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Metastatic testicular primitive neuroectodermal tumor of the orbit

Abstract:

Background: Orbital metastases are rare, occurring in 2–5% of systemic malignancies, and are exceptionally uncommon from testicular germ cell tumors (GCTs). Diagnosis is often delayed, as symptoms are nonspecific and precede primary cancer diagnosis in up to 32% of cases. No prior report has described orbital metastasis secondary to a testicular GCT arising from a teratoma.

Case: We present a novel case of a 21-year-old male with widespread metastatic testicular GCT involving the skull base, orbit, and optic canal, who developed acute, vision-threatening compressive optic neuropathy. After initial orchiectomy at outside hospital and liver biopsy on hospital day 8, initiation of oncologic therapy was delayed pending final pathology. Given a reassuring early ophthalmologic exam and suspected diagnosis of embryonal carcinoma—a chemosensitive tumor—surgical resection was initially deferred. However, daily ophthalmic monitoring revealed rapid visual decline by hospital day 20, prompting emergent multidisciplinary approach with orbitotomy and craniotomy for mass debulking and optic nerve decompression. Postoperative exams demonstrated immediate and sustained visual improvement. Molecular testing was concerning for medulloblastoma arising from a teratoma.

Conclusion: This is the first reported case of orbital metastasis from a testicular GCT of teratomatous origin, which required emergent surgical decompression for vision preservation. The case demonstrates the importance of comprehensive systemic evaluation, including of the genitourinary tract, in young men with nonspecific orbital symptoms. Also, close ophthalmic monitoring during diagnostic workup is essential to detect rapid visual decline and allow for timely surgical intervention in the setting of metastatic disease.

Biography

Megan is a second-year medical student at the Perelman School of Medicine, University of Pennsylvania, who aspires to be an ophthalmologist