NIHR | BioResource

SSC. Systemic Sclerosis

NIHR BioResource – Rare Diseases study project

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Summary

Systemic Sclerosis (SSc; aka scleroderma) is a rare, complex autoimmune condition. It causes fibrosis (scarring), typically of the skin and can affect internal organs (such as the gut, lungs, heart and kidney). It is also associated with blood vessel abnormalities (called vasculopathy) and immune system activation (in which specific autoantibodies are detected in the blood). These features make the disease extremely variable – with different onset of disease, course, severity and outcome. It can be relatively benign to life threatening with significant patient and medical burden. The exact basis for the development of SSc remains unclear, although we know a combination of genes and environment leads to its development.

The rare nature of the disease makes large studies and therefore progress more challenging. There is an urgent need to understand the genetic and other drivers of SSc, to improve the diagnosis of patients, prediction of disease course, and importantly, develop new therapies. The NIHR Bioresource provides an important and exciting opportunity for SSc/ rheumatology centres in the country to work together to achieve these goals.

Recruitment Criteria

Inclusion

For Pre-SSc:
- Clinical diagnosis of Raynaud's Phenomenon
- Nail-fold capillaroscopy findings: presence of Mega capillaries, haemorrhages or capillary loss
  AND/OR
- presence of SSc specific ANA (Anti Centromere, Anti Topoisomerase / SCL-70 or anti RNA pol III etc.)

For SSc diagnosis:
- Clinical diagnosis +/-
- 1980 ACR criteria +/-
- 2013 EULAR ACR classification criteria