A rare presentation: Presumed Susac Syndrome

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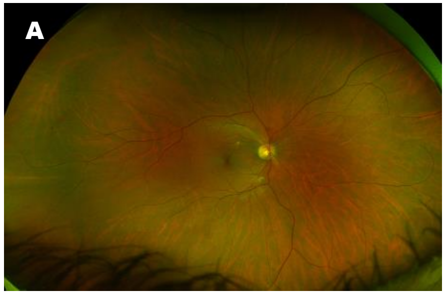
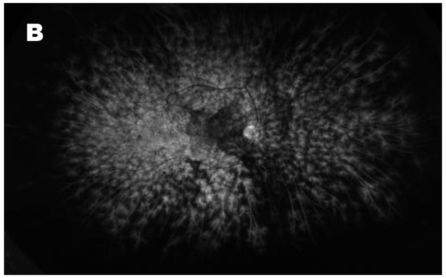
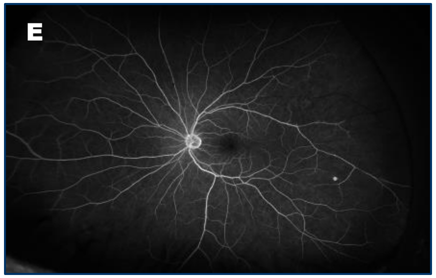
Clinical Presentation

A 37yo white male with PMH heavy EtOH abuse presented to ED with presumed withdrawal symptoms.
Given recurrent falls and worsening mentation, MRI brain was performed and showed multiple subcortical and brainstem areas of restricted diffusion raising concern for acute infarcts.
Further stroke work up was negative. Hypercoagulable and malignancy workups were negative.
Epileptiform activity was suspected and ruled out.
Worsening encephalopathy prompted repeat imaging which demonstrated spread and worsening of the lesions; there was concern for CNS vasculitis.
Extensive serum and CSF studies were unremarkable/inconclusive.
He was started on pulse dose solumedrol for 5 days followed by prednisone taper, as well as IVIG for suspected NMDA encephalitis, after which mental status improved.
Despite improvement, he did not follow verbal commands and was found to have profound sensorineural hearing loss on audiogram.
Given this development, ophthalmology was consulted to assess the fundus; initial evaluation at bedside was unremarkable other than two CWS OU.
Further evaluation in clinic was revealing of the findings presented to the right.
Communication was achieved by writing on a white board, and he speaks responses.
The current presumed diagnosis is Susac Syndrome.
Diagnosis and treatment have been challenging and have been led by Rheumatology and Neurology, with input from Medicine, ENT, Ophthalmology, and Infectious Disease.
Current proposed treatment is Prednisone taper, q4w IVIG infusion, Cellcept, and initiation of Rituximab. He is gradually improving. Prognosis is uncertain.

Fundus Photos and Fluorescein Angiography







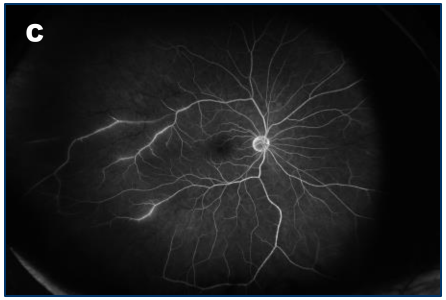


Figure D: Fundus photo OS demonstrating 2 CWS along ITA, occlusion of mid-distal arterioles along STA, incidental punched out RPE temporal retina.
Figure E: FA at 3m 57s demonstrating poor capillary perfusion, 360 midperipheral branch and twig vascular nonperfusion greatest in the ST retina and associated with venous attenuation and perivenular phlebitis.

Figure A: Fundus photo OD demonstrating resolving perivascular CWSs of the nasal macula, occlusion of the nasal arterioles.
Figure B: FA at 45s demonstrating severely delayed choroidal perfusion, focal patchy perfusion of choriocapillaris, delayed central retinal artery perfusion.
Figure C: FA at 7m 15s demonstrating severe nonperfusion of the midperiphery associated with frequent branch and twig vascular occlusions, temporal vasculitis of the venules.

Discussion

Susac Syndrome is a rare triad of encephalopathy, sensorineural hearing loss, and retinal microangiopathy. Pathogenesis is unknown but proposed to be autoimmunity against the endothelium of capillaries with anti-endothelial cell antibodies present in ~25% of cases[1], with preference for the pre-capillary arterioles[2,3]. Incidence is in the 2nd to 4th decades of life with caucasian and female predominance. Symptoms generally present sequentially with encephalopathy appearing first, followed by visual, then auditory deficits. Treatment usually includes high dose steroids and IVIG followed by immune suppression. Prognosis is variable and dependent on the severity and duration of presentation, as well as early aggressive initiation of treatment.

Differential diagnosis includes ADEM, MS, Vasculitides, Thromboembolic events, CADASIL (Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy).

This patient fits the typical SS demographic. His presentation differs somewhat given the presence of venous leakage, potentially indicating an underlying vasculitis, which is not well represented in the literature but has been reported[4]. While encephalopathy was clearly his leading manifestation, he next developed sensorineural hearing loss. Despite profound profusion deficits in his retinas, he has not thus far demonstrated objective or subjective visual deficits. VF perimetry is a reasonable diagnostic tool to monitor for the evolution of deficits, however his mental status is a limiting factor presently.

References

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