CASE REPORT

Urodynamic findings in Susac syndrome: first reported filling cystometrogram

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Accepted 20 June 2017

SUMMARY

Susac syndrome is a rare neurological disease, with only 300 cases reported in the literature. Lower urinary symptoms are not an uncommon feature of the disease, yet there is no information on specific dysfunction typical urodynamic findings associated with the disease. We present what we believe to be the first reported filling cystometrogram study of Susac syndrome for the evaluation of voiding dysfunction.

BACKGROUND

Susac syndrome was first described by Susac in 1979 in a young female patient who presented with a triad of encephalopathy, retinal arteriolar branch occlusion and sensorineural deafness.[1] Susac syndrome occurs three times more in women than men and usually occurs between the second and fourth decade of life.[2 3] It is an autoimmune disease, specifically an autoimmune endotheliopathy, which leads to damage of small blood vessels in the brain, retina and inner ear. The underlying cause of Susac syndrome is unknown. A variety of clinical manifestations of Susac syndrome outside the classic triad have been described and these include urological symptoms. These are predominantly related to urgency and urge incontinence, which occur in approximately 9% of the patients.[2]

CASE PRESENTATION

We present a 35-year-old man with a six history of Susac syndrome Initial symptoms included increasing confusion, ataxia and hearing loss. The patient had right macular, inferior, and superior branch retinal arterial occlusions, and left inferior branch occlusion. MRI study performed at the time of diagnosis demonstrated tiny lesions involving the periventricular white matter and central corpus collosum, consistent with the diagnosis of Susac syndrome.

The patient's main urinary symptoms included severe urgency and urge incontinence. He also complained of daytime frequency with small volume voids. He reported nocturia several times per night. The patient did however report good flow, without hesitancy or intermittency. He denied haematuria, urinary tract infections or dysuria. On examination his abdomen and external genitalia were normal. Digital rectal examination showed a small, normal prostate, while flexible cystoscopy demonstrated a normal appearing urethra and bladder.

Videourodynamic studies were carried out in accordance with good urodynamic practices. We were unable to obtain initial uroflowmetry due to low bladder volume. Filling cystometrogram demonstrated high-amplitude detrusor overactivity (DO) starting at 40 mL with pressures >200 cmH_2O. This was associated with urge leakage and complete bladder emptying secondary to DO (figure 1). A pressure flow study could not be satisfactorily performed, as the bladder could not be filled past 80 mL despite multiple fills at low fill rate to obtain an appropriate voiding phase study. Fluoroscopy showed a trabeculated bladder outline, but open bladder neck and patent urethra, without evidence of any vesicoureteric reflux (figure 2). A renal ultrasound confirmed no hydronephrosis. The patient was commenced on mirabegron 50 mg daily, and at 2 month follow-up, he had responded well to the beta-3 agonist (mirabegron 50 mg daily). He reported marked improvement in urgency, urge incontinence, frequency and nocturia. He is able to hold larger urinary volumes and reports no increased difficulty emptying his bladder. He is currently satisfied with this treatment. Should the current treatment fail, oral anticholinergics or intravesical onabotulinumtoxinA would be reasonable next steps in the treatment algorithm.

DISCUSSION

Susac syndrome is associated with the clinical triad of encephalopathy, retinal arteriolar branch occlusions and sensorineural deafness.[1 4] However, the triad is rarely complete at the onset of disease thus diagnosis does not rely solely on the presence of the clinical triad.[4 5] Most patients present with central nervous system symptoms initially, and there is usually a lag of approximately 5 months before the triad is noted to be complete.[4 5] Diagnostic investigations often used include: MRI brain, retinal fluorescein angiography of the retina and audiometry. Classical MRI findings include T2 hyperintense lesions in the corpus callosum, periventricular area and cerebellum, among other areas in the brain.[6] Fluorescein angiography is used to help detect retinal artery involvement in Susac syndrome.[2] Treatment strategies in Susac syndrome are aimed at immunosuppressive and immunomodulatory therapy.[2 4] In the case described, the treatment regime included infliximab infusions, prednisolone and mycophenolate.

