



Co-Existence of Multiple Sclerosis and Germinoma in an Adult Male: A Case Report

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INTRODUCTION

- Central nervous system (CNS) germinoma
 - Found in suprasellar, infundibular and pineal regions of brain
 - Patients present with headaches, vision changes, fatigue
 - Diagnosis based on clinical presentation and neuroimaging, then confirmed with biopsy
- Multiple sclerosis (MS)
 - Autoimmune disease of the CNS characterized by inflammation and demyelination
 - Diagnosis depends on clinical presentation, MRI and CSF findings
- Concurrent diagnosis has not been reported in the literature

PURPOSE

The purpose of this study is to describe the rare dual presentation of MS and CNS germinoma in an adult male and discuss the complexities of treating both diseases simultaneously.

CLINICAL PRESENTATION

- 28-year-old man with history of hypertension initially presents to Ophthalmology with a 2-year history of progressive, bilateral vision loss
 - Poor visual acuity and bilateral optic atrophy on exam
- Referred to Neurology for evaluation of possible MS
 - T2 FLAIR hyperintense lesions involving the corpus callosum and the periventricular white matter on brain MRI, suggestive of demyelinating plaques
 - Elevated IgG index and 4 CNS specific oligoclonal bands, normal protein and cells on lumbar puncture
 - Patient diagnosed with MS
- 3 months later, patient presents to outside hospital with severe right-sided headache
- Patient transferred to our hospital after CT imaging revealed hydrocephalus and intracranial mass
 - Intraventricular mass in posterior third ventricle, obstructive hydrocephalus, and worsening hyperintense lesions on MRI
- Endoscopic third ventriculostomy performed to treat hydrocephalus
 - Biopsy of mass confirmed CNS germinoma diagnosis

