Rate of progression in USH2A related retinal degeneration

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To report the natural history of retinal degeneration in patients with biallelic mutations in the USH2A gene; to identify sensitive structural and functional outcome measures to use for future multicenter clinical trials in USH2A-related retinal...

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Inner ear and VIIIth cranial nerve disorders
Study type	Observational non invasive

Summary

ID

NL-OMON46590

Source
ToetsingOnline

Brief title
RUSH2A

Condition

- Inner ear and VIIIth cranial nerve disorders
- Retina, choroid and vitreous haemorrhages and vascular disorders

Synonym
Retinal dystrophy, retinitis pigmentosa

Research involving
Human

Sponsors and support

Primary sponsor: Jaeb Center For Health Research
Source(s) of monetary or material Support: Foundation Fighting Blindness USA

Intervention

Keyword: Natural history, Retinitis Pigmentosa, USH2A, Usher syndrome

Outcome measures

Primary outcome

Visual field sensitivity measured by static perimetry, best corrected visual acuity, mean retinal sensitivity as measured by fundus-guided microperimetry, ellipsoid zone area as measured by spectral-domain optical coherence tomography, retinal function using full-field electroretinography amplitudes and timing in response to rod- and cone-specific stimuli

Secondary outcome

Auditory function, patient reported outcomes

Study description

Background summary

Usher syndrome represents a leading cause of autosomal recessive deaf-blindness. Usher syndrome is divided into 3 types, based on the severity and onset of hearing loss. The most common gene mutated in patients with Usher syndrome type 2 is USH2A. USH2A mutations may also cause retinitis pigmentosa (RP) without congenital hearing loss. As new treatments for USH2A-related retinal degeneration are developed, a clear understanding of the natural history of disease progression of USH2A-related retinal degeneration is necessary. Limited natural history data are available from patients with Usher syndrome type 2. Natural history studies of patients with USH2A mutations are needed to accelerate the development of outcome measures for clinical trials. Sensitive, objective outcome measures of retinal degeneration will greatly facilitate development of treatments for Usher syndrome patients. Together these approaches are expected to have an impact on understanding USH2A-related retinal degeneration, developing experimental treatment protocols, and assessing their effectiveness.

Study objective

To report the natural history of retinal degeneration in patients with biallelic mutations in the USH2A gene; to identify sensitive structural and functional outcome measures to use for future multicenter clinical trials in USH2A-related retinal degeneration; to identify well-defined subpopulations for future clinical trials of investigative treatments for USH2A-related retinal degeneration

Study design

This study is designed as a multicenter, longitudinal, prospective natural history study. A second cohort of eyes with more severe vision impairment will enroll into a cross-sectional baseline study only.

Study burden and risks

We anticipate that study enrollment will be representative of the population of patients with biallelic mutations in the USH2A gene, including those under the age of 18. Biallelic mutations in the USH2A gene can impact individuals in the first or second decade of life, therefore it is imperative that children are included in this natural history study in order to adequately characterize the natural history of retinal degeneration in patients with USH2A mutations.

Participants do not benefit, risks are considered negligible, procedures are non-invasive and take 3 to 6 hours extra time from patient (and parent) per visit, one visit per year. It is anticipated that, in the future, patients with USH2A-related retinal degeneration will benefit from newly developed therapeutic strategies.

Contacts

Public
Jaeb Center For Health Research

Amberly Drive, suite 350 15310
Tampa FL33647
US
Scientific
Jaeb Center For Health Research

Amberly Drive, suite 350 15310
Tampa FL33647
US

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

2.2.1 Study Participant Inclusion Criteria
1. Willing and able to complete the informed consent process
2. Ability to return for all study visits over 48 months if in the natural history study
3. Age * 8 years
4. At least 2 pathogenic or likely pathogenic mutations in USH2A gene from a clinically certified lab report

Exclusion criteria

2.2.2 Study Participant Exclusion Criteria
1. Mutations in genes that cause autosomal dominant RP, X-linked RP, or presence of biallelic mutations in autosomal recessive RP/retinal dystrophy genes other than USH2A
2. Expected to enter experimental treatment trial at any time during this study
3. History of more than 1 year of cumulative treatment, at any time, with an agent associated with pigmentary retinopathy (including hydroxychloroquine, chloroquine, thioridazine, and deferoxamine)

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-08-2018
Enrollment:	25
Type:	Actual

Medical products/devices used

Generic name:	Octopus 900 Pro
Registration:	Yes - CE intended use

Ethics review

Approved WMO Date:	26-04-2018
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	14-04-2021
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	03-05-2021
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	NL62954.091.17