

# Matthew A Brown

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

492  
papers

59,009  
citations

93  
h-index

237  
g-index

532  
ext. papers

68,065  
ext. citations

9.3  
avg, IF

8  
L-index

#	Paper	IF	Citations
492	Effect of short-term hindlimb immobilization on skeletal muscle atrophy and the transcriptome in a low compared with high responder to endurance training model.. <i>PLoS ONE</i> , <b>2022</b> , 17, e0261723	3.7	0
491	Vaccine hesitancy and access to psoriasis care in the COVID-19 pandemic: findings from a global patient-reported cross-sectional survey.. <i>British Journal of Dermatology</i> , <b>2022</b> ,	4	1
490	Substitution mutational signatures in whole-genome sequenced cancers in the UK population. <i>Science</i> , <b>2022</b> , 376,	33.3	7
489	Multiple Endocrine Tumors Associated with Germline MAX Mutations: Multiple Endocrine Neoplasia Type 5?. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2021</b> , 106, 1163-1182	5.6	13
488	Humoral and cellular immunogenicity to a second dose of COVID-19 vaccine BNT162b2 in people receiving methotrexate or targeted immunosuppression: a longitudinal cohort study. <i>Lancet Rheumatology</i> , <b>2021</b> ,	14.2	12
487	Performance Evaluation of Multiple Ultrasonographical Methods for the Detection of Primary Sjögren's Syndrome. <i>Frontiers in Immunology</i> , <b>2021</b> , 12, 777322	8.4	
486	Polygenic risk scores and rheumatic diseases. <i>Chinese Medical Journal</i> , <b>2021</b> , 134, 2521-2524	2.9	1
485	The P4 study: Subsequent pregnancy maternal physiology after hypertensive and normotensive pregnancies. <i>Pregnancy Hypertension</i> , <b>2021</b> , 27, 29-34	2.6	0
484	The impact of Marfan syndrome on an Aboriginal Australian family: 'I don't like it as much as I don't like cancer'. <i>Journal of Genetic Counseling</i> , <b>2021</b> ,	2.5	1
483	Risk-mitigating behaviours in people with inflammatory skin and joint disease during the COVID-19 pandemic differ by treatment type: a cross-sectional patient survey. <i>British Journal of Dermatology</i> , <b>2021</b> , 185, 80-90	4	17
482	Comprehensive analysis of the major histocompatibility complex in systemic sclerosis identifies differential HLA associations by clinical and serological subtypes. <i>Annals of the Rheumatic Diseases</i> , <b>2021</b> ,	2.4	4
481	Polygenic Risk Scores have high diagnostic capacity in ankylosing spondylitis. <i>Annals of the Rheumatic Diseases</i> , <b>2021</b> , 80, 1168-1174	2.4	8
480	Functional Genomic Analysis of a RUNX3 Polymorphism Associated With Ankylosing Spondylitis. <i>Arthritis and Rheumatology</i> , <b>2021</b> , 73, 980-990	9.5	3
479	Repeated Spinal Mobility Measures and Their Association With Radiographic Damage in Ankylosing Spondylitis. <i>ACR Open Rheumatology</i> , <b>2021</b> , 3, 413-421	3.5	
478	Inflammasome Activation in Ankylosing Spondylitis Is Associated With Gut Dysbiosis. <i>Arthritis and Rheumatology</i> , <b>2021</b> , 73, 1189-1199	9.5	3
477	Genome wide association study of response to interval and continuous exercise training: the Predict-HIIT study. <i>Journal of Biomedical Science</i> , <b>2021</b> , 28, 37	13.3	2
476	Ankylosing spondylitis: an autoimmune or autoinflammatory disease?. <i>Nature Reviews Rheumatology</i> , <b>2021</b> , 17, 387-404	8.1	11