CASE CONTINUED

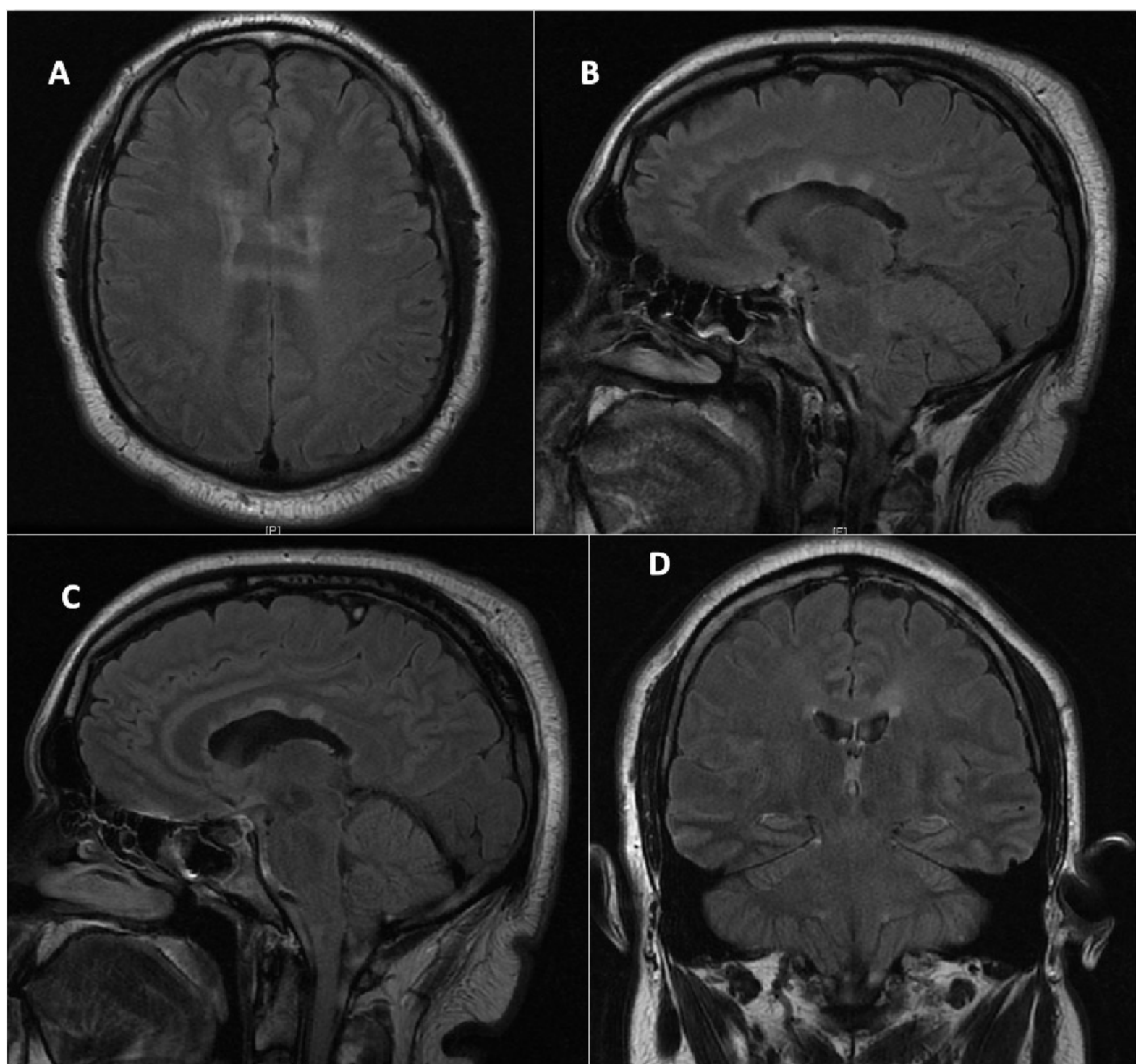


Figure 1: Initial MRI showing hyperintense corpus callosal and periventricular lesions

