Proton MR Spectroscopy in The Diagnostic Evaluation of Suspected Mitochondrial Disease in Iran

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Mitochondrial diseases are a group of inherited disorders Caused by a derangement of mitochondrial respiration. The clinical manifestations are heterogeneous, and the diagnosis is often based on information acquired from multiple levels of inquiry that require a variety of diagnostic tests for proper determination.

Neuroimaging may play a significant role in diagnosis. The various modalities of nuclear magnetic resonance imaging (MRI) allow for multiple independent detection procedures that can give important anatomical and metabolic clues for diagnosis when the constellation of symptoms is suggestive of mitochondrial disease, neuroimaging features may be diagnostic and suggestive, can help direct further workup, and can help to further characterize the underlying brain abnormalities. The non-invasive nature of neuroimaging also allows for longitudinal studies. To date, no pathognomonic correlation between specific genetic defect and neuroimaging findings have been described. The definitive diagnosis of a mitochondrial disorder can be difficult to establish. Magnetic resonance imaging changes may be nonspecific; however, certain neuroimaging results can give important clues that a patient may have a mitochondrial disease. Conventional MRI may show Brain atrophy, supratentorial white matter lesions, basal ganglia involvement deep gray structural abnormalities, and delayed myelination are the most frequent anomalies in the definite cases.

Also they may represent clearly characteristic of a given disorder such as stroke-like lesions that do not respect vascular boundaries in mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episode (MELAS). White matter hyper intensities with or without associated gray matter involvement may also be observed or stroke-like lesions that do not respect vascular territories.

Chemical techniques such as proton magnetic resonance spectroscopy (MRS) may demonstrate high levels of lactate or succinate. When found, these results are suggestive of a mitochondrial disease. MRI and MRS studies may also show non-specific findings such as delayed myelination or non-specific leukodystrophies picture. However, in the context of other biochemical, structural, and clinical findings, even non-specific findings may support further diagnostic testing for potential mitochondrial disease. Once a diagnosis has been established, these non-invasive tools can also aid in following disease progression and evaluate the effects of therapeutic interventions.

Keywords: Pediatrics; Mitochondrial disease; Neuroimaging; Proton MR Spectroscopy