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CEREBRAL VENOUS SINUS THROMBOSIS AS AN INITIAL MANIFESTATION OF MIXED CONNECTIVE TISSUE DISEASE: A CASE REPORT

General Medicine

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ABSTRACT

Mixed connective tissue disease (MCTD) is a rare autoimmune overlap disorder characterized by features of multiple connective tissue diseases along with anti-U1 ribonucleoprotein antibody positivity (Sharp et al., 1972). Neurological involvement is uncommon, and cerebral venous sinus thrombosis (CVST) as an initial presentation is rarely reported (Sanna et al., 2003).We present a case of a 35-year-old female who presented with acute onset severe headache and generalized seizures, preceded by a history of polyarthralgia, body swelling, and sicca symptoms. Neuroimaging revealed cerebral venous sinus thrombosis with haemorrhagic venous infarction (Stam, 2005). Further evaluation showed hypothyroidism and positive autoimmune markers including anti-U1 RNP, SSA, and SSB antibodies, confirming MCTD (Ortega-Hernandez & Shoenfeld, 2012). Due to the presence of haemorrhagic infarction, anticoagulation was withheld, and the patient was managed with corticosteroids, antiepileptics, osmotherapy, and supportive care. The patient improved clinically on follow-up. This case highlights the importance of considering autoimmune etiologies in patients presenting with CVST, especially when associated with systemic features (Bousser & Ferro, 2007).

KEYWORDS

Mixed Connective Tissue Disease, Cerebral Venous Sinus Thrombosis, Autoimmune Disorder, Anti-u1 Rnp

INTRODUCTION

Mixed connective tissue disease (MCTD) is a systemic autoimmune condition characterized by overlapping features of systemic lupus erythematosus, systemic sclerosis, and polymyositis, along with anti-U1 ribonucleoprotein antibody positivity *(Sharp et al., 1972)*. Common manifestations include arthralgia, Raynaud's phenomenon, and pulmonary involvement *(Ortega-Hernandez & Shoenfeld, 2012)*. Neurological manifestations are relatively rare in MCTD *(Sanna et al., 2003)*. Cerebral venous sinus thrombosis (CVST) is an uncommon cerebrovascular disorder caused by thrombosis of dural venous sinuses, leading to impaired venous drainage and raised intracranial pressure *(Stam, 2005)*. Its association with autoimmune diseases is recognized but uncommon *(Bousser & Ferro, 2007)*.

We report a rare case of MCTD presenting initially as CVST with haemorrhagic venous infarction.

Case Report

A 35-year-old female presented with generalized body swelling for 3–4 months, initially involving the face and lower limbs and later becoming generalized. She also reported generalized body ache, multiple joint pains, and dryness of eyes. She was a known case of hypothyroidism with irregular treatment.

She developed a severe holocranial headache for 3–4 days, throbbing in nature, associated with vomiting and worsening on bending forward. One day prior to admission, she had generalized tonic–clonic seizures with altered sensorium.

On examination, pulse was 56/min and blood pressure was 140/90 mmHg. Glasgow Coma Scale was E4V5M6. No focal neurological deficits were noted. Clinical findings suggested raised intracranial pressure.

Ophthalmological evaluation revealed severe dry eyes and early papilledema.

MRI brain showed haemorrhagic venous infarction. MR venography demonstrated partial thrombosis of posterior dural venous sinuses and straight sinus. The left transverse sinus was hypoplastic.

Table 1: Laboratory Investigations

Parameter	Findings
Haemoglobin	Low
ESR / CRP	Elevated
Ferritin	Elevated
Thyroid Profile	Hypothyroidism
ANA	Positive
Anti-U1 RNP	Positive
SSA / SSB	Positive
Rheumatoid Factor	Elevated
Urine Examination	Trace proteinuria

Based on clinical, radiological, and immunological findings, a diagnosis of MCTD presenting as CVST was made.Due to haemorrhagic infarction, anticoagulation was withheld. The patient was treated with corticosteroids, antiepileptics, mannitol, thyroid hormone replacement, and supportive care. She showed gradual improvement.

DISCUSSION

MCTD is a rare autoimmune overlap syndrome with variable clinical presentation *(Ortega-Hernandez & Shoenfeld, 2012)*. Neurological involvement is uncommon and may present diagnostic challenges *(Sanna et al., 2003)*.