Other manifestations of Susac syndrome have been described that include urinary symptoms mentioned in a number of cases.[2] However,

To cite: Nzenza TC, Hoag N, Hennessey DB, *et al*. *BMJ Case Rep* Published Online First: [*please include* Day Month Year]. doi:10.1136/bcr-2017-220200

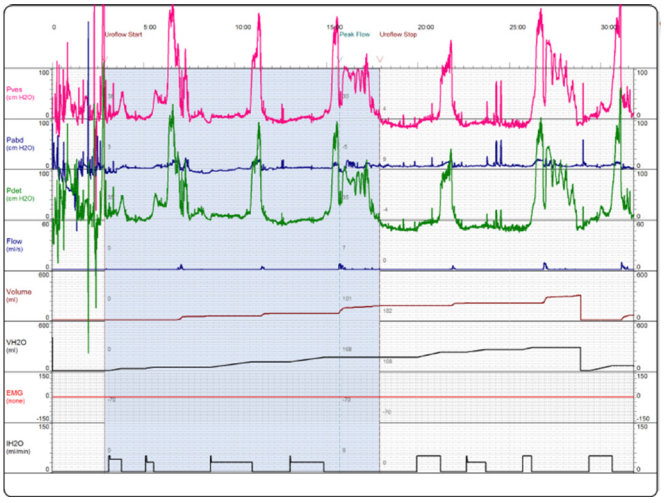

Figure 1 Filling cystometrogram demonstrating high-amplitude detrusor overactivity with leakage.

minimal information exists in the literature with respect to the extent of urinary symptoms in Susac syndrome. It appears that the most common urinary symptom is urgency, which has been hypothesised to be associated with encephalopathy. To the best of our knowledge, this is the first report of urodynamic findings in a case of Susac syndrome. Cystometrogram showed severe DO with urge incontinence at low volumes and a synergic bladder and an external urethral sphincter on fluoroscopic imaging. Unfortunately, the severity of DO precluded adequate urinary volume for pressure flow studies. We would postulate that voiding function is maintained in the patient presented and that his symptoms relate entirely to urine storage. It is not clear whether this is representative of voiding dysfunction seen in Susac syndrome. However, the filling cystometrogram represents the first reported urodynamic tracing in a patient with Susac syndrome. More cases describing urological manifestations of Susac syndrome are needed to appreciate the true extent of urinary involvement.

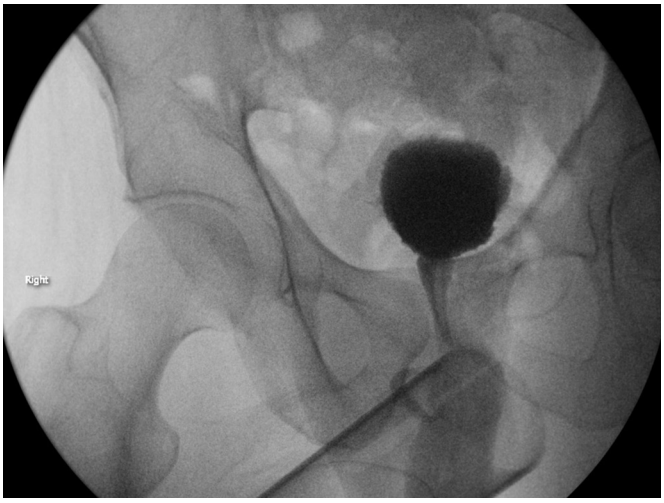

Figure 2 Fluoroscopic image demonstrating a trabeculated bladder during detrusor overactivity with open bladder neck and patent urethra.

Learning points

- Susac syndrome has a clinical triad of encephalopathy, retinal arteriolar branch occlusions and sensorineural deafness.
- A variety of clinical manifestations of Susac syndrome outside the classic triad have been described which include urological symptoms.
- First reported urodynamic tracing in a patient with Susac syndrome.

Contributors All authors: involved in analysing the case and results and explaining the results; preparing the manuscript; critical revision of the manuscript; provided final approval of the version to be published and are guarantors. JG and NH: case management.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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