**INTRODUCTION & AIM**

Gaucher disease is the most prevalent inherited, lysosomal storage disease. It results from mutations that confer a deficient level of activity of β-glucocerebrosidase, a membrane-bound lysosomal enzyme. This deficiency leads to accumulation of the lipid glucocerebroside in the lysosomes of monocytes and macrophages, called Gaucher cells. The symptoms and pathology of Gaucher disease result from the accumulation of Gaucher cells in various organ system. The disease has traditionally been classified into three clinical phenotypes: type I-adult, non-neuronopathic; type II-infantile or acute neuronopathic form (rapidly progressive neurovisceral storage disease, with death during infancy); and type III-juvenile or chronic neuronopathic (less rapidly progressive neurovisceral storage disease). Type I is the most prevalent form of Gaucher disease. The clinical features of type I disease encompass hepatosplenomegaly, cytopenia, and bone involvement. Bone disease is one of the most debilitating features of type I Gaucher disease and consists of atypical bone pain, osteonecrosis, pathologic fractures, and bone crises. Osteonecrosis is probably the most clinically significant and disabling skeletal manifestation in Gaucher disease; it is bone death, believed to be secondary to ischaemia due to chronic infarction, and once the necrotic process starts, it cannot be reversed. It affects predominantly the femoral head, proximal humerus, and vertebral bodies, and can result in fracture and joint collapse. Magnetic Resonance (MR) is the modality of choice to depict bone marrow abnormalities. It is extremely sensitive to the skeletal pathologies found in Gaucher disease including acute bone infarction, infection, trauma, marrow infiltration with Gaucher cells and avascular necrosis. The infiltration of Gaucher cells, which replace the fat marrow, dramatically changes the signal to hypointense on T1-, T2- and T2*-weighted images. In some patients with Gaucher disease, the T1-weighted signal is hypointense and the T2-weighted images with or without fat suppression or STIR sequences show hyperintense inclusions. This may indicate an active or “complicated” bone marrow process, such as acute bone crisis, occult fracture, infection or bone infarction. To evaluate the biochemical process underlying the infiltration of Gaucher cells, single voxel MR spectroscopy (1H-MRS) on vertebral bone marrow has been acquired in normal and pathological conditions at different age.

**MATERIAL AND METHODS**

12 patients were affected by Gaucher disease (8 male and 4 female; aged between 13 to 47 years) and 20 subjects (11 male and 9 female; aged between 17 to 75 years), with chronic lumbar pain but without bone abnormalities, were investigated on a 1.5-T scanner (GE Signa Excite HDx 1.5T). The MRI protocol included SE T1-weighted, SE T2-weighted, and STIR sequences on the sagittal and axial plane for morphological evaluation, followed by a 1H-MRS acquisition consisting of a single voxel PRESS sequence VOI covering the spongiosa of L3 with the following parameters: TR=2000ms, TE=35 ms, 128 signals, bandwidth 2500 Hz, 2048 data points. 1H-MRS spectra were analyzed offline using dedicated software (SaGe) and the data-processing included zero filling, spectral apodization using a Gaussian function with 2.5 Hz linewidth. Signal amplitudes of the water signal and of the main lipid peak at 1.3 ppm were measured. The fat to water signal ratio (FWR) and the Fat Fraction (FF) (according to 100 x FWR x FWR/(FWR+1)) have been calculated.

**RESULTS**

Good quality spectra were acquired in all the subjects taken as control sample, and in 10 out of 12 patients (2 spectra have been discharged for motion artifact). An age-related increase of fat contents was detected in the normal sample: the fat fraction, as well the FWR, increase with age, ranging from from 31 % at 17 years to 67 % at 63 years of age. In comparison with spectra of normal individuals of any age range, spectra acquired on patients with Gaucher disease showed a significant reduction of fat content (below 29% down to non-detectability of the resonance signal at 1.3 ppm). Furthermore no age-related increase of fat contents has been found in Gaucher patients.

![Figure 1: VOI location and 1H-MRS spectrum (left); plot of FF values in function of age (right)](image)

**CONCLUSION**

Vertebral bone marrow 1H-MRS is feasible at 1.5 T and yields additional information about bone marrow structure and aging. The aging vertebral fat increase detected in vivo by means IH MRS is in according to quantitative histologic studies on age-related changes in bone, showing that the change from hematopoietic to fatty marrow is gradual, steady, and progressive. The accurate determination of fat fractions can be useful also to detect biochemical abnormalities in pathological conditions. The fat fraction depletion found in Gaucher disease could reflect the accumulation of Gaucher cells in the bone marrow. The absence of the normal developmental and aging change in Gaucher patients could reflect the more severity of bone involvement during childhood. Thanks to its quantitative properties, MR spectroscopy can yield additional information about the bone marrow structure and its biochemical degeneration and can be used as an adjunct to conventional MRI of the spine.