CVST is a rare form of stroke, commonly presenting with headache, seizures, and altered consciousness *(Stam, 2005)*. Its association with autoimmune diseases may be explained by chronic inflammation, endothelial dysfunction, and prothrombotic mechanisms *(Bousser & Ferro, 2007)*.

In this case, the presence of anti-U1 RNP antibodies and overlapping autoimmune features confirmed MCTD *(Sharp et al., 1972)*. Sicca symptoms and thyroid dysfunction further supported autoimmune involvement.

Management of CVST typically involves anticoagulation; however, in cases with haemorrhagic infarction, treatment must be individualized *(Bousser & Ferro, 2007)*. Our patient responded well to conservative management.

CONCLUSION

Cerebral venous sinus thrombosis can be an unusual initial presentation of mixed connective tissue disease. Early recognition of underlying autoimmune etiology is important for appropriate management and improved outcomes.

Declaration

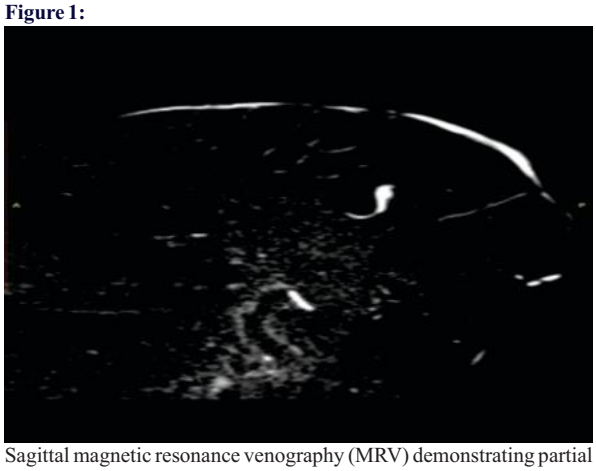
The authors declare that there is no conflict of interest regarding the publication of this paper. This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

Patient Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. Efforts have been made to ensure patient anonymity, and no identifying information has been disclosed.

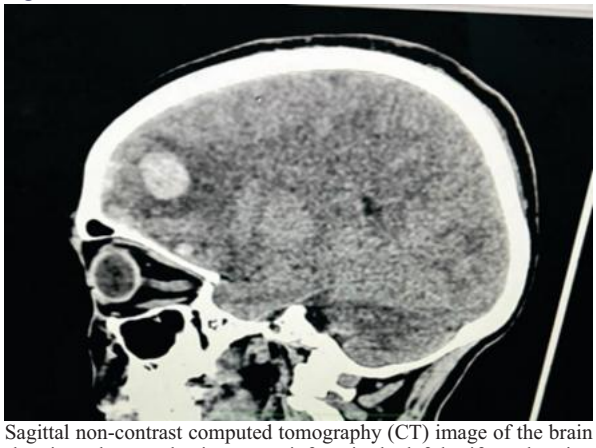
IMAGES

Figure 1:



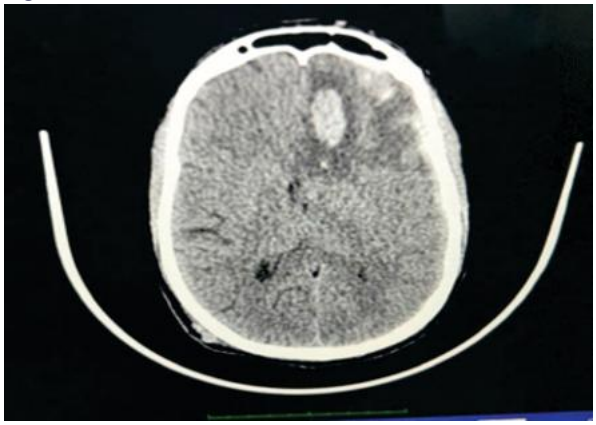
Sagittal magnetic resonance venography (MRV) demonstrating partial thrombosis of the superior sagittal sinus with reduced flow signal, consistent with superior sagittal sinus thrombosis.

Figure 2:



Sagittal non-contrast computed tomography (CT) image of the brain showing a haemorrhagic venous infarct in the left basifrontal region with surrounding mild vasogenic edema.

Figure 3:



Axial non-contrast computed tomography (CT) image of the brain showing a haemorrhagic venous infarct in the left high frontal region with surrounding edema.

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