David L Duffy

List of Publications by Year in Descending Order

272 papers	15,065 citations	67 h-index	115 g-index
284 ext. papers	16,816 ext. citations	6.8 avg, IF	5.74 L-index

#	Paper	IF	Citations
272	Genome-Wide Association Study Suggests the Variant rs7551288*A within the DHCR24 Gene Is Associated with Poor Overall Survival in Melanoma Patients. *Cancers*, **2022**, 14, 2410	6.6	0
271	The Heritability of Twinning in Seven Large Historic Pedigrees.. *Twin Research and Human Genetics*, **2022**, 1-4	2.2	0
270	Germline variants are associated with increased primary melanoma tumor thickness at diagnosis. *Human Molecular Genetics*, **2021**, 29, 3578-3587	5.6	1
269	Genome-wide association study in almost 195,000 individuals identifies 50 previously unidentified genetic loci for eye color. *Science Advances*, **2021**, 7,	14.3	11
268	Essential TremorÕPhenotype in FMR1 Premutation/Gray Zone Sibling Series: Exploring Possible Genetic Modifiers. *Twin Research and Human Genetics*, **2021**, 24, 95-102	2.2	1
267	Genome-wide association study identifies 48 common genetic variants associated with handedness. *Nature Human Behaviour*, **2021**, 5, 59-70	12.8	33
266	Candidate Glycoprotein Biomarkers for Canine Visceral Hemangiosarcoma and Validation Using Semi-Quantitative Lectin/Immunohistochemical Assays. *Veterinary Sciences*, **2021**, 8,	2.4	1
265	Addressing Delicate and Variable Cancer Morphology in Spectral Histopathology Using Canine Visceral Hemangiosarcoma. *Analytical Chemistry*, **2021**, 93, 12187-12194	7.8	3
264	Gene Discovery Using Twins. *Twin Research and Human Genetics*, **2020**, 23, 90-93	2.2	
263	CDKN2A testing threshold in a high-risk Australian melanoma cohort: number of primaries, family history and young age of onset impact risk. *Journal of the European Academy of Dermatology and Venereology*, **2020**, 34, e797-e798	4.6	
262	Genome-wide association meta-analyses combining multiple risk phenotypes provide insights into the genetic architecture of cutaneous melanoma susceptibility. *Nature Genetics*, **2020**, 52, 494-504	36.3	39
261	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants. *PLoS ONE*, **2020**, 15, e0238529	3.7	5
260	The interplay of sun damage and genetic risk in Australian multiple and single primary melanoma cases and controls. *British Journal of Dermatology*, **2020**, 183, 357-366	4	7
259	Multiplex melanoma families are enriched for polygenic risk. *Human Molecular Genetics*, **2020**, 29, 2976-2985	5.6	3
258	Genes Determining Nevus Count and Dermoscopic Appearance in Australian Melanoma Cases and Controls. *Journal of Investigative Dermatology*, **2020**, 140, 498-501.e17	4.3	6
257	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		
256	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		

255	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		
254	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		
253	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		
252	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		
251	Germline and somatic albinism variants in amelanotic/hypomelanotic melanoma: Increased carriage of TYR and OCA2 variants **2020**, 15, e0238529		
250	Phenotypic and genotypic analysis of amelanotic and hypomelanotic melanoma patients. *Journal of the European Academy of Dermatology and Venereology*, **2019**, 33, 1076-1083	4.6	6
249	Noncoding Variations in the Gene Encoding Ceramide Synthase 6 are Associated with Type 2 Diabetes in a Large Indigenous Australian Pedigree. *Twin Research and Human Genetics*, **2019**, 22, 79-87	2.2	1
248	High naevus count and MC1R red hair alleles contribute synergistically to increased melanoma risk. *British Journal of Dermatology*, **2019**, 181, 1009-1016	4	15
