Mutations in X-linked cosmc gene and their role in IgA nephropathy

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Ethical review Approved WMO

Status Pending

Health condition type Chromosomal abnormalities, gene alterations and gene variants

Study type Observational non invasive

Summary

ID

NL-OMON29999

Source

ToetsingOnline

Brief title

The course of IgA nephropathy

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Nephropathies

Synonym

IgA-glycosylation and kidney inflammation

Research involving

Human

Sponsors and support

Primary sponsor: Nierstichting Nederland

Source(s) of monetary or material Support: Nierstichting Nederland

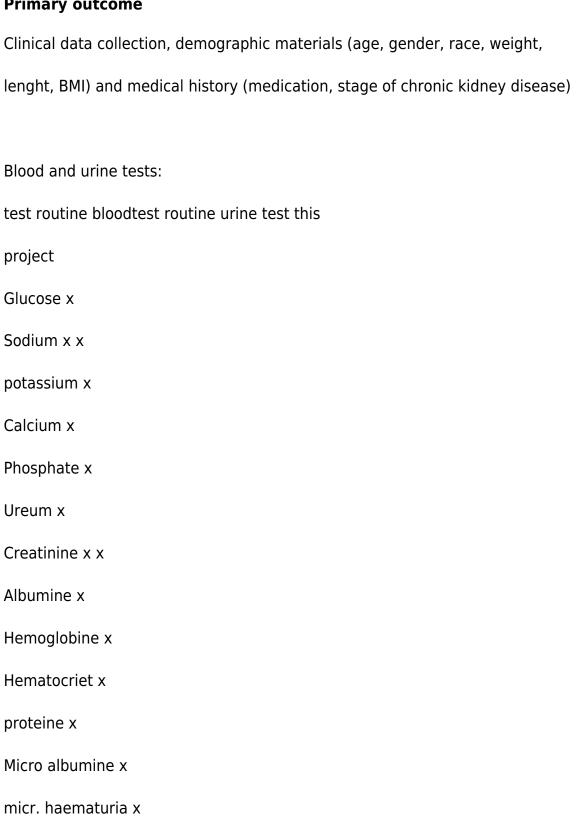
Intervention

Keyword: Chronic Kidney Disease, cosmc gene, Glycosylation, IgA nephropathy

Outcome measures

Immunoglobulines/Totaal IgA x

Primary outcome



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bloodgroup x

nucleotide sequence

and expression analyse from

cosmc gene in white

bloodcells and mucus x

DNA sequence or expression

of the cosmc

gene

Х

RT-PCR from cosmc mRNA x

Secondary outcome

not applicable.

Study description

Background summary

IgA nephropathy (IgAN) is the most common form of glomerulonephritis worldwide. The disease is characterized by the precipitation of IgA1, a defense protein that is synthesized by leucocytes, in the glomeruli. The damage caused by the precipitated IgA1 may develop into chronic renal failure.

The molecular basis of the development of IgAN is not known. It has been shown that serum from patients with IgA nephropathy contains aberrant IgA1 molecules. Our hypothesis is that the formation of the aberrant IgA1 is due to a defect in the biosynthesis of this immunoglobulin, and that this defect causes the IgA1 to precipitate in the glomeruli.

Study objective

The major aim of this project is to elucidate the molecular basis of IgAN. A large cohort of IgAN patients will be screened on aberrancies in the DNA sequence or expression of the gene encoding Cosmc, a protein that is essential for the biosynthesis of normal IgA1. Aberrancies that are found will be related

to the observed clinical parameters of the respective patients. Increased insight and understanding of the molecular basis of IgAN is important to enable the future development of improved diagnostics and therapy for this disease.

Study design

Patients will be screened on aberrancies in the DNA sequence or expression of the gene encoding Cosmc.

Patients in different stages of IgAN, patients with proteinuria and healthy persons will be tested,

In this project 50 patients with IgAN, 50 patients with proteinuria and 50 healthy controls will be tested, matched for age, gender and race. The samples will be gathered in the coming 1,5 year.

Study burden and risks

The burden and risk are minimized because bloodsample will be taken only when routine bloodcontrole will take place.

Mucus samples are painless and will take minimal time.

Contacts

Public

Nierstichting Nederland

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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

Age 0-75 years

Exclusion criteria

Diabetes Mellitus, malignancy

Study design

Design

Study type: Observational non invasive

Intervention model: Other

Allocation: Non-randomized controlled trial

Masking: Open (masking not used)

Control: Active

Primary purpose: Basic science

Recruitment

NL

Recruitment status: Pending

Start date (anticipated): 01-06-2006

Enrollment: 150

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 NL12328.029.06