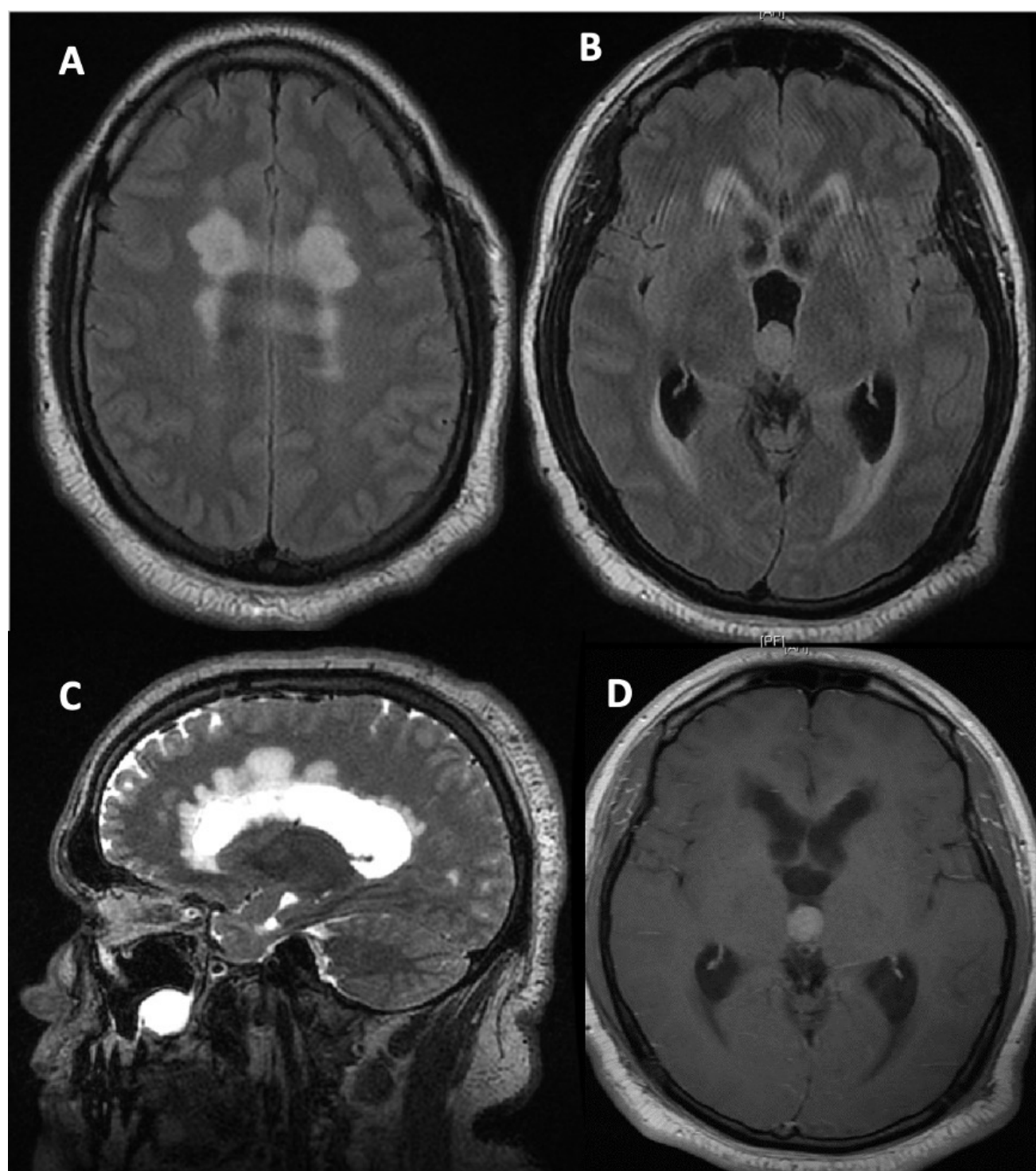


Figure 2: Subsequent MRI showing worsening lesions, ovoid 3rd ventricular mass, & obstructive hydrocephalus

