The effect of one mutation in the ATP8B1 gene on hearing.

Published: 05-05-2009
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The aim of this study is to investigate the effect of ATP8B1 carrier on hearing in human.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Hepatobiliary disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON33871

Source
ToetsingOnline

Brief title
ATP8B1 and hearing

Condition

- Hepatobiliary disorders congenital
- Hepatic and hepatobiliary disorders

Synonym
bile accumulation, liver disease

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Utrecht
Source(s) of monetary or material Support: Ministerie van OC&W,BRIC patiënten vereniging

Intervention

Keyword: ATP8B1, Familiar intrahepatic cholestasis, Hearing

Outcome measures

Primary outcome

Hearing loss

Secondary outcome

Not applicable

Study description

Background summary

Familiar intrahepatic cholestasis type 1 (FIC1) is caused by mutations in the ATP8B1 gene and is characterized by intrahepatic cholestasis, either in episodes (BRIC1) or progressive (PFIC1). The exact mechanism of a deficiency of FIC1 leading to cholestasis is still unknown.
Because ATP8B1 expression is also found in other organs than the liver, patients with FIC1 may also present with extrahepatic symptoms. Some are well known, such as pancreatitis, failure to thrive and diarrhea. Recently we also found, that patients with BRIC1 do have hearing loss. Despite a mild phenotype in patients with BRIC1 the degree of hearing loss seems to be similar to the hearing loss in patients with PFIC1. Furthermore, also family members with one mutation in ATP8B1 complain about hearing problems.

Study objective

The aim of this study is to investigate the effect of ATP8B1 carrier on hearing in human.

Study design

About ten patients with BRIC1 will be asked permission to contact family members with the question to whether they want to participate in this study, consisting of four audiometric tests: tone and speach audiometry, tympanometry and measuring of OtoAcoustic Emissions. These tests are without risk or side effects.
If it's unknown whether they are carrier or not, we will first ask permission

to take blood for DNA analysis.

Study burden and risks

No risks

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

One mutation in ATP8B1 gene
Adults (18 years or older)
No congenital hearing disorder or hearing loss with a known cause

Exclusion criteria

Congenital hearing disorder or hearing loss with a known cause
Children

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	10-06-2009
Enrollment:	10
Type:	Actual

Ethics review

Approved WMO

Date:	05-05-2009
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Utrecht (Utrecht)

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	NL25723.041.08