

Norwegian families multiplex for primary Sjögren's syndrome

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Introduction. Our collection of Norwegian families multiplex for primary Sjögren's syndrome (pSS) was initiated in 1996 with the long term purpose of identifying susceptibility genes that may contribute to the development of pSS. By January 1999 we had completed the collection of blood samples, labial salivary gland biopsies and clinical information, and have now fully characterised 6 families, all with Norwegian ancestors.

Patients and methods. The data collection from the patients and their healthy relatives included demographic and medical information. To be included in the study, the patients had to fulfil the European Community criteria for the classification of Sjögren's syndrome and have at least one close relative with pSS. Blood samples were requested from the patients and their siblings, parents, uncles, aunts and grandparents. Labial salivary gland biopsies were desired for inclusion. Serum was collected from patients and healthy relatives, and whole blood MNC has been immortalised by EBV transfection for subsequent access to DNA.

Results. The families included 14 affected with a mean age of 45.3 years (range 28-74, median 44.5 years) and 36 healthy relatives. All patients had oral symptoms, 78.6% had ocular symptoms, 21.4% has ocular signs, 57.1% has positive focus score, 28.6% had other forms of salivary gland involvement, 50% were anti-Ro/SSA positive and 28.4% anti-La/SSB positive. 71.4% were ANA positive, 57.1% were positive for rheumatoid factor, and 71.4% had symptoms of extraordinary fatigue. DNA are extracted and ready for genotyping using a set of 390 fluorescence-labelled primers for polymorphic microsatellite markers covering the complete human genome with a spacing between 10-20 cM. The set consists mainly of tetra- and tri-nucleotide repeats.

Discussion. This initial report describes a collection of pSS families in Norway with the purpose of performing a full genome scan to search for susceptibility genes for pSS. Since pSS is a complex disease, a very large cohort of families is required to get reliable results. Before starting the genetic analysis, attempts will therefore be made to collect more Scandinavian families.