Genetics of childhood glaucoma in the Netherlands

Published: 11-09-2024
Last updated: 18-01-2025

The goal is to study the genetic background of childhood glaucoma in the Netherlands. We aim to study which mutations play a role in the pathogenesis of childhood glaucoma in the Netherlands and their prevalence. We will study the genetic mutations...

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

Summary

ID

NL-OMON56998

Source
ToetsingOnline

Brief title
Genetics of childhood glaucoma

Condition

- Eye disorders congenital
- Glaucoma and ocular hypertension

Synonym
childhood glaucoma, glaucoma in children

Research involving
Human

Sponsors and support

Primary sponsor: Amsterdam UMC
Source(s) of monetary or material Support: Stichting Louise Rottinghuisfonds

Intervention

Keyword: childhood glaucoma, genetics

Outcome measures

Primary outcome

Primary outcomes are a mutation in one of the 26 known glaucoma genes of the gene panel, a new gene mutations found with whole exome sequencing (WES), or no gene mutation.

Secondary outcome

Secondary study parameters are the genotype-phenotype study where we link the clinical findings to the gene mutation

Study description

Background summary

Childhood glaucoma is a rare but sight threatening disease where an increased intraocular pressure causes harmful changes to the eye. The genetic background plays a role in the disease risk and severity for childhood glaucoma. In primary congenital glaucoma (PCG), mutations in CYP1B1 gene are frequently found (15-90% depending on the population). Patients with a CYP1B1 mutation present often with a more severe phenotype of the disease, which is less easily to treat. Quite a lot is known about the CYP1B1 gene in relation to childhood glaucoma. However, other genes are also involved, and mutations in these other genes might play a more prominent role in Caucasian and Azian children. Not much is known about these other gene mutations, their prevalence and phenotype effect. Furthermore, not all cases with childhood glaucoma can be explained with the known (childhood) glaucoma genes. Knowledge and widening of the scope of disease genes will help to better understand the disease and improve treatment strategies.

Study objective

The goal is to study the genetic background of childhood glaucoma in the Netherlands. We aim to study which mutations play a role in the pathogenesis of childhood glaucoma in the Netherlands and their prevalence. We will study the

genetic mutations in relation to the clinical findings. With this phenotype-genotype study we will ultimately come to personalized medicine, which make each treatment path per patient optimal and significant. Finally we aim to find new gene mutations that are involved in childhood glaucoma.

Study design

diagnostic cohort study

Study burden and risks

The study's risk is minimal, and the burden is low, as most participants already have stored DNA. If not, a small blood sample is taken, preferably during planned surgeries for children.
Participation to the study might result in the finding of a gene mutation that causes the disease. This finding could give more context of the disease and more information about the heritability, which might be beneficial for patients/parents.
Global research on childhood glaucoma may not fully apply to the Netherlands due to differing surgical techniques and genetic factors. Deepening our understanding of the genetic background in the Netherlands is important to develop personalized medicine. It is important to include children into this study, since it is a childhood disease and children need to be treated (usually surgically) at young age. The childhood glaucoma population in the Amsterdam UMC is relatively young and it would take around 8 years before half of these patients are > 16 years old.

Contacts

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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)
Babies and toddlers (28 days-23 months)
Newborns

Inclusion criteria

Patients with childhood glaucoma
- without a glaucoma gene panel screening; or
- with a negative glaucoma gene panel

Exclusion criteria

patients with childhood glaucoma where a gene mutation have been found

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	02-09-2024
Enrollment:	437
Type:	Anticipated

Ethics review

Approved WMO

Date:	11-09-2024
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	NL81534.029.22