247	Genome-Wide Association Studies Identify Multiple Genetic Loci Influencing Eyebrow Color Variation in Europeans. *Journal of Investigative Dermatology*, **2019**, 139, 1601-1605	4.3	10
246	Variation at and Asthma on the Island of Tristan da Cunha. *Twin Research and Human Genetics*, **2019**, 22, 277-282	2.2	0
245	Genetics and Gene-Environment Interactions in Childhood and Adult Onset Asthma. *Frontiers in Pediatrics*, **2019**, 7, 499	3.4	30
244	IRF4 rs12203592*T/T genotype is associated with nodular melanoma. *Melanoma Research*, **2019**, 29, 445-446	3.3	1
243	MC1R minor variants and the multiple pathways to melanoma. *The Lancet Child and Adolescent Health*, **2019**, 3, 287-288	14.5	
242	Genome-wide association meta-analysis of individuals of European ancestry identifies new loci explaining a substantial fraction of hair color variation and heritability. *Nature Genetics*, **2018**, 50, 652-656	36.3	59
241	Iris pigmented lesions as a marker of cutaneous melanoma risk: an Australian case-control study. *British Journal of Dermatology*, **2018**, 178, 1119-1127	4	12
240	Accuracy of Inferred APOE Genotypes for a Range of Genotyping Arrays and Imputation Reference Panels. *Journal of Alzheimeris Disease*, **2018**, 64, 49-54	4.3	5
239	The Relationship Between Personality and Somatic and Psychological Distress: A Comparison of Chinese and Australian Adolescents. *Behavior Genetics*, **2018**, 48, 315-322	3.2	5
238	Genome-wide association study in 176,678 Europeans reveals genetic loci for tanning response to sun exposure. *Nature Communications*, **2018**, 9, 1684	17.4	51

237	Novel pleiotropic risk loci for melanoma and nevus density implicate multiple biological pathways. *Nature Communications*, **2018**, 9, 4774	17.4	47
236	Cell-type-specific eQTL of primary melanocytes facilitates identification of melanoma susceptibility genes. *Genome Research*, **2018**, 28, 1621-1635	9.7	33
235	Mind your Moles study: protocol of a prospective cohort study of melanocytic naevi. *BMJ Open*, **2018**, 8, e025857	3	8
234	GSTP1 does not modify MC1R effects on melanoma risk. *Experimental Dermatology*, **2017**, 26, 730-733	4	9
233	Genome-Wide Association Shows that Pigmentation Genes Play a Role in Skin Aging. *Journal of Investigative Dermatology*, **2017**, 137, 1887-1894	4.3	30
232	Analysis of Quantitative Trait Loci. *Methods in Molecular Biology*, **2017**, 1526, 191-203	1.4	1
231	Shared genetic origin of asthma, hay fever and eczema elucidates allergic disease biology. *Nature Genetics*, **2017**, 49, 1752-1757	36.3	256
230	Classifying dermoscopic patterns of naevi in a case-control study of melanoma. *PLoS ONE*, **2017**, 12, e0186647	3.7	3
229	Familial aggregation of albuminuria and arterial hypertension in an Aboriginal Australian community and the contribution of variants in ACE and TP53. *BMC Nephrology*, **2016**, 17, 183	2.7	10
228	Variation in Latent Classes of Adult Attention-Deficit Hyperactivity Disorder by Sex and Environmental Adversity. *Journal of Attention Disorders*, **2016**, 20, 934-945	3.7	1
227	Head Motion and Inattention/Hyperactivity Share Common Genetic Influences: Implications for fMRI Studies of ADHD. *PLoS ONE*, **2016**, 11, e0146271	3.7	41
226	Cysteinyl Leukotrienes Pathway Genes, Atopic Asthma and Drug Response: From Population Isolates to Large Genome-Wide Association Studies. *Frontiers in Pharmacology*, **2016**, 7, 299	5.6	21
225	Heritability of naevus patterns in an adult twin cohort from the Brisbane Twin Registry: a cross-sectional study. *British Journal of Dermatology*, **2016**, 174, 356-63	4	12
