Bilateral Retinal Vein Thrombosis and Cerebral Amyloidoma: Unraveling a Complex CAA-ri Case

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Aim

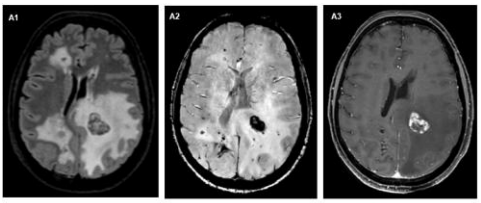
To describe a case of CAA-related inflammation (CAA-ri) presenting with a tumor-like mass lesion and bilateral retinal vein thrombosis.

Methods

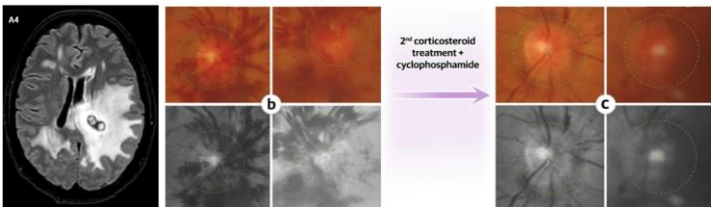
Cerebral Amyloid Angiopathy (CAA) is a small vessel disease characterized by cerebrovascular deposition of β-amyloid, typically associated with lobar intracerebral hemorrhage (ICH) and cognitive impairment. A rare inflammatory subtype, CAA-related inflammation (CAA-ri), presents with rapid cognitive deterioration, seizures, headaches, and distinctive MRI findings, including T2-hyperintense lesions. Here, we report the case of a 56-year-old woman who presented initially with transient postural instability.

Results

Neurological examination was unremarkable, except brisk deep tendon reflexes on the right arm and leg. Brain MRI revealed a left parietal cortico-subcorotical lesion, hyperintense on T2/FLAIR and hemosiderin deposits on SWI sequences, suggestive of CAA-ri. Due to mild symptoms, regular neuroradiological monitoring was advised. However, in the following months, the patient developed gait disturbances, right lower limb weakness, and confusion. Repeat MRI showed a left centrum semiovale lesion with contrast enhancement and perilesional edema. Amyloid-PET confirmed focal cerebral amyloidosis in the left temporo-occipital region. Cerebrospinal fluid analysis revealed low Aβ42 and elevated Tau, supporting amyloid pathology. Genetic analysis revealed ApoE genotype e3/e3. Empirical high-dose IV steroids led to initial clinical improvement, but new symptoms occurred during steroid tapering (visual field deficits and left arm paresthesias). Ophthalmologic evaluation revealed bilateral retinal vein thrombosis with retinal haemorrhages and papilledema, a rare CAA-ri manifestation. Therefore, steroids were escalated, followed by mycophenolate mofetil (2 g/day). A brain biopsy confirmed amyloidoma with gliosis, microglial activation, rare lymphocytic infiltrates and Thioflavine S-positive amyloid deposits. However, due to a lack of clinico-radiological response to immunosuppressive therapies and worsening of ocular symptoms, cyclophosphamide (1000 mg/m²) was initiated, resulting in complete resolution of retinal vein thrombosis despite stable brain MRI findings.



Axial T2/FLAIR (A1) brain MRI showing asymmetric bilateral parietal and right frontal hyperintensities within the subcortical white matter, that reduced after treatment (A4). The left parietal alteration is consistent with an amyloidoma, showing mass effect, signal loss on SWI (A2), and irregular ring-enhancement on post-contrast T1-weighted images (A3). Axial SWI scan (A4) also showing multiple microbleeds in the subcortical white matter bilaterally and one right parieto-occipital intracerebral hemorrhage (A2). FLAIR = fluid-attenuated inversion recovery; SWI = susceptibility-weighted imaging.



Fundus photographs centred in both visible-light colour and near-infrared monochrome modalities showing hemorrhagic retinal manifestations and papilledema during corticosteroid tapering (B); marked improvement after intravenous cyclophosphamide at 1 month follow-up (C).

Discussion

Cerebral amyloidoma is characterized by a focal, tumor-like accumulation of amyloid glycoproteins in neural tissue and has a benign prognosis. CAA-ri may have various clinical and radiological presentations, including retinal vein thrombosis and may co-exist with cerebral amyloidoma. The variable response to immunosuppression underscores the need for a biopsy in steroid-refractory cases and atypical radiological presentations to rule out mimics. Steroids and MMF provided transient benefit, while cyclophosphamide addressed refractory inflammation.

References

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