DETECTION OF GENE VARIANTS IN CHRONIC FATIGUE SYNDROME PATIENTS

Arthy Narayanan (Alan Light)

Department of Anesthesiology

Myalgic Encephalomyelitis/Chronic Fatigue Syndrome (ME/CFS) and Fibromyalgia (FM) are chronic conditions often present in the same patient, characterized by severe physical fatigue, widespread muscle and joint pain, mental fog and post-exertional worsening of symptoms. Patients with ME/CFS and FM experience difficulty in performing daily activities and in some cases are wheelchair-bound. In a pilot sample, the majority of ME/CFS patients had elevated autoantibodies to autonomic receptors and also showed variants (mutations) in genes linked to cellular energy (mitochondria). This study aims to identify some of the gene variants that may contribute to ME/CFS and FM by comparing the genetic sequences of these patients with that of the controls. Our hypothesis is that ME/CFS is both an autoimmune disorder and an energy deficiency disorder, so we have focused on both immune and mitochondrial/energy genes. Blood samples were drawn from ME/CFS patients and controls and the leukocytes were separated from it. The mRNA of these leukocytes, which represents the transcriptome of the cells, was extracted and then sequenced using the RNA-Seq method. The mutations in the genes were ranked as low, moderate or high impact on the function of the gene. In this initial sample of 44 of the planned 340 participants, we identified 3 automunity/immune function variants (LILRA6, RFTN1 and MARCH8) and several different multigene pattern for mitochondrial variants with moderate and high impact that occurred with greater frequency in the ME/CFS patients vs. controls. When the data from all 340 participants is complete, it will help identify biomarkers to make diagnosis easier and may also provide targets for developing new treatments for ME/CFS and FM.

References:

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