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ADULT ONSET LEUKOENCEPHALOPATHY DUE TO CSF1R GENE MUTATION

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We report on a 44 year old Afro-Caribbean lady who presented with progressive personality change and cognitive impairment over one year. She had progressive difficulty managing household tasks, lost interest in her hobbies and spoke very little. Her family described her behaviour as 'child-like'. She had been diagnosed with anti-Ro positive mild Lupus 10 years previously and was taking mepacrine. Examination suggested a predominantly frontal subcortical pattern of cognitive impairment. MRI/A revealed confluent symmetrical white matter changes in the frontal and parietal lobes with patchy restricted diffusion in the basal ganglia. Extensive other investigations including

metabolic tests, autoimmune tests, and CSF analysis were unrevealing. A right frontal brain biopsy revealed non-specific gliosis. She was treated empirically with steroids and cyclophosphamide for the possibility of cerebral vasculitis but there was no subsequent clinical improvement. She tested positive for a heterozygous mutation in the CSF1R gene, first described in Hereditary Diffuse Leukoencephalopathy with Spheroids. Adult onset leukoencephalopathies are difficult to investigate given their broad clinical presentation and wide variety of acquired and rare genetic aetiologies. This case is instructive because it highlights a novel genetic test which may obviate the need for brain biopsy or harmful immunosuppressive agents in rapidly progressive leukoencephalopathies.