



**Application number/Title:** 21024 - Genetics of ankylosing spondylitis

**Applicant PI:** Professor Matthew Brown

**Applicant institution:** Queensland University of Technology, Translational Research Institute, 37 Kent Street, Woolloongabba, Brisbane QLD 4102, Australia

**Lead Collaborators:** 1) Professor David Evans

**Collaborating Institutions and Addresses:** 1) University of Queensland, Diamantina Institute, 37 Kent Street, Woolloongabba, Brisbane QLD 4102, Australia

**Keywords provided by the Applicant PI to describe the research project:**

Arthritis, genetic risk prediction,,ankylosing spondylitis

**Application Lay Summary:**

**1a:** 1. To identify genes involved in ankylosing spondylitis  
2. To test genetic risk prediction for ankylosing spondylitis  
3. To test whether HLA-B27 influences the risk of ischaemic heart disease and mortality.

**1b:** The study aims to improve treatment and potentially prevention of AS both by identifying novel therapeutic targets, and developing methods to improve early diagnosis of the disease, which currently has a mean 10 year diagnostic delay.

**1c:** Ankylosing spondylitis cases will be identified from the registry. The accuracy of the diagnosis will be tested by investigating the prevalence of HLA-B27 in cases, which should be >80%. If that threshold is achieved then case genotypes will be compared with controls by standard GWAS approaches. Genetic risk scores developed using a 25000 case GWAS we are

currently completing will be tested for their positive and negative predictive value in the UK Biobank cohort.

**1d:** Full cohort