475	A polygenic resilience score moderates the genetic risk for schizophrenia. <i>Molecular Psychiatry</i> , <b>2021</b> , 26, 800-815	15.1	15
474	Low responders to endurance training exhibit impaired hypertrophy and divergent biological process responses in rat skeletal muscle. <i>Experimental Physiology</i> , <b>2021</b> , 106, 714-725	2.4	3
473	Germline ERBB3 mutation in familial non-small-cell lung carcinoma: expanding ErbB's role in oncogenesis. <i>Human Molecular Genetics</i> , <b>2021</b> , 30, 2393-2401	5.6	0
472	A KCNK16 mutation causing TALK-1 gain of function is associated with maturity-onset diabetes of the young. <i>JCI Insight</i> , <b>2021</b> , 6,	9.9	3
471	Identifying Trajectories of Radiographic Spinal Disease in Ankylosing Spondylitis: A 15-year follow up study of the PSOAS Cohort. <i>Rheumatology</i> , <b>2021</b> ,	3.9	2
470	Describing the burden of the COVID-19 pandemic in people with psoriasis: findings from a global cross-sectional study. <i>Journal of the European Academy of Dermatology and Venereology</i> , <b>2021</b> , 35, e636-e640	4.6	3
469	The effect of methotrexate and targeted immunosuppression on humoral and cellular immune responses to the COVID-19 vaccine BNT162b2: a cohort study. <i>Lancet Rheumatology, The</i> , <b>2021</b> , 3, e627-e637	14.2	47
468	Estimates of the rate of infection and asymptomatic COVID-19 disease in a population sample from SE England. <i>Journal of Infection</i> , <b>2020</b> , 81, 931-936	18.9	32
467	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , <b>2020</b> , 583, 90-95	50.4	69
466	Complement genes contribute sex-biased vulnerability in diverse disorders. <i>Nature</i> , <b>2020</b> , 582, 577-581	50.4	71
465	Causal Attributions in an Australian Aboriginal Family With Marfan Syndrome: A Qualitative Study. <i>Frontiers in Genetics</i> , <b>2020</b> , 11, 461	4.5	1
464	Normal human enthesis harbours conventional CD4+ and CD8+ T cells with regulatory features and inducible IL-17A and TNF expression. <i>Annals of the Rheumatic Diseases</i> , <b>2020</b> , 79, 1044-1054	2.4	33
463	Genomewide Association Study of Acute Anterior Uveitis Identifies New Susceptibility Loci <b>2020</b> , 61, 3		20
462	A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. <i>Nature Communications</i> , <b>2020</b> , 11, 3150	17.4	41
461	Altered Repertoire Diversity and Disease-Associated Clonal Expansions Revealed by T Cell Receptor Immunosequencing in Ankylosing Spondylitis Patients. <i>Arthritis and Rheumatology</i> , <b>2020</b> , 72, 1289-1302	9.5	16
460	Significant out-of-sample classification from methylation profile scoring for amyotrophic lateral sclerosis. <i>Npj Genomic Medicine</i> , <b>2020</b> , 5, 10	6.2	11
459	Longitudinal associations between depressive symptoms and clinical factors in ankylosing spondylitis patients: analysis from an observational cohort. <i>Rheumatology International</i> , <b>2020</b> , 40, 1053-1061	3.6	4
458	Biomarker development for axial spondyloarthritis. <i>Nature Reviews Rheumatology</i> , <b>2020</b> , 16, 448-463	8.1	20

