

Genetic clue to development of mouth ulcers in lupus

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The results of a study presented today at the European League Against Rheumatism Annual Congress (EULAR 2016) showed for the first time an association between a specific genetic pathway and the development of mouth ulcers in patients with Systemic Lupus Erythematosus (SLE). Linking the vascular endothelial growth factor (VEGF) genetic pathway with a specific disease characteristic among SLE patients represents an important step towards unravelling the genetic basis of different SLE clinical presentations.

SLE is a genetically complex chronic relapsing autoimmune disease characterised by inflammation that may affect different tissues, including the skin, joint linings, lungs, kidneys and other organs. The disease is highly variable in the way it may present, and also in its outcome among individuals and across different ancestral groups. It often progresses to organ dysfunction and failure.²

"Understanding the relationships between specific SLE risk genes and different manifestations of the disease should help elucidate the underlying [disease](#) mechanisms and pathways," said Dr Antonio Julià of the Vall d'Hebron Research Institute, Barcelona, Spain. "Understanding more about the genetic pathways which underlie different manifestations of SLE is an important step towards the goal of improving the management of SLE, and ultimately to offer preventative care to individuals at increased risk of SLE," he added.

To identify new genetic variations associated with different SLE phenotypes, a total of 598,258 different regions on the genome (known as single-nucleotide polymorphisms) were genotyped in a population of 482 Caucasian European SLE patients of Spanish origin recruited from the rheumatology departments of 15 Spanish university hospitals belonging to the IMID Consortium (SLE group). A total of 11 clinically relevant SLE phenotypes were tested for association with over 700 reference genetic

pathways. In this discovery stage, two genetic pathways were significantly associated with the presence of mouth ulcers and the presence of antinuclear antibodies in SLE.

These two genetic pathways were then tested for validation in a second independent population of 425 SLE patients of the same southern European ancestry recruited from the same Spanish hospitals. In this replication stage, the significant association between mouth ulcers and the VEGF [genetic pathway](#) was confirmed.

"Finally, analysing the transcriptional effect of the topical immunotherapies used for the treatment of mouth ulcers in SLE, we found a significant differential expression of VEGF pathway genes," Dr Julià concluded.

Provided by European League Against Rheumatism

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