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Incidental Detection of Wyburn-Mason Syndrome Mimicking Extranodal Involvement in a Patient with Hodgkin Lymphoma: A Diagnostic Challenge

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Abstract

Wyburn-Mason syndrome (WMS) is an extremely rare congenital, non-hereditary neurocutaneous disorder characterized by arteriovenous malformations (AVMs) involving the retina, midbrain, and occasionally the facial structures. Although often asymptomatic, such vascular anomalies may incidentally mimic malignant lesions on functional imaging. A sixteen-year-old girl was diagnosed with stage IIB nodular sclerosis-type Hodgkin lymphoma (HL). During initial staging, positron emission tomography-computed tomography (PET-CT) revealed increased fluorodeoxyglucose uptake and thickening along the left optic nerve, suggesting extranodal involvement. Orbital magnetic resonance imaging demonstrated a stable vascular lesion consistent with WMS. No histopathologic confirmation was possible due to the lesion's location. Following standard European Network for Paediatric HL-C1 therapy (oncovin, etoposide, prednisone, adriamycin / cyclophosphamide, oncovin, prednisone, dacarbazine) and standard involved-site radiotherapy, the lesion remained unchanged. To our knowledge, the coexistence of WMS and HL has not been previously reported. This case highlights the diagnostic challenge posed by congenital AVMs that can mimic malignant infiltration on functional imaging. Recognition of this potential overlap is crucial to avoid overstaging and unnecessary therapeutic escalation. This case highlights a diagnostic pitfall in pediatric oncology and emphasizes the importance of correlating multimodal imaging findings to differentiate congenital vascular malformations from malignant extranodal disease, thereby avoiding overstaging or unnecessary treatment escalation.

Keywords: Wyburn-Mason syndrome, Hodgkin lymphoma, arteriovenous malformation, PET-CT, orbital MRI, diagnostic pitfall

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Introduction

Wyburn-Mason syndrome (WMS) is a rare neurocutaneous disorder characterized by unilateral vascular malformations involving the retina, midbrain, and sometimes facial structures[1,2]. The condition arises from an embryologic defect in the development of the optic pathway and its associated vasculature.

Hodgkin lymphoma (HL) is a highly curable pediatric malignancy in which accurate staging is critical for risk-adapted therapy. Fluorodeoxyglucose (FDG) positron emission tomography-computed tomography (PET-CT) is the cornerstone of initial staging and response assessment in HL and plays a decisive role in treatment stratification[3]. However, FDG uptake is not specific to malignancy and may also be observed in inflammatory or vascular conditions. Although rare, congenital vascular malformations may mimic extranodal lymphoma involvement and pose a significant diagnostic challenge[4].

Case Presentation

A sixteen-year-old girl presented with left supraclavicular lymphadenopathy measuring 3 cm. Excisional biopsy revealed nodular sclerosis-type HL. Laboratory studies, including erythrocyte sedimentation rate, were within normal limits. She reported no fever or night sweats but had a 12% weight loss in one month. Staging FDG PET-CT demonstrated increased uptake and thickening along the left optic nerve, raising concern for extranodal involvement. Quantitative maximum standardized uptake value measurements were not available, as the PET-CT images were reviewed retrospectively and the original report did not include numerical metabolic values. Orbital magnetic resonance imaging (MRI) revealed contrast-enhancing vascular structures in the same region, consistent with a congenital arteriovenous malformation (AVM) (**Figure 1**).

According to the Ann Arbor staging system, the patient was classified as stage IIB and was treated with the European Network for Paediatric HL-C1 protocol, consisting of two cycles of oncovin, etoposide, prednisone, adriamycin followed by two cycles of cyclophosphamide, oncovin, prednisone, dacarbazine chemotherapy. Involved-site radiotherapy (21.6 Gy in 12 fractions) to the mediastinal and cervical regions was administered as part of standard treatment for stage IIB HL, rather than because of an inadequate metabolic response.