457	Relapse Patterns in NMOSD: Evidence for Earlier Occurrence of Optic Neuritis and Possible Seasonal Variation. <i>Frontiers in Neurology</i> , <b>2020</b> , 11, 537	4.1	12
456	Whole-Exome Sequencing Reveals a Rare Missense Variant in in a Pedigree with Early-Onset Gout. <i>BioMed Research International</i> , <b>2020</b> , 2020, 4321419	3	5
455	Hippocampal plasticity underpins long-term cognitive gains from resistance exercise in MCI. <i>NeuroImage: Clinical</i> , <b>2020</b> , 25, 102182	5.3	25
454	Compound heterozygous mutations in FBN1 in a large family with Marfan syndrome. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2020</b> , 8, e1116	2.3	4
453	Use of the arm-span to height ratio as a criterion for Marfan syndrome in Aboriginal Australians: Diagnostically challenging. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 829-830	2.5	
452	Comparison of methods to construct a genetic risk score for prediction of rheumatoid arthritis in the population-based Nord-Trøndelag Health Study, Norway. <i>Rheumatology</i> , <b>2020</b> , 59, 1743-1751	3.9	2
451	Circular RNA sequencing indicates circ-IQGAP2 and circ-ZC3H6 as noninvasive biomarkers of primary Sjögren's syndrome. <i>Rheumatology</i> , <b>2020</b> , 59, 2603-2615	3.9	9
450	Multiple sclerosis risk variants regulate gene expression in innate and adaptive immune cells. <i>Life Science Alliance</i> , <b>2020</b> , 3,	5.8	10
449	A latent class based imputation method under Bayesian quantile regression framework using asymmetric Laplace distribution for longitudinal medication usage data with intermittent missing values. <i>Journal of Biopharmaceutical Statistics</i> , <b>2020</b> , 30, 160-177	1.3	1
448	Update on stem cell technologies in congenital heart disease. <i>Journal of Cardiac Surgery</i> , <b>2020</b> , 35, 174-179		1
447	Shotgun metagenomics reveals an enrichment of potentially cross-reactive bacterial epitopes in ankylosing spondylitis patients, as well as the effects of TNFi therapy upon microbiome composition. <i>Annals of the Rheumatic Diseases</i> , <b>2020</b> , 79, 132-140	2.4	47
446	Nonsteroidal Antiinflammatory Drug Use and Association With Incident Hypertension in Ankylosing Spondylitis. <i>Arthritis Care and Research</i> , <b>2020</b> , 72, 1645-1652	4.7	11
445	MRI compared with low-dose CT scanning in the diagnosis of axial spondyloarthritis. <i>Clinical Rheumatology</i> , <b>2020</b> , 39, 1295-1303	3.9	11
444	Genetic risk scores in inflammatory arthritis: a new era?. <i>Nature Reviews Rheumatology</i> , <b>2020</b> , 16, 545-548	1	2
443	Factors influencing cancer genetic somatic mutation test ordering by cancer physician. <i>Journal of Translational Medicine</i> , <b>2020</b> , 18, 431	8.5	4
442	Septic Shock: A Genomewide Association Study and Polygenic Risk Score Analysis. <i>Twin Research and Human Genetics</i> , <b>2020</b> , 23, 204-213	2.2	1
441	Patients with ACVR1 mutations have an increased prevalence of cardiac conduction abnormalities on electrocardiogram in a natural history study of Fibrodysplasia Ossificans Progressiva. <i>Orphanet Journal of Rare Diseases</i> , <b>2020</b> , 15, 193	4.2	6
440	Identification of susceptibility variants to benign childhood epilepsy with centro-temporal spikes (BECTS) in Chinese Han population. <i>EBioMedicine</i> , <b>2020</b> , 57, 102840	8.8	4

