High resolution genotyping with immunochip in idiopathic retroperitoneal fibrosis patients

Published: 08-12-2015
Last updated: 19-04-2024

The study is designed to evaluate the possible association between genetic variants and susceptibility to IRF, based on previous studies in autoimmune-inflammatory diseases (celiac disease, type 1 diabetes and rheumatoid arthritis).

Ethical review	Approved WMO
Status	Pending
Health condition type	Other condition
Study type	Observational non invasive

Summary

ID

NL-OMON42725

Source
ToetsingOnline

Brief title
Genotyping in idiopathic retroperitoneal fibrosis patients

Condition

- Other condition

Synonym
connective tissue disease; chronic fibrosing disorder

Health condition

idiopathische retroperitoneale fibrose

Research involving
Human

Sponsors and support

Primary sponsor: Albert Schweitzer Ziekenhuis
Source(s) of monetary or material Support: Kosten worden betaald vanuit de Universiteit Parma;Italie

Intervention

Keyword: Genotyping, Retroperitoneal fibrosis

Outcome measures

Primary outcome

Identification of genetic markers associated with IRF, providing new insights

into the etiology of this complex disease.

Secondary outcome

1. Creation of a database on the basis of a possible new classification of the

various forms of the IRF on the basis of genetic data;

2. Creation of a public database for the dissemination of the results.

Study description

Background summary

IRF is a rare disease with an estimated annual incidence of 1:100,000/inhabitants, most commonly affects between 40 and 60 years and more frequently males (M:F=2:1).

To date, the pathogenesis of IRF is unknown, but is considered a multifactorial disease in which genetic and environmental factors interact determining susceptibility to the disease. Given the extreme rarity of the disease, the genetics of IRF is still poorly investigated, for which the associated genes are few; among them, there are genes related to major histocompatibility complex, in particular HLA-DRB1*03 and HLA-B*27.
Because many patients are seen in two centers in Italy (Parma, Prof. Dr. Vaglio; Milaan, Dr. Moroni) and in our center in the Netherlands, we collectively wanted to study the possible genetic basis of iRPF.

Study objective

The study is designed to evaluate the possible association between genetic variants and susceptibility to IRF, based on previous studies in autoimmune-inflammatory diseases (celiac disease, type 1 diabetes and rheumatoid arthritis).

Study design

In this study, the method used involves the genotyping of polymorphic DNA markers using case-control study to compare and evaluate allele frequencies of the marker in patients and healthy controls. For healthy controls will be used genotypes from data of 200 individuals of the Caucasian population deposited anonymously in online databases (http://www.1000genomes.org/; http://www.ncbi.nlm.nih.gov/;http://hapmap.ncbi.nlm.nih.gov/). There will not be recruitment of healthy individuals.

The study involves the use of the Immunochip platform (Illumina), which can simultaneously genotype 196,524 polymorphic markers, it includes 195.806 SNPs and 718 small insertions and deletions. The polymorphic markers included in the chip offer a high level of coverage in the region of the major histocompatibility complex (MHC).

To perform a genetic study with rare diseases, it is necessary to design a multicenter project; the entitycenter responsible for the sample recruitment is the Unit of Nephrology of the University Hospital of Parma (Prof. Dr. Augusto Vaglio, Dr. Palmisano, Dr. Maritati, Dr. Urban). The recruitment of samples will also include University of Milano-Bicocca (Dr. Gabriella Moroni) and the Albert Schweitzer hospital in the Netherlands (Dr. Eric van Bommel). These specialised centers follow patients in their clinical course and treatment.

Blood samples will be sent to the laboratory of Medical Genetics of the University Hospital of Parma (Responsible for sample storage Dr. Davide Martorana). DNA extraction (performed with QIAamp DNA Blood Mini Kit, Qiagen, Valencia, CA) will be performed by Dr. Davide Martorana.
Once extracted, the DNA samples will be sent for genotyping at the Institute of Parasitology and Biomedicine López Neyra, the Superior Council for Scientific Investigations, Armilla, Granada, Spain). The responsible for the execution of genetic testing is Prof Dr. Javier Martin.

Final bioinformatic and statistic analysis will be performed by Dr David Carmona (Institute of Parasitology and Biomedicine López Neyra, the Superior Council for Scientific Investigations, Armilla, Granada, Spain). The statistical analysis includes a case-control association test using a standard test Cochran-Armitage using PLINK v1.07.

At the end of the study, DNA samples will be destroyed.

Study burden and risks

There is no particular risk or burden for the patient when participating in this study, hence, we think performing this study is justified. There is a single extra collection of a 10 cc blood sample, which is not part of the standard care of the patient. There is no other burden for the patient, other than the regular visits to the outpatient department, blood sampling and radiologic investigations as part of the standard care and as such depicted in the 'Zorgpad RPF'.

Contacts

Public
Albert Schweitzer Ziekenhuis

Albert Schweitzer Plaats 25
Dordrecht 3318 AT
NL
Scientific
Albert Schweitzer Ziekenhuis

Albert Schweitzer Plaats 25
Dordrecht 3318 AT
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

Patients with idiopathic retroperitoneal fibrosis will be included

Exclusion criteria

Patients with secondary RPF will be excluded

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	05-12-2015
Enrollment:	100
Type:	Anticipated

Ethics review

Approved WMO

Date:	08-12-2015
Application type:	First submission
Review commission:	TWOR: Toetsingscommissie Wetenschappelijk Onderzoek Rotterdam e.o. (Rotterdam)

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	NL54831.101.15