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Supporting Patients through Education & Research

Lay Summary - Myositis Autoantibodies- Dr Zoe Betteridge

Myositis is a rare autoimmune condition hallmarked by muscle inflammation, weakness, a characteristic skin rash, and in some cases associated cancer and lung disease. Due to the wide range of presenting features the diagnosis of myositis can be problematic; however the screening of myositis autoantibodies is increasingly viewed as a key tool in diagnosis. At the University of Bath we have now established a clinical service screening patient blood samples for myositis autoantibodies using the 'gold standard' technique termed radio-immunoprecipitation (IPP). Through this service and with our continuing collaborations with the UKMyoNet, EuMyonet and Juvenile Dermatomyositis Cohort Biomarker Studies we have now screened over 3000 myositis patients for autoantibodies using IPP. These results, alongside the corresponding clinical data have now been extensively analysed in collaboration with the Department of Mathematics, and a manuscript detailing the percentage of myositis patients with each particular autoantibody, the number of cases with more than one type of autoantibody and the clinical features associated with each specific autoantibody is now in preparation for submission. Additionally, an abstract regarding the mutual exclusivity of myositis specific autoantibodies has been accepted as a poster presentation at the British Society of Rheumatology conference (April 2016) and work from this project has been presented as an oral presentation at the 1st International Myositis meeting (May 2015). This work has also led to a review in the Journal of Internal Medicine and a chapter in 'The Myositis Handbook: An Inclusive Guide to the Inflammatory Myopathies'.

Data from this study has also been used in additional publications including an analysis of the autoantibodies in the Hungarian cohort and a study of HMGCR autoantibodies in patients from the Czech Republic. Additionally, work has been completed on a revised manuscript regarding the novel autoantibody; anti-EIF3 in patients with mild polymyositis, and the continued development of rapid tests for determining the levels of anti-TIF1 gamma, anti-NXP2 and anti-MDA5 autoantibodies. Furthermore, with the aid of two final year Pharmacology Students, preliminary work has been completed in the development of tests for anti-TIF1 alpha and beta autoantibodies, a study due to be continued as part of a BIRD studentship in the Summer (2016).

Furthermore as part of a Medical Research Council grant we have been able to complete preliminary work investigating the accuracy of a commercially available myositis autoantibody assay in comparison with our IPP data, as well as collaborating with a new industrial partner (Genalyte) in the development and validation of rapid myositis test. The preliminary results from this study have been presented by Prof Neil McHugh at the American College of Rheumatology meeting and will form the basis of a grant application to 'The Myositis Association' investigating the standardisation of commercial myositis autoantibody tests. Additionally, the line blot versus IPP data is currently being analysed and prepared for submission.

Scleroderma Autoantibody Studies

Our publication on the novel anti-EIF2B autoantibody, associated with diffuse systemic sclerosis and lung disease, has been accepted by Arthritis and Rheumatology. Work is now on going determining the presence of this autoantibody in a US cohort.

Autoantibodies in Lung Disease

Finally, as part of a collaboration with the University of Liverpool, 250 idiopathic pulmonary fibrosis (IPF) patients have been screened for autoantibodies by IPP, with 2% resulting in a known autoantibody and a further 40% of patients demonstrating strong bands of unknown specificity, (some of which are now being investigated further as potentially novel lung disease markers). Since patients with IPF have a poor prognosis, the detection of autoantibodies, demonstrating a potential misdiagnosis, has clinical relevance. These preliminary findings are forming part of an MRC Partnership Grant application investigating the use of autoantibody screening as part of IPF / CTD diagnosis in a larger cohort of lung disease patients.