224	Genome-wide meta-analysis identifies five new susceptibility loci for cutaneous malignant melanoma. *Nature Genetics*, **2015**, 47, 987-995	36.3	162
223	Contrast effects and sex influence maternal and self-report dimensional measures of attention-deficit hyperactivity disorder. *Behavior Genetics*, **2015**, 45, 35-50	3.2	10
222	The prognostic significance of the 2014 International Society of Urological Pathology (ISUP) grading system for prostate cancer. *Pathology*, **2015**, 47, 515-9	1.6	39
221	Genetics of Eye Colour **2015**, 1-9		3
220	Potential Modifying Loci Associated With Primary Lens Luxation, Pedal Hyperkeratosis, and Ocular Phenotypes in Miniature Bull Terriers **2015**, 56, 8288-96		1

219	Genetics of skin color variation in Europeans: genome-wide association studies with functional follow-up. *Human Genetics*, **2015**, 134, 823-35	6.3	97
218	Meta-analysis identifies seven susceptibility loci involved in the atopic march. *Nature Communications*, **2015**, 6, 8804	17.4	105
217	Molecular analysis of common polymorphisms within the human Tyrosinase locus and genetic association with pigmentation traits. *Pigment Cell and Melanoma Research*, **2014**, 27, 552-64	4.5	26
216	Genome-wide association analysis identifies 11 risk variants associated with the asthma with hay fever phenotype. *Journal of Allergy and Clinical Immunology*, **2014**, 133, 1564-71	11.5	143
215	Early life environmental predictors of asthma age-of-onset. *Immunity, Inflammation and Disease*, **2014**, 2, 141-51	2.4	7
214	Distribution analyses of acquired melanocytic naevi on the trunk. *Dermatology*, **2014**, 228, 269-75	4.4	2
213	Identification of a melanoma susceptibility locus and somatic mutation in TET2. *Carcinogenesis*, **2014**, 35, 2097-101	4.6	38
212	Phenotypic characterization of nevus and tumor patterns in MITF E318K mutation carrier melanoma patients. *Journal of Investigative Dermatology*, **2014**, 134, 141-149	4.3	49
211	Meta-analysis of genome-wide association studies identifies ten loci influencing allergic sensitization. *Nature Genetics*, **2013**, 45, 902-906	36.3	191
210	First all-in-one diagnostic tool for DNA intelligence: genome-wide inference of biogeographic ancestry, appearance, relatedness, and sex with the Identitas v1 Forensic Chip. *International Journal of Legal Medicine*, **2013**, 127, 559-72	3.1	38
209	Investigation of diabetes mellitus in Burmese cats as an inherited trait: a preliminary study. *New Zealand Veterinary Journal*, **2013**, 61, 354-8	1.7	9
208	A new regulatory variant in the interleukin-6 receptor gene associates with asthma risk. *Genes and Immunity*, **2013**, 14, 441-6	4.4	25
207	Genome-wide association study of inattention and hyperactivity-impulsivity measured as quantitative traits. *Twin Research and Human Genetics*, **2013**, 16, 560-74	2.2	46
206	Atopic dermatitis in West Highland white terriers is associated with a 1.3-Mb region on CFA 17. *Immunogenetics*, **2012**, 64, 209-17	3.2	20
205	Human pigmentation genes under environmental selection. *Genome Biology*, **2012**, 13, 248	18.3	120
204	Genome-wide association study to identify genetic determinants of severe asthma. *Thorax*, **2012**, 67, 762-8	7.3	139
203	Meta-analysis combining new and existing data sets confirms that the TERT-CLPTM1L locus influences melanoma risk. *Journal of Investigative Dermatology*, **2012**, 132, 485-7	4.3	38
202	Genome-wide association studies of asthma in population-based cohorts confirm known and suggested loci and identify an additional association near HLA. *PLoS ONE*, **2012**, 7, e44008	3.7	89

