Advanced liver imaging in Gaucher Disease: a pilot study

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To determine whether different MR-based imaging techniques and FibroScan can be used for the purpose of identifying and monitoring Gaucher Disease patients at high risk of developing disease related liver complications.

Ethical review	Approved WMO
Status	Pending
Health condition type	Endocrine disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON34763

Source
ToetsingOnline

Brief title
Liver imaging in Gaucher Disease

Condition

- Endocrine disorders congenital
- Hepatic and hepatobiliary disorders

Synonym
Gaucher Disease, lysosomal storage disorder

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Fibroscan, Gaucher Disease, Liver, Magnetic Resonance Imaging

Outcome measures

Primary outcome

The differences between Gaucher Disease patients with high risk versus normal risk and Gaucher Disease patients versus healthy volunteers in:

-Elasticity measurements of the liver obtained with MR Elastography and FibroScan

-Iron accumulation in the liver assessed by MRI and biochemical values.

Secondary outcome

The differences between Gaucher Disease patients with high risk versus normal risk and Gaucher Disease patients versus healthy volunteers in: The spectrum of glucosylceramide in the liver obtained with 1H-MR Spectroscopy.

Study description

Background summary

Gaucher disease is a hereditary lysosomal storage disorder. The deficiency in glucocerebrosidase leads to accumulation of glucocerebroside or "gaucheromas" within the body. Gaucher disease patients typically show high serum ferritin levels as well. One of the long term complications of the disease within the liver are increased risk of fibrosis, cirrhosis and hepatocellular carcinoma (HCC). The pathophysiology behind this phenomenon is still largely unknown. We hypothesize that iron accumulation and/ or glucocerebroside storage cause fibrotic and cirrhotic changes to occur and that this mechanism is responsible for the increased risk of HCC as seen in Gaucher disease. However, this has never been investigated due to a lack of safe and reliable diagnostic tools. Newly available imaging techniques could be of value in the early detection and subsequent monitoring of these complications. MR Elastography as well as FibroScan can detect liver fibrosis by measuring liver elasticity. Also, an algorithm was designed to asses liver iron content from MRI measurements.

Proton-MR Spectroscopy (1H-MRS) is another MRI based method which can quantify different metabolites within tissues. 1H -MRS is most commonly used for the quantification of fat within the liver. It is unknown whether 1H -MRS can measure the amount of glucocerebroside within liver tissue. It would therefore be valuable to test our hypotheses by investigating the value of these non-invasive imaging techniques in patients with Gaucher disease.

Study objective

To determine whether different MR-based imaging techniques and FibroScan can be used for the purpose of identifying and monitoring Gaucher Disease patients at high risk of developing disease related liver complications.

Study design

This is a single-centre observational pilot study. Both patients and healthy volunteers will be included. The study is not randomized and patients will be non-consecutively included. Patients with from the Endocrinology outpatient clinic who meet the inclusion criteria will be informed about the study. After giving informed consent they will be included in the study.
Healthy volunteers will be recruted by means of sending out flyers thoughout the AMC. Healthy volunteers will be age and sex matched with the mild Gaucher patient group. After giving informed consent they will be included in the study.

Study burden and risks

There are no significant risks associated with the interventions. The procedures will require:
-10-20 minutes in the 1.5 Tesla MRI scanner
-30-40 minutes in the 3 Tesla MRI scanner
-15 minutes for FibroScan at the Gastroenterology Department
-Drawing of 2-3 extra tubes of blood voor Gaucher Disease patients added to regular blood drawing.

Due to logistics, patients might have some waiting time in between the different procedures. We will try to plan all three interventions on the same day, and keep waiting time as short as possible.

Participants will fill out a checklist to identify any possible contra-indications for MRI scanning. Participants with contra-indications for MRI will excluded from this study. During MRI scanning, the patient will have to lie still on his or her back in a MRI scanner. Both MRI and FibroScan are non-invasive, non-ionizing examinations. No contrast medium will be administered. Patients with Gaucher Disease routinely visit the outpatient clinic in the AMC four times a year. Whenever possible, we will try to combine

the two MRIs and FibroScan with visiting the outpatient clinic. However, due to scanning capacity this might not always be achievable. Therefore, participating in this pilot study might require an extra visit to the AMC. The best available option will be considered for each participant separately.

Participating in this study has no direct advantage for the patient, except extra insight in their disease. Patients are not delayed in treatment for their disease. Apart from the possible extra visit to the hospital and lying in the MRI scanner, there will be little extra physical and psychological discomfort associated with participation.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

Patients with Gaucher Disease;
Over 18 years of age;
BMI under 27 kg/m2;
At least 2 years of therapy;
Written informed consent

Exclusion criteria

Known history of chronic liver disease other than caused by Gaucher Disease;
Alcohol more than 3 units per day for males and more than 2 units per day for females;
Contra-indications for MRI scanning (standard checklist);
younger than 18 years of age;
use of medications that are known to affect the liver (see protocol)

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-03-2010
Enrollment:	21
Type:	Anticipated

Ethics review

Approved WMO
Application type: First submission

Review commission: METC Amsterdam UMC

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL30863.018.09