

MRI Spectroscopy: A Case of Mitochondrial Disorder in a 13 Year-old Boy.

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A 13-year-old boy with movement disorder was evaluated at Massachusetts General Hospital for mitochondrial disease.

The patient underwent conventional Magnetic Resonance Imaging (MRI) of the brain which showed normal white matter, but failed to find a lesion to explain the patient's clinical symptoms (Figure 1). However, Magnetic Resonance Spectroscopy (MRS) on a region of white matter in the left frontal lobe, revealed an abnormality in the spectrum (Figure 2). The ratios of choline to creatine peak amplitude and N-acetyl-aspartate to creatine peak amplitude were found to be increased. Together these increases are consistent with diminished creatine levels. Reduced creatine levels are indicative of a metabolic abnormality in this region of white matter and in particular, of a mitochondrial metabolic disorder.

The clinical picture, along with MRS findings, allowed a diagnosis of mitochondrial disease to be made.

This case illustrates the power of MRS as a non-invasive means of obtaining neurological and metabolic information in the presence of a normal MRI. It may be possible to use MRS to diagnose disease states which currently are missed with conventional MRI.

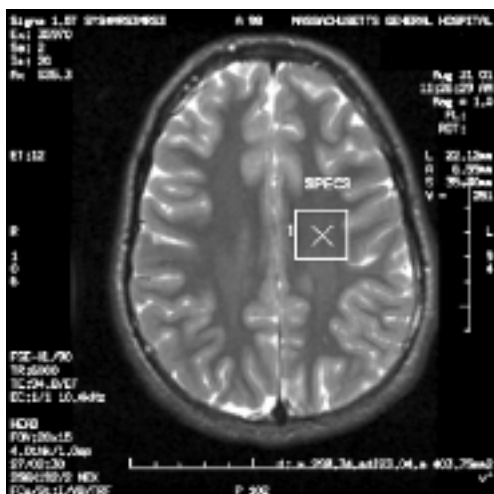


Figure 1. Brain MRI of patient showing no focal areas of abnormality in white matter. X marks area used for MRS.

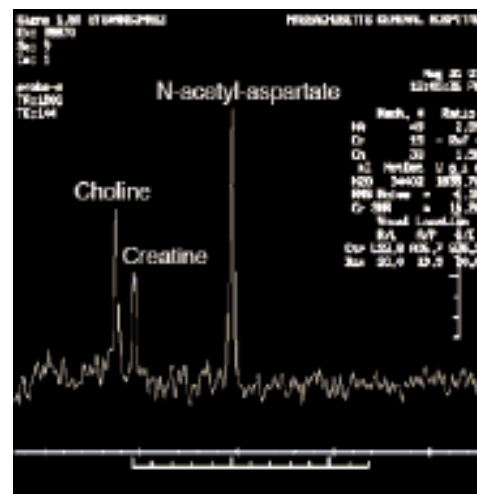


Figure 2. Magnetic Resonance Spectroscopy taken from region of the brain marked X. Three resonance peaks are shown at 2.0, 3.0 and 3.2 ppm, corresponding to N-acetyl-aspartate, creatine and choline respectively. The amplitude of the resonance peak reflects the concentration of the molecule in the region sampled.