201	Attention deficit hyperactivity disorder in Australian adults: prevalence, persistence, conduct problems and disadvantage. *PLoS ONE*, **2012**, 7, e47404	3.7	67
200	Meta-analysis of genome-wide association studies identifies three new risk loci for atopic dermatitis. *Nature Genetics*, **2011**, 44, 187-92	36.3	244
199	Genome-wide association study identifies three new melanoma susceptibility loci. *Nature Genetics*, **2011**, 43, 1108-13	36.3	203
198	Real-time PCR quantification of the canine filaggrin orthologue in the skin of atopic and non-atopic dogs: a pilot study. *BMC Research Notes*, **2011**, 4, 554	2.3	8
197	Identification of IL6R and chromosome 11q13.5 as risk loci for asthma. *Lancet, The*, **2011**, 378, 1006-14	40	298
196	Pathway-based analysis of a melanoma genome-wide association study: analysis of genes related to tumour-immunosuppression. *PLoS ONE*, **2011**, 6, e29451	3.7	16
195	Tertiary Gleason pattern 5 on needle biopsy predicts greater tumour volume on radical prostatectomy. *Pathology*, **2011**, 43, 693-6	1.6	4
194	Risk of asthma in adult twins with type 2 diabetes and increased body mass index. *Allergy: European Journal of Allergy and Clinical Immunology*, **2011**, 66, 562-8	9.3	29
193	Relationship between type 1 diabetes and atopic diseases in a twin population. *Allergy: European Journal of Allergy and Clinical Immunology*, **2011**, 66, 645-7	9.3	21
192	High allergen-specific serum immunoglobulin E levels in nonatopic West Highland white terriers. *Veterinary Dermatology*, **2011**, 22, 257-66	1.8	21
191	Association between ORMDL3, IL1RL1 and a deletion on chromosome 17q21 with asthma risk in Australia. *European Journal of Human Genetics*, **2011**, 19, 458-64	5.3	92
190	GWAS findings for human iris patterns: associations with variants in genes that influence normal neuronal pattern development. *American Journal of Human Genetics*, **2011**, 89, 334-43	11	47
189	PTPN22 polymorphisms may indicate a role for this gene in atopic dermatitis in West Highland white terriers. *BMC Research Notes*, **2011**, 4, 571	2.3	11
188	Genome-wide association study identifies novel loci predisposing to cutaneous melanoma. *Human Molecular Genetics*, **2011**, 20, 5012-23	5.6	164
187	Polymorphisms in nevus-associated genes MTAP, PLA2G6, and IRF4 and the risk of invasive cutaneous melanoma. *Twin Research and Human Genetics*, **2011**, 14, 422-32	2.2	34
186	IgE responsiveness to Dermatophagoides farinae in West Highland white terrier dogs is associated with region on CFA35. *Journal of Heredity*, **2011**, 102 Suppl 1, S74-80	2.4	10
185	Variation in BMPR1B, TGFRB1 and BMPR2 and control of dizygotic twinning. *Twin Research and Human Genetics*, **2011**, 14, 408-16	2.2	19
184	A novel recurrent mutation in MITF predisposes to familial and sporadic melanoma. *Nature*, **2011**, 480, 99-103	50.4	335

183	Report of Endometrial Cancer in Australian BRCA1 and BRCA2 mutation-positive Families. *Twin Research and Human Genetics*, **2011**, 14, 111-8	2.2	12
182	Genome-wide association study identifies a new melanoma susceptibility locus at 1q21.3. *Nature Genetics*, **2011**, 43, 1114-8	36.3	126
181	A non-synonymous mutation in the canine Pkd1 gene is associated with autosomal dominant polycystic kidney disease in Bull Terriers. *PLoS ONE*, **2011**, 6, e22455	3.7	10
180	Meta-analysis of 20 genome-wide linkage studies evidenced new regions linked to asthma and atopy. *European Journal of Human Genetics*, **2010**, 18, 700-6	5.3	44
179	Characterization of the methylation patterns of MS4A2 in atopic cases and controls. *Allergy: European Journal of Allergy and Clinical Immunology*, **2010**, 65, 333-7	9.3	8
