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MOVING TO CONNECTIVE TISSUE DISEASE

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A 76 year old retired seamstress presented with jerky limb movements, increasing difficulty swallowing solids more than liquids, and progressive weight loss of 4 stone over 8 months. She had presented to medical services on 3 occasions during that time, but each time was discharged home. Investigations had included; plain chest radiograph, oesophago-gastro-duodenoscopy(OGD), CT-body, tumour markers, and CT-PET scan, all of which were unremarkable. Repeat OGD now revealed oesophageal dysmotility.

She had stigmata of connective tissue disease; small joint swelling, sclerodactyly, raynaud's phenomenon, malar rash, and microstomia. Additionally, she had stimulus-sensitive myoclonus, and evidence of peripheral neuropathy. MRI brain showed widespread small vessel disease including brainstem disease. Serum ANA was positive, with a high titre of anti-dsDNA antibodies. Anti-histone antibodies, cANCA and pANCA were negative, ESR was less than 5. Therefore, given her clinical stigmata, mixed connective tissue disease (CTD) was confirmed by Rheumatology and she is being evaluated for immunosuppression.

CTDs present with a wide spectrum of neurological disease, the most common being encephalopathy (35–75%), psychosis, seizures, stroke due to small vessel vasculitis, non-compressive myelopathy, peripheral neuropathy, myopathy, cranial nerve involvement and fatigue.

We present an approach to the neurological presentations of CTD and reveal possible manifestations and important treatment considerations.