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Genetic profiling and analysis for the presence of auto-antibodies in patients with axial spondylarthritis (Bechterew's disease)

Project details

Research Leader	Doctor Michael NISSEN, Department of Rheumatology, Geneva University Hospital (HUG)
Participating institutes	Department of Rheumatology, Geneva University Hospital and all Swiss Rheumatology units participating in the SCQM registry (8).
Relevant conditions	Axial spondyloarthritis (axSpA) - also known as ankylosing spondylarthritis or Bechterew's disease
Project duration	2 years
Total project cost	88,000 CHF

What are the aims of the research?

This is a prospective observational cohort study which studies patients with inflammatory rheumatic diseases who are registered in the Swiss Clinical Quality Management (SCQM) data base. The aim is to identify certain autoantibodies, which are specifically linked to AS, and others that are not. Some are well-known, (e.g. anti-CCP), and some are newly discovered (e.g. anti-CarP and anti-PAD3). The study will then correlate these autoantibodies with clinical characteristics, data on the distribution and determinants of disease (including environmental factors such as smoking), radiographic data and genetic data (using the genome-wide association study - GWAS).

Why is this research important?

Up to 2% of the population suffers from axial spondylarthritis (axSpA), which is also known as ankylosing spondylarthritis or Bechterew's disease. AxSpA is a painful, chronic, incurable and debilitating rheumatic disease which affects predominantly young adults and leads to fusion of the spine and sacroiliac joints in many patients.

Most axSpA patients do not demonstrate signs of inflammation in blood tests and standard radiographs are often normal. As early treatment is impeded by a delayed diagnosis, it is critical to identify new biomarkers, such as autoantibodies that improve early diagnosis. This knowledge may also contribute to a better assessment of disease activity, prognosis and response to therapy.

While axSpA is generally not considered to be an auto-immune disease, the presence of auto-antibodies is becoming increasingly well recognised. It is believed that AxSpA is caused by a combination of genetic, immunological and environmental factors, but most of these have still not been identified. A variation of the HLA-B gene called HLA-B27 increases the risk of developing the disease and is currently the only genetic test utilized in routine practice. However, not all people with axSpA have HLA-B27, and 90% of healthy people with this gene will never develop the disorder. There are over a hundred other genes known to be associated with axSpA that are not routinely tested and require further characterization, because they may be useful in diagnosis.

How will the findings benefit patients?

If biomarkers are found that could be used to test patients, clinicians could establish the diagnosis of axial spondylarthritis at an earlier stage and even identify predictive markers in people at risk, before disease breaks out. Thus, it would become possible to characterize the group of patients requiring early and intensive therapy.

As a result, patients will be relieved of their symptoms more rapidly and will present fewer complications in the future. In addition, all these data will be accessible for future research projects performed by other Swiss investigators using the SCQM database.