178	Multiple pigmentation gene polymorphisms account for a substantial proportion of risk of cutaneous malignant melanoma. *Journal of Investigative Dermatology*, **2010**, 130, 520-8	4.3	144
177	Evaluation of candidate stromal epithelial cross-talk genes identifies association between risk of serous ovarian cancer and TERT, a cancer susceptibility "hot-spot". *PLoS Genetics*, **2010**, 6, e1001016	6	42
176	Association of MC1R variants and host phenotypes with melanoma risk in CDKN2A mutation carriers: a GenoMEL study. *Journal of the National Cancer Institute*, **2010**, 102, 1568-83	9.7	81
175	Digital quantification of human eye color highlights genetic association of three new loci. *PLoS Genetics*, **2010**, 6, e1000934	6	135
174	A genome wide linkage scan for dizygotic twinning in 525 families of mothers of dizygotic twins. *Human Reproduction*, **2010**, 25, 1569-80	5.7	27
173	Genetic influence on the age at onset of asthma: a twin study. *Journal of Allergy and Clinical Immunology*, **2010**, 126, 626-30	11.5	50
172	Any proportion of ductal adenocarcinoma in radical prostatectomy specimens predicts extraprostatic extension. *Human Pathology*, **2010**, 41, 281-5	3.7	44
171	IRF4 variants have age-specific effects on nevus count and predispose to melanoma. *American Journal of Human Genetics*, **2010**, 87, 6-16	11	100
170	Mutations at KCNQ1 and an unknown locus cause long QT syndrome in a large Australian family: implications for genetic testing. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 613-21	2.5	3
169	Unexpectedly severe acute radiotherapy side effects are associated with single nucleotide polymorphisms of the melanocortin-1 receptor. *International Journal of Radiation Oncology Biology Physics*, **2010**, 77, 1486-92	4	16
168	Heritability and linkage analysis of appendicitis utilizing age at onset. *Twin Research and Human Genetics*, **2009**, 12, 150-7	2.2	9
167	Exploring the association between severe respiratory syncytial virus infection and asthma: a registry-based twin study. *American Journal of Respiratory and Critical Care Medicine*, **2009**, 179, 1091-7	10.2	146
166	Genetic influences on handedness: data from 25,732 Australian and Dutch twin families. *Neuropsychologia*, **2009**, 47, 330-7	3.2	205

165	Association study of common mitochondrial variants and cognitive ability. *Behavior Genetics*, **2009**, 39, 504-12	3.2	5
164	No evidence that CDKN1B (p27) polymorphisms modify breast cancer risk in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2009**, 115, 307-13	4.4	9
163	Family-based mitochondrial association study of traits related to type 2 diabetes and the metabolic syndrome in adolescents. *Diabetologia*, **2009**, 52, 2359-2368	10.3	3
162	Haplotype sharing excludes orthologous COL4A3, COL4A4 or MYH9 loci in hereditary nephritis in bull terriers. *Animal Genetics*, **2009**, 40, 252-3	2.5	1
161	Linkage confirms canine pkd1 orthologue as a candidate for bull terrier polycystic kidney disease. *Animal Genetics*, **2009**, 40, 543-6	2.5	4
160	Haplotype sharing excludes canine orthologous Filaggrin locus in atopy in West Highland White Terriers. *Animal Genetics*, **2009**, 40, 793-4	2.5	27
159	Analysis of cultured human melanocytes based on polymorphisms within the SLC45A2/MATP, SLC24A5/NCKX5, and OCA2/P loci. *Journal of Investigative Dermatology*, **2009**, 129, 392-405	4.3	84
158	A population-based study of Australian twins with melanoma suggests a strong genetic contribution to liability. *Journal of Investigative Dermatology*, **2009**, 129, 2211-9	4.3	23
157	Genome-wide association study identifies variants at 9p21 and 22q13 associated with development of cutaneous nevi. *Nature Genetics*, **2009**, 41, 915-9	36.3	186
