Hereditary hearing impairment in the Netherlands: elucidation of genetic causes, and clinical characterization.

Published: 31-05-2011
Last updated: 04-05-2024

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Ethical review	Approved WMO
Status	Recruiting
Health condition type	Ear and labyrinthine disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON47077

Source
ToetsingOnline

Brief title
Hereditary hearing impairment in the Netherlands

Condition

- Ear and labyrinthine disorders congenital

Synonym
hereditary deafness, hereditary hearing impairment

Research involving
Human

Sponsors and support

Primary sponsor: KNO
Source(s) of monetary or material Support: NWO (ZONMW),Het Heinsius Houbolt fonds;Fonds NutsOHRA

Intervention

Keyword: genotype, hearing impairment, inheritance, phenotype

Outcome measures

Primary outcome

1, pathologic genetic variants that are known to or are likely to be causative

for hearing impairment. 2, genotype-phenotype correlations for at least part of

the genetic subtypes leading to improved counselling.

Secondary outcome

A strategy for DNA-diagnostics for hearing impairment in the Dutch population.

Study description

Background summary

Currently, many of the causative genes for syndromic hearing impairment are known. However, especially for non-syndromic hereditary hearing impairment (NSHI) the majority of the genes remain to be identified which means that in ~70% of NSHI patients, the genetic defect remains unknown in routine DNA-diagnostics. Knowledge of the genetic cause is important to patients in order to distinguish between genetic and non-genetic hearing impairment, to give them a more grounded genetic counselling and prognosis, to distinguish in an early stage between syndromic and non-syndromic hearing impairment, and to determine whether treatments such as cochlear implantation may be helpful to the patient.

Study objective

The goal of this study is to collect a large cohort of patients mainly with NSHI. In the study cohort, the genetic causes underlying the NSHI will be determined which can be either in known or novel deafness genes. Also, the clinical characteristics associated with the specific genetic defects will be determined. Collecting a large cohort of patients will help us to provide 1. accurate genetic counselling to patients, 2. a more reliable prognosis, 3. to discover new genes associated with hearing impairment, 4. to develop a strategy for future routine DNA diagnostics for hereditary hearing impairment and 5.

insight in inner ear (dys)function.

Study design

Patients will be informed about the study by their ENT specialist, clinical geneticist or audiologist. The usual diagnostic examinations for hearing impairment will be performed (otoscopy, tympanometry, pure tone audiometry, speech audiometry), and a blood sample will be drawn for usual DNA diagnostic screening for hereditary hearing impairment.
Patient will receive a questionnaire, with questions about their hearing, balance and origin of their grandparents. Besides, the patient will be asked to give consent on the further genetic research when no mutations in known deafnessgenes can be found.
A small number of patients will be invited for more psychophysical hearing tests, in order to get a more detailed phenotype.

Study burden and risks

The only burden for a patient could be the possible extra examinations (psychophysical testing, takes 1 hour). The other examinations would be performed anyway for diagnostic purposes.

Contacts

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Selecteer

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

patients with hearing impairment in whom common non-genetic causes for hearing impairment are excluded.

Exclusion criteria

patients with age-related hearing impairment

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	15-06-2011
Enrollment:	2050
Type:	Actual

Ethics review

Approved WMO Date:	31-05-2011
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	14-01-2015
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	29-07-2015
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	30-12-2015
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	27-07-2017
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	28-12-2017
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	02-05-2018
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

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	NL33648.091.10