



Project Summary

How common is VEXAS syndrome in an adult rheumatology population? Multicentre study over 3 sites in South West England.

Dr Ben Mulhearn, RNHRD & Uni of Bath - £20,000.00

The VEXAS syndrome is a recently discovered condition which causes difficult-to-treat inflammatory disease and eventually leads to bone marrow failure. It is caused by a mutation on the X-chromosome which occurs later in life and therefore largely affects men over the age of 50. It can lead to several diseases seen in the rheumatology clinic including vasculitis and relapsing polychondritis. We therefore envisage that some patients with rheumatic diseases may have the VEXAS mutation and be 'hiding in plain sight'.

Patients with VEXAS syndrome may not respond as expected to immune-suppressing drugs. Furthermore, immune-suppressing drugs may even cause harm to these patients.

This project proposes to:

1. Identify potential VEXAS patients from current rheumatology clinics and invite them for genetic testing
2. Ensure patients with VEXAS syndrome are not taking any immune-suppressing drugs known to cause harm
3. Use blood from VEXAS patients in experiments to study this new condition further with the aim of finding better treatments
4. Follow patients up to investigate their long-term outcomes
5. Provide support to patients and their families

This study therefore has the potential to improve current individual patients' care and improve the prospects of future patients in general.