156	Association and interaction analyses of eight genes under asthma linkage peaks. *Allergy: European Journal of Allergy and Clinical Immunology*, **2009**, 64, 1623-8	9.3	13
155	Common variants in the trichohyalin gene are associated with straight hair in Europeans. *American Journal of Human Genetics*, **2009**, 85, 750-5	11	200
154	Polymorphisms in the syntaxin 17 gene are not associated with human cutaneous malignant melanoma. *Melanoma Research*, **2009**, 19, 80-6	3.3	6
153	Linkage and association analysis of spectrophotometrically quantified hair color in Australian adolescents: the effect of OCA2 and HERC2. *Journal of Investigative Dermatology*, **2008**, 128, 2807-14	4.3	19
152	Common sequence variants on 20q11.22 confer melanoma susceptibility. *Nature Genetics*, **2008**, 40, 838-40	36.3	188
151	Spectrophotometric methods for quantifying pigmentation in human hair-influence of MC1R genotype and environment. *Photochemistry and Photobiology*, **2008**, 84, 719-26	3.6	30
150	Examination of chromosome 7p22 candidate genes RBaK, PMS2 and GNA12 in familial hyperaldosteronism type II. *Clinical and Experimental Pharmacology and Physiology*, **2008**, 35, 380-5	3	36
149	Cyclooxygenase-2 gene polymorphisms in an Australian population: association of the -1195G > A promoter polymorphism with mild asthma. *Clinical and Experimental Allergy*, **2008**, 38, 913-20	4.1	19
148	Investigation of the relationship between smoking and appendicitis in Australian twins. *Annals of Epidemiology*, **2008**, 18, 631-6	6.4	35

147	Variation in bone morphogenetic protein 15 is not associated with spontaneous human dizygotic twinning. *Human Reproduction*, **2008**, 23, 2372-9	5.7	29
146	The Queensland Study of Melanoma: environmental and genetic associations (Q-MEGA); study design, baseline characteristics, and repeatability of phenotype and sun exposure measures. *Twin Research and Human Genetics*, **2008**, 11, 183-96	2.2	38
145	A study of diabetes mellitus within a large sample of Australian twins. *Twin Research and Human Genetics*, **2008**, 11, 28-40	2.2	25
144	Skewed X chromosome inactivation and breast and ovarian cancer status: evidence for X-linked modifiers of BRCA1. *Journal of the National Cancer Institute*, **2008**, 100, 1519-29	9.7	26
143	Further evidence for linkage of familial hyperaldosteronism type II at chromosome 7p22 in Italian as well as Australian and South American families. *Journal of Hypertension*, **2008**, 26, 1577-82	1.9	71
142	Progesterone receptor polymorphisms and risk of breast cancer: results from two Australian breast cancer studies. *Breast Cancer Research and Treatment*, **2008**, 109, 91-9	4.4	31
141	Red hair is the null phenotype of MC1R. *Human Mutation*, **2008**, 29, E88-94	4.7	64
140	Calculation of IBD probabilities with dense SNP or sequence data. *Genetic Epidemiology*, **2008**, 32, 513-9	2.6	5
139	A single SNP in an evolutionary conserved region within intron 86 of the HERC2 gene determines human blue-brown eye color. *American Journal of Human Genetics*, **2008**, 82, 424-31	11	275
138	A genome-wide association study identifies novel alleles associated with hair color and skin pigmentation. *PLoS Genetics*, **2008**, 4, e1000074	6	373
137	A haplotype spanning KIAA0319 and TTRAP is associated with normal variation in reading and spelling ability. *Biological Psychiatry*, **2007**, 62, 811-7	7.9	79
136	Evidence of genetic effects on blood lead concentration. *Environmental Health Perspectives*, **2007**, 115, 1224-30	8.4	31
