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Institute of Rheumatology Research
Institut für Rheumaforschung
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Discovering new biomarkers to identify individuals in pre-clinical stages of rheumatoid arthritis

Project details

Research Leader	Prof. Axel Finckh, Division of Rheumatology, University Hospitals of Geneva
Participating Institutes	Prof. Emmanouil Dermitzakis, iGE3 genomics platform of Geneva University In collaboration with all Swiss academic Rheumatology units through the cohort study group of close relatives: www.arthritis-checkup.ch
Relevant conditions	Rheumatoid arthritis (RA)
Project duration	3 Jahre
Total project cost	479'127 CHF (159'709 per year)

What are the aims of this research?

The cause and subsequent development of rheumatoid arthritis (RA) is only partially understood but is believed to result from a multi-step process. The immune onset (i.e. development of autoimmunity associated by RA) is followed by a pre-clinical phase, which will eventually develop into symptomatic RA.

The first aim of this research is to pinpoint biomarkers for pre-clinical stages of RA, and in particular for "imminent RA", in order to identify candidates for a preventive therapeutic intervention, before these individuals ever develop arthritis. While no single genetic variant can sufficiently predict RA development, we hypothesize that a combination of different known and novel genetic variants, and their interactions with specific environmental factors, would be highly predictive in determining whether an individual will develop RA or not.

In a second step, using genome-wide association studies the research aims to integrate the insights into a genomic risk score for RA.

A third aim is to analyse the genetic expression signature of individuals at different stages of RA disease development using the RNAseq technology. We hope to be able to identify individuals with "imminent RA", or patients who will develop the disease during the next 12 months.

Why is this research important?

Autoimmune diseases, once fully developed, are often difficult to treat and generally require lifelong expensive therapy. Therapeutic interventions in the very early phases of the disease, or possibly before the clinical onset of the disease, can control the disease more effectively and may be curative. However, it is currently still unclear how to identify patients in the preclinical stages of the disease with sufficient precision to warrant starting healthy individuals on potentially dangerous immunomodulatory drugs.

Knowing the genetic associations and expression signature of RA and using existing knowledge of clinical risk factors would make it possible to identify individuals who are highly likely to develop RA (sometimes called "pre-RA"). Such candidates could be treated with a preventive therapy intervention, before they ever develop arthritis.

How will the findings benefit patients?

The project goals of a better understanding of the risk factors for RA, creating a genomic risk score and analysing the genetic expression signature are the first steps towards developing a screening strategy which aims to identify an imminent disease outbreak, or predict the future occurrence of RA. This would have huge potential benefits for individuals at risk.