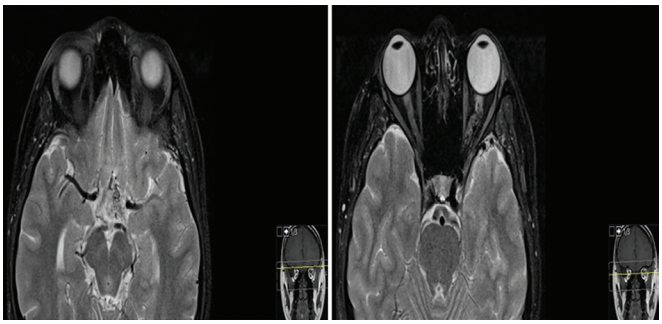

Figure 1. Axial T2-weighted orbital MRI images demonstrating vascular malformation along the left optic nerve, consistent with Wyburn-Mason syndrome
MRI; Magnetic resonance imaging

Follow-up MRI showed no change in the optic lesion, confirming a congenital etiology. Given the asymptomatic nature of WMS, no specific treatment was initiated. Written informed consent was obtained from the patient's parents for publication of this case report and accompanying images.

Discussion

WMS is an exceptionally rare, non-hereditary congenital vascular disorder characterized by AVMs involving the retina, brain, and occasionally orbital or facial structures. Fewer than 100 cases have been reported in the literature, and the clinical presentation varies widely depending on the location and extent of vascular involvement[5-7]. Many patients remain asymptomatic, and the condition is often detected incidentally during imaging performed for unrelated reasons.

The clinical spectrum varies depending on the site and extent of AVMs. Ocular or orbital lesions may cause proptosis or visual disturbances, but many patients remain asymptomatic[7,8].

In pediatric HL, FDG PET-CT plays a central role in initial staging and response assessment, directly influencing risk stratification and treatment planning. However, FDG uptake is not specific to malignancy. Increased metabolic activity may also be observed in inflammatory processes or benign vascular anomalies, creating potential diagnostic pitfalls. In this case, focal FDG uptake along the optic nerve initially raised concern for extranodal involvement by lymphoma, which could have led to disease upstaging and treatment intensification if it had been interpreted in isolation.

Subsequent orbital MRI demonstrated imaging features consistent with a congenital vascular malformation, and the lesion showed no radiologic progression during chemotherapy and follow-up. Importantly, the optic nerve lesion remained morphologically unchanged despite the administration of systemic chemotherapy and standard involved-site radiotherapy, which were given as part of routine treatment for stage IIB HL, rather than in response to inadequate metabolic remission. This stability strongly supported a benign congenital etiology and argued against malignant infiltration.

The underlying mechanism of FDG uptake in AVMs is not fully understood, but is thought to be related to increased regional blood flow, endothelial metabolic activity, and low-grade inflammatory processes rather than true neoplastic involvement. Similar cases of benign vascular lesions mimicking malignancy on PET-CT have been reported sporadically in adults; reports in pediatric patients, particularly in association with HL, are exceedingly rare[6].

This case underscores the importance of multimodal imaging correlation in pediatric oncology. Functional imaging findings should always be interpreted alongside high-resolution anatomic imaging, especially when lesions are located in atypical sites for lymphoma involvement. Awareness of congenital vascular malformations as potential mimics of extranodal disease is essential to prevent overstaging, unnecessary invasive procedures, or overtreatment.

Although angiography remains the diagnostic gold standard for vascular malformations, a non-invasive diagnostic approach was considered appropriate in this asymptomatic pediatric patient. The characteristic MRI findings, combined with long-term radiologic stability during and after lymphoma therapy, supported the diagnosis of WMS without exposing the patient to additional procedural risk.

To our knowledge, this is one of very few reported pediatric cases of incidental detection of WMS during staging for HL. The case highlights an important diagnostic consideration for pediatric oncologists and radiologists, and emphasizes the need for cautious interpretation of FDG PET-CT findings in the presence of rare congenital vascular anomalies.

Conclusion

This case illustrates an important diagnostic pitfall in pediatric HL, where a rare congenital vascular malformation may mimic extranodal disease on FDG PET-CT. The incidental detection of WMS in our patient underscores the limitations of functional imaging when used in isolation. Careful correlation with high-resolution anatomic imaging, particularly MRI, is essential to distinguish benign congenital anomalies from malignant involvement. Awareness of such rare mimickers is crucial to avoid disease overstaging, unnecessary invasive procedures, and inappropriate treatment escalation in pediatric oncology practice.

Ethics

Informed Consent: Written informed consent was obtained from the patient's parents for publication of this case report and accompanying images.

Footnotes

Author Contributions: Yiğit Y: Concept, Design, Data Collection or Processing, Analysis or Interpretation, Literature Search, Writing; Çınar C: Surgical and Medical Practices, Concept; Bahar S: Literature Search; Özgüven AA: Literature Search.

Conflict of Interest: The authors have no conflicts of interest to declare.

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