439	Genetics and the axial spondyloarthritis spectrum. <i>Rheumatology</i> , <b>2020</b> , 59, iv58-iv66	3.9	7
438	HLA-A alleles including HLA-A29 affect the composition of the gut microbiome: a potential clue to the pathogenesis of birdshot retinochoroidopathy. <i>Scientific Reports</i> , <b>2020</b> , 10, 17636	4.9	3
437	Epistatic interactions between killer immunoglobulin-like receptors and human leukocyte antigen ligands are associated with ankylosing spondylitis. <i>PLoS Genetics</i> , <b>2020</b> , 16, e1008906	6	8
436	Response to: Imputation-based analysis of MICA alleles in the susceptibility to ankylosing spondylitis by Zhou. <i>Annals of the Rheumatic Diseases</i> , <b>2020</b> , 79, e2	2.4	2
435	Prediction of Ankylosing Spondylitis in the HUNT Study by a Genetic Risk Score Combining 110 Single-nucleotide Polymorphisms of Genome-wide Significance. <i>Journal of Rheumatology</i> , <b>2020</b> , 47, 204-210	4.1	9
434	Gender Disparity in Inpatient Mortality After Transjugular Intrahepatic Portosystemic Shunt Creation in Patients Admitted With Hepatorenal Syndrome: A Nationwide Study. <i>Journal of the American College of Radiology</i> , <b>2020</b> , 17, 231-237	3.5	1
433	A Rare Mutation in SMAD9 Associated With High Bone Mass Identifies the SMAD-Dependent BMP Signaling Pathway as a Potential Anabolic Target for Osteoporosis. <i>Journal of Bone and Mineral Research</i> , <b>2020</b> , 35, 92-105	6.3	19
432	Genes Determining Nevus Count and Dermoscopic Appearance in Australian Melanoma Cases and Controls. <i>Journal of Investigative Dermatology</i> , <b>2020</b> , 140, 498-501.e17	4.3	6
431	MHC associations of ankylosing spondylitis in East Asians are complex and involve non-HLA-B27 HLA contributions. <i>Arthritis Research and Therapy</i> , <b>2020</b> , 22, 74	5.7	5
430	Epistatic interactions between killer immunoglobulin-like receptors and human leukocyte antigen ligands are associated with ankylosing spondylitis <b>2020</b> , 16, e1008906		
429	Epistatic interactions between killer immunoglobulin-like receptors and human leukocyte antigen ligands are associated with ankylosing spondylitis <b>2020</b> , 16, e1008906		
428	Epistatic interactions between killer immunoglobulin-like receptors and human leukocyte antigen ligands are associated with ankylosing spondylitis <b>2020</b> , 16, e1008906		
427	Epistatic interactions between killer immunoglobulin-like receptors and human leukocyte antigen ligands are associated with ankylosing spondylitis <b>2020</b> , 16, e1008906		
426	Vitamin D-Binding Protein Deficiency and Homozygous Deletion of the GC Gene. <i>New England Journal of Medicine</i> , <b>2019</b> , 380, 2584-2585	59.2	3
425	Best practices in DNA methylation: lessons from inflammatory bowel disease, psoriasis and ankylosing spondylitis. <i>Arthritis Research and Therapy</i> , <b>2019</b> , 21, 133	5.7	15
424	Elevated / Expression During the Disease Process of Primary Sjögren's Syndrome. <i>Frontiers in Immunology</i> , <b>2019</b> , 10, 795	8.4	14
423	Natural history of fibrodysplasia ossificans progressiva: cross-sectional analysis of annotated baseline phenotypes. <i>Orphanet Journal of Rare Diseases</i> , <b>2019</b> , 14, 98	4.2	20
422	HLA Alleles Associated With Risk of Ankylosing Spondylitis and Rheumatoid Arthritis Influence the Gut Microbiome. <i>Arthritis and Rheumatology</i> , <b>2019</b> , 71, 1642-1650	9.5	63

