Does MR elastography determined liver stiffness correlate with disease severity score in type 1 Gaucher disease patients?

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Background
Gaucher disease (GD) is an autosomal recessive disorder causedby a mutation in the enzyme glucocerebrosidase resulting in a failure to metabolize glycolipids. Patients with GD1 often present with hepatosplenomegaly. Because liver biopsies are not routinely performed, it is not known how many patients are at risk for developing liver fibrosis, however cases of advanced fibrosis and cirrhosis have been reported. The exact mechanism of liver fibrosis in this disease has not yet been elucidated,and it is not well understood how liver disease may influence GD1 disease severity.

Aims/Hypothesis
The aim of this study was to determine the relationship between liver stiffness, measured by magnetic resonance elastography (MRE) and global GD1 disease severity, measured by a validated disease severity scoring system (DS3). We hypothesized that higher liver stiffness would directly correlate with increasing disease severity scores.

Methods
Data were gathered retrospectively on 15 patients with GD1 (9 men, 6 women) and used to calculate avalidated GD1 disease severity scoring score(DS3). Liver stiffness values (in kPa) were calculated from routine clinical MRE scans . Liver stiffness values were plotted against DS3 scores and a linear regression model was generated and plotted with 95% confidence bands. The model was then used to predict liver stiffness values using the patients' DS3 scores. The predicted stiffness values were then plotted against the actual stiffness values, and a Pearson's r value was calculated.

Results
The linear regression model showed a significant correlation between disease severity and Gaucher disease (Pearson's r=0.64, p=0.014). Correlation between predicted and actual stiffness values was also determined to be significant (Pearson's r = 0.64, p=0.014).

Conclusion
Our model suggests that increasingliver stiffness is associated with more severe GD1. Future prospective studies are required to validate these findings in a larger cohort of patients and to determine if treatment of GD1 with enzyme replacement reduces the liver stiffness and likelihood of progression to liver fibrosis.

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