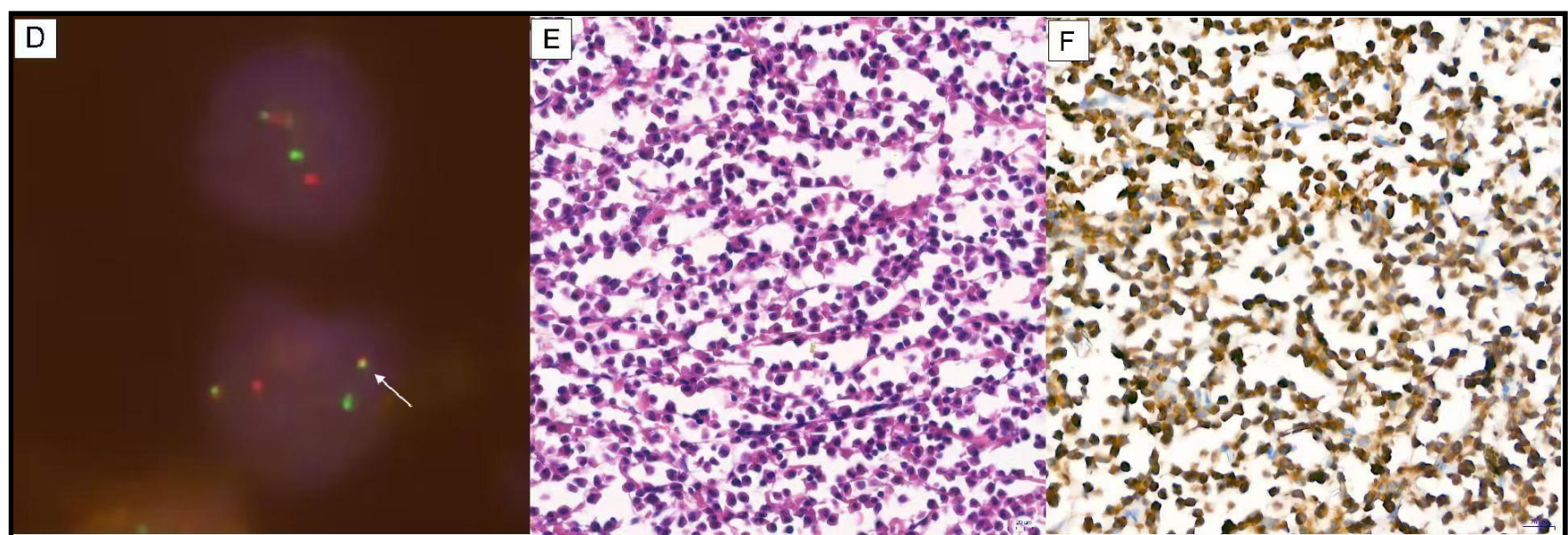


Figure 2: Histopathological and molecular characterization of brain tissue.

D: Fluorescence in situ hybridization analysis of resected brain tissue demonstrating positivity for the PML:RAR α fusion gene (white arrow).

E: Hematoxylin and eosin (H&E) staining ($\times 40$) showing diffuse infiltration of abnormal promyelocytes with moderate cell size, abundant eosinophilic cytoplasm, and folded nuclei in a subset of cells.

F: Myeloperoxidase (MPO) staining ($\times 40$) demonstrating MPO-positive tumor cells.

Postoperatively, headache symptoms improved; however, swelling of the right temporal region, diplopia caused by impaired abduction of the right eye, and numbness of the right facial area persisted for several months. Treatment following the CCCG-APL-2017 protocol was initiated [3]. Radiotherapy was not administered due to the complete surgical resection of the tumor. Three courses of intrathecal triplet therapy consisting of methotrexate, cytarabine, and dexamethasone were administered. CSF cytology, protein analysis, and flow immunophenotyping remained normal. After one cycle of chemotherapy with arsenic trioxide (ATO) and all-trans-retinoic acid (ATRA), swelling and sensory symptoms resolved gradually, and complete molecular remission was maintained at 14-month follow-up.

Discussion

This case was exceptional in that isolated intracranial MS with PML:RAR α fusion was identified in a pediatric patient, a presentation not previously reported in the published literature (PubMed). Acute myeloid leukemia (AML) presenting as isolated MS is rare, accounting for approximately 1% of cases [4]. In acute promyelocytic leukemia (APL), MS occurs infrequently and most often at relapse, whereas presentation as isolated extramedullary disease without bone marrow involvement is exceedingly uncommon [5]. The PML:RAR α fusion is typically associated with APL, an aggressive leukemia subtype for which durable remission is achieved in 98% of patients treated with ATRA/ATO therapy [6, 8]. In contrast, isolated intracranial MS with PML:RAR α fusion represents an exceptionally rare clinical scenario, and histopathology alone lacks sufficient specificity for definitive diagnosis. Although H&E staining may

suggest promyelocytic morphology, morphological features alone are inadequate to establish the diagnosis in the absence of molecular confirmation. Molecular profiling using next-generation sequencing and related assays therefore plays a critical role in accurate classification of this disease. The favorable therapeutic response observed in this case supports the concept that MS with PML:RAR α fusion shares core differentiation and apoptosis mechanisms with APL and responds effectively to ATRA/ATO-based therapy [7]. This outcome provides a rational treatment reference for future patients presenting with similar clinicopathological features.

Conclusion

Isolated MS typically progresses to systemic disease within three to six months in the absence of treatment [9, 11]. This case underscores the importance of combining surgical resection with systemic therapy guided by APL protocols to achieve durable remission and offers a therapeutic framework for similar future cases.

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participants for their contributions to the study.

Conflicts Of Interest: The authors of this manuscript declare no potential conflicts of interest.

Ethics Consideration

This study was approved by the IRB of the Institute of Hematology & Blood Diseases Hospital, Chinese Academy of Medical Sciences (Approval No. IIT2016008-EC-3). Written informed consent-to-disclose was obtained from the patient.

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Author Contributions

Ye Guo: Contributed to study design and drafting of the article.

Shuchun Wang and Ning Wang: Contributed to patient management and drafting of the article.

Xiaojuan chen: Contributed to study design, drafting of the article, and data analysis and interpretation.

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