Unusual ocular cases: Systemic illnesses, diagnostic challenges.

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Introduction

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This collection of case reports highlights a diverse range of ophthalmic conditions, often presenting with unusual manifestations or linked to systemic diseases, underscoring the complexities of ocular diagnostics and treatment. One case describes an atypical presentation of chronic ocular toxoplasmosis in an immunocompetent adult, showing peripheral retinal non-perfusion and optic neuropathy, which are uncommon for the disease. This emphasizes the need for extensive diagnostic workup, including multimodal imaging, to accurately identify rare ocular infections and guide treatment approaches [1].

Another report details chronic progressive external ophthalmoplegia (CPEO) associated with mitochondrial DNA deletions, stressing the importance of genetic investigation for neuromuscular disorders impacting ocular motility. It provides a review of clinical aspects, diagnostic methods, and management strategies for this intricate condition [2].

Similarly, a patient experienced bilateral optic neuritis following a SARS-CoV-2 infection, suggesting a potential connection between the virus and demyelinating neurological complications affecting the optic nerve. This adds to our growing understanding of COVID-19's varied ophthalmic sequelae and calls for increased clinician awareness [3].

Ocular involvement can often be the initial sign of severe systemic conditions, a theme explored across several reports. For instance, acute myeloid leukemia (AML) presented as bilateral orbital infiltration in a rare case. This particular finding compels ophthalmologists and oncologists to consider systemic hematological malignancies when unusual or rapidly progressing orbital lesions are encountered, as timely diagnosis profoundly affects patient outcomes [4].

An infrequent case of sarcoidosis exclusively affecting the optic nerve, without other systemic symptoms, further illustrates diagnostic challenges. It requires a high level of suspicion and advanced imaging to distinguish it from other optic neuropathies and enable prompt sight-preserving treatment [5].

In another example, acute lymphoblastic leukemia (ALL) in an adult first manifested with ophthalmic symptoms, specifically subretinal fluid and optic disc edema. This scenario stresses that ocular signs can be crucial initial indicators of serious systemic illnesses, prompting ophthalmologists to investigate hematological malignancies in patients with unexplained papilledema or retinal fluid for earlier intervention [6].

Vascular complications and their management are also key areas of focus. One report discusses acute bilateral central retinal artery occlusion (CRAO) occurring shortly after COVID-19 vaccination, suggesting a possible adverse event and contributing to the knowledge base of post-vaccination ocular complications. It highlights the need for vigilance and further research into immune-mediated vascular events linked to vaccinations [7].

A complex case of retinal arteriolar macroaneurysm, complicated by both vitreous and submacular hemorrhage, demonstrates successful surgical management. The combined approach of vitrectomy and subretinal tissue plasminogen activator proved effective in resolving severe hemorrhagic issues and maintaining macular function, providing a valuable treatment paradigm for challenging retinal vascular conditions [8].

Additionally, an unusual instance of bilateral spontaneous central retinal vein occlusion (CRVO) was documented as a complication of Graves' disease. This points to a potential link between autoimmune thyroid disease and retinal vascular occlusions, advising clinicians to evaluate for underlying systemic autoimmune conditions in patients with atypical or bilateral CRVO [9].

Finally, the diagnostic challenges of rare ocular infections are brought to light. An extremely rare case of fungal keratitis caused by Lecythophora mutabilis, a pathogen seldom implicated in human ocular infections, underscores the difficulty in identifying uncommon fungal agents. This emphasizes the critical importance of laboratory identification and targeted antifungal therapy for effective management of such unique and potentially sight-threatening ocular infections [10].

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Conclusion

This collection of ten case reports provides a comprehensive look into various unusual ophthalmic presentations, encompassing rare infections, systemic diseases with ocular manifestations, and challenging vascular conditions. The reports highlight an atypical chronic ocular toxoplasmosis with peripheral retinal non-perfusion and optic neuropathy, and a case of chronic progressive external ophthalmoplegia linked to mitochondrial DNA deletions. They also detail bilateral optic neuritis following a SARS-CoV-2 infection, emphasizing the evolving understanding of COVID-19's impact on the eye.

Crucially, several cases illustrate ocular symptoms as initial indicators of severe systemic illnesses, such as acute myeloid leukemia presenting with bilateral orbital infiltration and acute lymphoblastic leukemia manifesting with subretinal fluid and optic disc edema. An unusual instance of bilateral central retinal vein occlusion secondary to Graves' disease further reinforces this connection. Additionally, the collection covers vascular complications like acute bilateral central retinal artery occlusion after COVID-19 vaccination and the surgical management of a complex retinal arteriolar macroaneurysm. The diagnostic challenges of rare infections are also underscored by a case of fungal keratitis caused by Lecythophora mutabilis. Together, these reports stress the importance of thorough diagnostic workups, interdisciplinary awareness, and advanced treatment strategies for complex and rare ocular disorders, ultimately aiming to preserve vision and improve patient outcomes.

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