421	Response to Comment on Johnson et al. Cost-effectiveness Analysis of Routine Screening Using Massively Parallel Sequencing for Maturity-Onset Diabetes of the Young in a Pediatric Diabetes Cohort: Reduced Health System Costs and Improved Patient Quality of Life. <i>Diabetes Care</i> 2019;42:69-76. <i>Diabetes Care</i> , 2019, 42, e79-e80	14.6	
420	Genetics of Axial Spondyloarthritis 2019, 67-85		
419	The Gut Microbiome and Ankylosing Spondylitis 2019, 87-95		1
418	Mice with a Brd4 Mutation Represent a New Model of Nephrocalcinosis. <i>Journal of Bone and Mineral Research</i> , 2019, 34, 1324-1335	6.3	3
417	Genetic susceptibility to cervical neoplasia. <i>Papillomavirus Research (Amsterdam, Netherlands)</i> , 2019, 7, 132-134	4.6	6
416	Genome-wide association study in Turkish and Iranian populations identify rare familial Mediterranean fever gene (MEFV) polymorphisms associated with ankylosing spondylitis. <i>PLoS Genetics</i> , 2019, 15, e1008038	6	22
415	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. <i>American Journal of Human Genetics</i> , 2019, 104, 948-956	11	17
414	Population-based identity-by-descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2019, 180, 223-231	3.5	2
413	Association of Genetic Variants in NUDT15 With Thiopurine-Induced Myelosuppression in Patients With Inflammatory Bowel Disease. <i>JAMA - Journal of the American Medical Association</i> , 2019, 321, 773-783	27.4	75
412	Comparative performances of machine learning methods for classifying Crohn Disease patients using genome-wide genotyping data. <i>Scientific Reports</i> , 2019, 9, 10351	4.9	44
411	Association of Schizophrenia Risk With Disordered Niacin Metabolism in an Indian Genome-wide Association Study. <i>JAMA Psychiatry</i> , 2019, 76, 1026-1034	14.5	24
410	GWAS for systemic sclerosis identifies multiple risk loci and highlights fibrotic and vasculopathy pathways. <i>Nature Communications</i> , 2019, 10, 4955	17.4	46
409	Comprehensive genetic screening: The prevalence of maturity-onset diabetes of the young gene variants in a population-based childhood diabetes cohort. <i>Pediatric Diabetes</i> , 2019, 20, 57-64	3.6	13
408	An N-Ethyl-N-Nitrosourea (ENU)-Induced Tyr265Stop Mutation of the DNA Polymerase Accessory Subunit Gamma 2 (Polg2) Is Associated With Renal Calcification in Mice. <i>Journal of Bone and Mineral Research</i> , 2019, 34, 497-507	6.3	2
407	HLA class I and II alleles in susceptibility to ankylosing spondylitis. <i>Annals of the Rheumatic Diseases</i> , 2019, 78, 66-73	2.4	31
406	Cost-effectiveness Analysis of Routine Screening Using Massively Parallel Sequencing for Maturity-Onset Diabetes of the Young in a Pediatric Diabetes Cohort: Reduced Health System Costs and Improved Patient Quality of Life. <i>Diabetes Care</i> , 2019, 42, 69-76	14.6	15
405	Analysis of the genetic component of systemic sclerosis in Iranian and Turkish populations through a genome-wide association study. <i>Rheumatology</i> , 2019, 58, 289-298	3.9	8
404	Special considerations for clinical trials in fibrodysplasia ossificans progressiva (FOP). <i>British Journal of Clinical Pharmacology</i> , 2019, 85, 1199-1207	3.8	14

403	Somatic POLE exonuclease domain mutations are early events in sporadic endometrial and colorectal carcinogenesis, determining driver mutational landscape, clonal neoantigen burden and immune response. <i>Journal of Pathology</i> , <b>2018</b> , 245, 283-296	9.4	43
402	Diffusion-weighted Imaging Is a Sensitive and Specific Magnetic Resonance Sequence in the Diagnosis of Ankylosing Spondylitis. <i>Journal of Rheumatology</i> , <b>2018</b> , 45, 771-778	4.1	28
401	Whole-exome sequencing for mutation detection in pediatric disorders of insulin secretion: Maturity onset diabetes of the young and congenital hyperinsulinism. <i>Pediatric Diabetes</i> , <b>2018</b> , 19, 656-662	3.6	13
400	Point mutation in p14 -specific exon 1 of CDKN2A causing familial melanoma and astrocytoma. <i>British Journal of Dermatology</i> , <b>2018</b> , 178, e263-e264	4	0
399	Clinical usefulness of comprehensive genetic screening in maturity onset diabetes of the young (MODY): A novel ABCC8 mutation in a previously screened family. <i>Journal of Diabetes</i> , <b>2018</b> , 10, 764-767	3.8	10
398	An -Ethyl-Nitrosourea (ENU) Mutagenized Mouse Model for Autosomal Dominant Nonsyndromic Kyphoscoliosis Due to Vertebral Fusion. <i>JBMR Plus</i> , <b>2018</b> , 2, 154-163	3.9	1
397	Imputation-based analysis of alleles in the susceptibility to ankylosing spondylitis. <i>Annals of the Rheumatic Diseases</i> , <b>2018</b> , 77, 1691-1692	2.4	10
396	Genetic variation in is associated with bacteremia secondary to diverse pathogens in African children. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2018</b> , 115, E3601-E3603	11.5	3
395	Loss-of-function nuclear factor B subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. <i>Journal of Allergy and Clinical Immunology</i> , <b>2018</b> , 142, 1285-1296	11.5	109
394	Evidence for a second ankylosing spondylitis-associated regulatory polymorphism. <i>RMD Open</i> , <b>2018</b> , 4, e000628	5.9	12
393	Evaluation of the effect of baseline MRI sacroiliitis and C reactive protein status on etanercept treatment response in non-radiographic axial spondyloarthritis: a post hoc analysis of the EMBARK study. <i>Annals of the Rheumatic Diseases</i> , <b>2018</b> , 77, 1091-1093	2.4	10
392	Solving the pathogenesis of ankylosing spondylitis. <i>Clinical Immunology</i> , <b>2018</b> , 186, 46-50	9	4
391	Genome-Wide Association Studies <b>2018</b> , 33-41		0
390	Genome-wide association study in Guillain-Barré syndrome. <i>Journal of Neuroimmunology</i> , <b>2018</b> , 323, 109-114	3.5	9
389	Longitudinal expression profiling of CD4+ and CD8+ cells in patients with active to quiescent giant cell arteritis. <i>BMC Medical Genomics</i> , <b>2018</b> , 11, 61	3.7	8
388	Non-classical human leucocyte antigens in ankylosing spondylitis: possible association with HLA-E and HLA-F. <i>RMD Open</i> , <b>2018</b> , 4, e000677	5.9	11
387	A multiple imputation method based on weighted quantile regression models for longitudinal censored biomarker data with missing values at early visits. <i>BMC Medical Research Methodology</i> , <b>2018</b> , 18, 8	4.7	8
386	Type 1 diabetes susceptibility alleles are associated with distinct alterations in the gut microbiota. <i>Microbiome</i> , <b>2018</b> , 6, 35	16.6	40