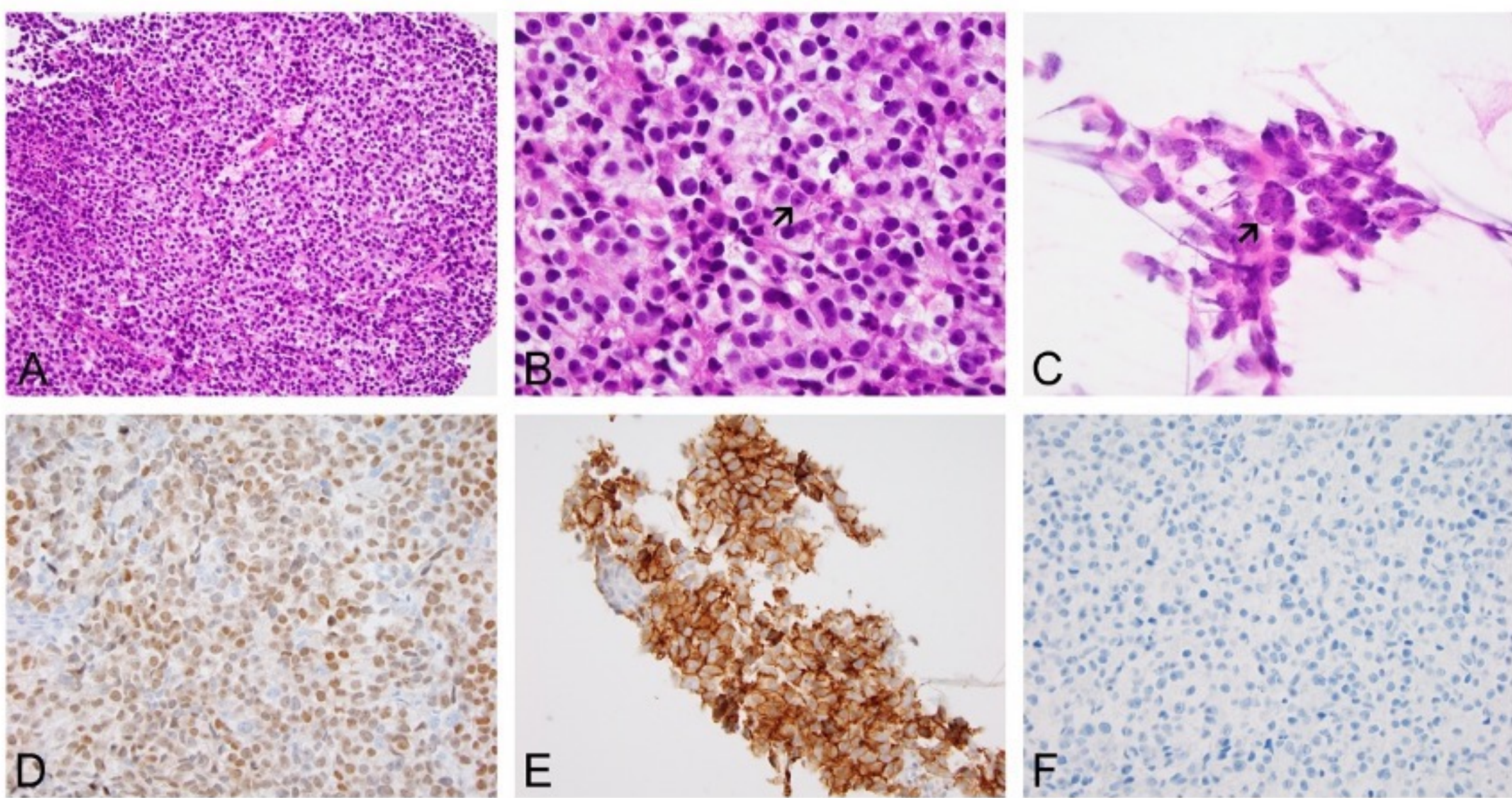


Figure 3: (a) and (b) are low- and high-magnification. Prominent nucleoli highlighted by arrows in (b) and (c). (d), (e) and (f) are immunohistochemistry for CNS germinoma markers OCT3/4, CD117, and H3 K27M, respectively

CASE CONTINUED

- Post-Operative Course
 - Neurologic exam normal post-operatively & discharged after 2 days
 - Carboplatin + etoposide administered for cytoreduction
- Brain MRI at 4-months post-op
 - Showed resolution of hydrocephalus, periventricular T2 hyperintense changes and decreased size of corpus callosum plaques
- Serum studies to rule out paraneoplastic optic neuritis
 - Markers anti-CRMP-5 and anti-amphiphysin were negative
- At the time of this presentation, patient had not undergone adjuvant radiation treatment nor started disease modifying treatment for MS

DISCUSSION

- To our knowledge, no instance of true simultaneous presentation of CNS germinoma and MS in the same patient has been described in the scientific literature.
- Due to the rarity of this dual diagnosis, a broad differential was considered:
 - Paraneoplastic optic neuritis
 - Germinoma as an MS mimic
- Treatment plan
 - Focused on addressing the CNS germinoma before managing MS
 - Completed four cycles of carboplatin + etoposide with near complete response
 - Patient is slated to begin radiation therapy shortly, followed by IVIG therapy to slow vision loss in MS.

CONCLUSIONS

Concurrent presentation of MS and CNS germinoma is exceptionally rare. After careful consideration of the clinical presentation of our patient, combined with imaging and biopsy, a final dual diagnosis was made. Development of a treatment plan for co-existing MS and CNS germinoma is complex, but emphasis was ultimately placed on treating the tumor, followed with targeted MS therapy.

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