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Genomics reveals key macrophages' involvement in systemic sclerosis

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Singapore - A new international study has made an important discovery about the key role of macrophages, a type of immune cell, in systemic sclerosis (SSc), a chronic autoimmune disease which currently has no cure.

The research led by Enrico Petretto, Associate Professor at Duke-NUS Medical School (Duke-NUS), along with Dr Jacques Behmoaras at Imperial College London and collaborators from University College London in the UK, established for the first time a decisive link between immune cells, specifically the macrophages derived from SSc patients and systemic sclerosis. The study also demonstrated the role played by macrophages in the development of the disease due to known genetic factors, such as the case of the susceptibility gene *GSDMA,* which has been involved in cell death in the skin and was associated to the disease in 4,436 SSc patients, but whose function in macrophages from SSc patients was unclear.

"In the long quest for finding therapies for systemic sclerosis, our findings have implications for understanding the genetic basis of the disease, and we believe our discovery will prompt detailed functional studies in macrophages and immune cells, hopefully providing a starting point to develop greatly needed treatments for this disease" explained Professor Petretto, co-lead principal investigator and coordinator of the study.

Published in the Annals of Rheumatic Diseases (ARD*)* — the highest ranked journal in Rheumatology — in addition to confirming many genes previously implicated in the genetic predisposition to the disease, the study further discovered hundreds of genes that are previously not known to be associated with SSc. This provides a new starting point to better understand the disease aetiology, its genetic causes and develop therapies for SSc.

The study was supported by the Medical Research Council, UK, the National Research Foundation Singapore under its Cooperative Basic Research Grant (NMRC/CBRG/0106/2016) administered by the Singapore Ministry of Health's National Medical Research Council and Duke-NUS Medical School, as well as by the Arthritis Research UK, Scleroderma & Raynaud's UK and the Royal Free Charity, UK.