385	Genome-wide association studies for diabetic macular edema and proliferative diabetic retinopathy. <i>BMC Medical Genetics</i> , <b>2018</b> , 19, 71	2.1	32
384	HLA and KIR Associations of Cervical Neoplasia. <i>Journal of Infectious Diseases</i> , <b>2018</b> , 218, 2006-2015	7	15
383	Harmonization, data management, and statistical issues related to prospective multicenter studies in Ankylosing spondylitis (AS): Experience from the Prospective Study Of Ankylosing Spondylitis (PSOAS) cohort. <i>Contemporary Clinical Trials Communications</i> , <b>2018</b> , 11, 127-135	1.8	8
382	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. <i>Cell</i> , <b>2018</b> , 173, 1705-1715.e16	56.2	360
381	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 3-18	11	27
380	Genome-wide association study of extreme high bone mass: Contribution of common genetic variation to extreme BMD phenotypes and potential novel BMD-associated genes. <i>Bone</i> , <b>2018</b> , 114, 62-71	4.7	25
379	Association of Crohn's disease-related chromosome 1q32 with ankylosing spondylitis is independent of bowel symptoms and faecal calprotectin. <i>PeerJ</i> , <b>2018</b> , 6, e5088	3.1	2
378	The FOP Connection Registry: Design of an international patient-sponsored registry for Fibrodysplasia Ossificans Progressiva. <i>Bone</i> , <b>2018</b> , 109, 285-290	4.7	13
377	Identification of a novel locus on chromosome 2q13, which predisposes to clinical vertebral fractures independently of bone density. <i>Annals of the Rheumatic Diseases</i> , <b>2018</b> , 77, 378-385	2.4	15
376	Opioid Analgesic Use in Patients with Ankylosing Spondylitis: An Analysis of the Prospective Study of Outcomes in an Ankylosing Spondylitis Cohort. <i>Journal of Rheumatology</i> , <b>2018</b> , 45, 188-194	4.1	13
375	Genetic Variants in ERAP1 and ERAP2 Associated With Immune-Mediated Diseases Influence Protein Expression and the Isoform Profile. <i>Arthritis and Rheumatology</i> , <b>2018</b> , 70, 255-265	9.5	34
374	Invasive dermatophyte infection with <i>Trichophyton interdigitale</i> is associated with prurigo-induced pseudoperforation and a signal transducer and activator of transcription 3 mutation. <i>British Journal of Dermatology</i> , <b>2018</b> , 179, 750-754	4	9
373	Novel pleiotropic risk loci for melanoma and nevus density implicate multiple biological pathways. <i>Nature Communications</i> , <b>2018</b> , 9, 4774	17.4	47
372	Assessment of the genetic and clinical determinants of fracture risk: genome wide association and mendelian randomisation study. <i>BMJ, The</i> , <b>2018</b> , 362, k3225	5.9	114
371	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. <i>Nature Communications</i> , <b>2018</b> , 9, 1864	17.4	37
370	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 144-153	11	18
369	Rare variants in Fanconi anemia genes are enriched in acute myeloid leukemia. <i>Blood Cancer Journal</i> , <b>2018</b> , 8, 50	7	13
368	Ethics of genetic testing and research in sport: a position statement from the Australian Institute of Sport. <i>British Journal of Sports Medicine</i> , <b>2017</b> , 51, 5-11	10.3	34

