THE ROLE OF MAGNETIC RESONANCE SPECTROSCOPY IN EVALUATION OF PATIENTS WITH NEUROFIBROMATOSIS TYPE 1

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INTRODUCTION

Neurofibromatosis type 1 (NF1) is a common autosomal dominant disorderυ which predisposes patients to develop benign and malignant neoplasms, mostly due to a loss of function mutation in the neurofibromatosis type 1 (NF1) gene. NF1's protein product, neurofibromin, controls Ras pathway inactivation and so functions as a tumor suppressor. A decrease in neurofibromin, which is found in neurons, Schwann cells, and melanocytes, predisposes patients with NF1 to tumors in both the central and peripheral nervous systems. The most common focal brain lesions found in NF1 patients are hamartomas (FASI) and gliomas. Proton Magnetic resonance spectroscopy is one such technique which provides a noninvasive method for characterizing the cellular biochemistry which underlies brain pathologies, helping in differentiating between hamartomas (FASI) and gliomas in our study based on the spectroscopic analysis of different metabolites

AIM OF THE WORK

This study aimed to assess the role of MR spectroscopy in differentiating between hamartomas (FASI) & gliomas in NF1 patients. And assessing the metabolite differences in the normal appearing brain of NF1 patients and normal healthy volunteers.

PATIENTS AND METHODS

PATIENTS: The study included 15 patients diagnosed with NF1 with glioma lesions, hamartoma (FASI) lesions and without focal brain lesions at conventional MRI, and 15 healthy volunteers as a control group.

METHODS: **I.** Full history taking **II.** Thorough clinical examination

III. Imaging Conventional MRI: Brain MRI was performed on a 3 T (Signa HDxt, General Electric, Milwaukee, USA) closed configuration whole body scanner using a standard quadrature head coil in the 25 patients.

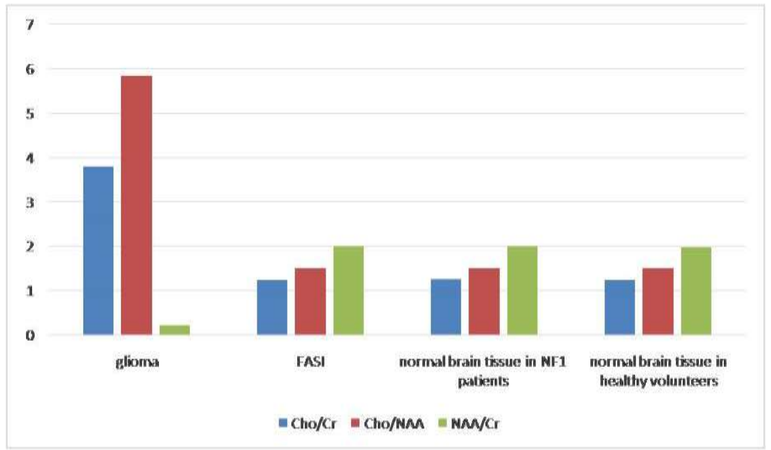
All patients and control group were subjected to the following MRI protocols:

1.Axial, coronal T2- weighted turbo spin echo (T2 TSE).
2. Axial FLAIR. 3. Pre-contrast axial and sagital T1 weighted spin echo (T1 SE).
4. Post contrast conventional axial, sagital and coronal T1 spin echo. 5. MR Spectroscopy.

IV. Spectroscopic analysis

RESULTS

Glioma lesions showed increased NAA/Cr and Cho/Cr ratios compared to those of hamartomas (FASI) lesions. And showed markedly lowered NAA/Cho ratios.
While hamartomas showed MRS results of NAA/Cr and Cho/Cr ratios lower than that of gliomas and NAA/Co ratios more or less similar to those of healthy volunteers.
There was no significant difference in the spectroscopic analysis of the normal appearing white matter in NF1 patients with focal lesions and NF1 patients without focal lesions and the normal white matter of the control group of healthy volunteers.



As shown in chart diagram there is no significant differences between hamartomas (FASI) and normal appearing brain in NF1 patients with and without focal lesion nor the normal brain tissue of healthy volunteers. However, gliomas show marked decrease in NAA/Cr ratio denotingυ demyelination and axonal loss and significant increase in Cho/Cr ratio denoting membrane turnover.

a)sagital T2WI showing hyper-intense lentiform nucleus and focal hyperintensities at left thalamic region

b&c) ***Multi-voxel MR spectroscopy analysis (from multiple samples obtained from the left lentiform nucleus lesion):***

-Persistent high Ch peak.
-Persistent high Ch/Cr ratio, reaching up to 2.8
-Persistent high Ch/NAA ratio reaching up to 9.8
-Low NAA peak.
-NAA/Ch Ratio is down to 0.1

d&e) ***Multi-voxel MR spectroscopy analysis of the left thalamic region shows:***

-Preserved choline, creatine and NAA
-No definite other abnormal metabolites.

CONCLUSION

-MR spectroscopy imaging technique can give important in vivo physiologic and metabolic information.
-MRI is a valuable tool in diagnosis of patients with NF1.
-MR spectroscopy is a valuable non-invasive modality in diagnosis of focal brain lesions in NF1 patients.
-MR spectroscopy is able to differentiate between hamartomas (FASI) and gliomas in patients with NF1.
-No significant difference in spectroscopic analysis of normal appearing brain in patients diagnosed with NF1 and healthy volunteers.