367	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 334-342	11	14
366	Genetic association of ankylosing spondylitis with TBX21 influences T-bet and pro-inflammatory cytokine expression in humans and SKG mice as a model of spondyloarthritis. <i>Annals of the Rheumatic Diseases</i> , <b>2017</b> , 76, 261-269	2.4	28
365	Homozygous variant in C21orf2 in a case of Jeune syndrome with severe thoracic involvement: Extending the phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1698-1704	2.5	9
364	Self-reported Diagnosis of Rheumatoid Arthritis or Ankylosing Spondylitis Has Low Accuracy: Data from the Nord-Trøndelag Health Study. <i>Journal of Rheumatology</i> , <b>2017</b> , 44, 1134-1141	4.1	21
363	Pathogenesis of ankylosing spondylitis - recent advances and future directions. <i>Nature Reviews Rheumatology</i> , <b>2017</b> , 13, 359-367	8.1	143
362	Epigenetic and gene expression analysis of ankylosing spondylitis-associated loci implicate immune cells and the gut in the disease pathogenesis. <i>Genes and Immunity</i> , <b>2017</b> , 18, 135-143	4.4	19
361	Incidence and prevalence of NMOSD in Australia and New Zealand. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2017</b> , 88, 632-638	5.5	72
360	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 75-90	11	235
359	Consensus statements on the imaging of axial spondyloarthritis in Australia and New Zealand. <i>Journal of Medical Imaging and Radiation Oncology</i> , <b>2017</b> , 61, 58-69	1.7	7
358	ERAP1 association with ankylosing spondylitis is attributable to common genotypes rather than rare haplotype combinations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2017</b> , 114, 558-561	11.5	25
357	Geo-epidemiology of temporal artery biopsy-positive giant cell arteritis in Australia and New Zealand: is there a seasonal influence?. <i>RMD Open</i> , <b>2017</b> , 3, e000531	5.9	9
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54	Whole-genome linkage analysis of rheumatoid arthritis susceptibility loci in 252 affected sibling pairs in the United Kingdom. <i>Arthritis and Rheumatism</i> , <b>2002</b> , 46, 632-9		171
53	Role of NOD2 variants in spondylarthritis. <i>Arthritis and Rheumatism</i> , <b>2002</b> , 46, 1629-33		77
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4	Pancytopenia after accidental overdose of methotrexate. A complication of low-dose therapy for rheumatoid arthritis. <i>Medical Journal of Australia</i> , <b>1991</b> , 155, 493-4	4	25
3	Concordance of B and T cell responses to SARS-CoV-2 infection, irrespective of symptoms suggestive of COVID-19		1
2	Shotgun metagenomics reveals an enrichment of potentially cross-reactive bacterial epitopes in ankylosing spondylitis patients, as well as the effects of TNFi therapy and the host's genotype upon microbiome composition		3
1	Missense mutations in the MLKL BACE1 region lead to lethal neonatal inflammation in mice and are present in high frequency in humans		4