135	Effects of SCA1, MJD, and DPRLA triplet repeat polymorphisms on cognitive phenotypes in a normal population of adolescent twins. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2007**, 144B, 95-100	3.5	2
134	A genome-wide scan for naevus count: linkage to CDKN2A and to other chromosome regions. *European Journal of Human Genetics*, **2007**, 15, 94-102	5.3	67
133	Effect of the BDNF V166M polymorphism on working memory in healthy adolescents. *Genes, Brain and Behavior*, **2007**, 6, 260-8	3.6	43
132	Estimation of variance components for age at menarche in twin families. *Behavior Genetics*, **2007**, 37, 668-77	3.2	55
131	Genomewide scans of red cell indices suggest linkage on chromosome 6q23. *Journal of Medical Genetics*, **2007**, 44, 24-30	5.8	10
130	Receptor function, dominant negative activity and phenotype correlations for MC1R variant alleles. *Human Molecular Genetics*, **2007**, 16, 2249-60	5.6	141

129	Genetic determinants of diabetes are similarly associated with other immune-mediated diseases. *Current Opinion in Allergy and Clinical Immunology*, **2007**, 7, 468-74	3.3	19
128	Significance of minute focus of adenocarcinoma on prostate needle biopsy. *Urology*, **2007**, 70, 299-302	1.6	6
127	A three-single-nucleotide polymorphism haplotype in intron 1 of OCA2 explains most human eye-color variation. *American Journal of Human Genetics*, **2007**, 80, 241-52	11	174
126	Recent human effective population size estimated from linkage disequilibrium. *Genome Research*, **2007**, 17, 520-6	9.7	297
125	The value of relatives with phenotypes but missing genotypes in association studies for quantitative traits. *Genetic Epidemiology*, **2006**, 30, 30-6	2.6	19
124	Rapid screening of 4000 individuals for germ-line variations in the BRAF gene. *Clinical Chemistry*, **2006**, 52, 1675-8	5.5	13
123	Increased DNA methylation at the AXIN1 gene in a monozygotic twin from a pair discordant for a caudal duplication anomaly. *American Journal of Human Genetics*, **2006**, 79, 155-62	11	116
122	HLA and genomewide allele sharing in dizygotic twins. *American Journal of Human Genetics*, **2006**, 79, 1052-8	11	7
121	Novel variants in growth differentiation factor 9 in mothers of dizygotic twins. *Journal of Clinical Endocrinology and Metabolism*, **2006**, 91, 4713-6	5.6	110
120	Handedness in Twins: Joint Analysis of Data From 35 Samples. *Twin Research and Human Genetics*, **2006**, 9, 46-53	2.2	62
119	Variance components analyses of multiple asthma traits in a large sample of Australian families ascertained through a twin proband. *Allergy: European Journal of Allergy and Clinical Immunology*, **2006**, 61, 245-53	9.3	22
118	Risk factors for asthma in young adults: a co-twin control study. *Allergy: European Journal of Allergy and Clinical Immunology*, **2006**, 61, 229-33	9.3	25
117	No evidence for coding region mutations in the retinoblastoma-associated Kruppel-associated box protein gene (RBaK) causing familial hyperaldosteronism type II. *Clinical Endocrinology*, **2006**, 65, 829-31	3.4	12
116	Linkage analysis excludes the involvement of the canine PKD2 homologue in bull terrier polycystic kidney disease. *Animal Genetics*, **2006**, 37, 527-8	2.5	3
115	A simple method to localise pleiotropic susceptibility loci using univariate linkage analyses of correlated traits. *European Journal of Human Genetics*, **2006**, 14, 953-62	5.3	7
114	ADAM33 haplotypes are associated with asthma in a large Australian population. *European Journal of Human Genetics*, **2006**, 14, 1027-36	5.3	53
113	A genome scan for epidermal skin pattern in adolescent twins reveals suggestive linkage on 12p13.31. *Journal of Investigative Dermatology*, **2006**, 126, 277-82	4.3	3
112	Linkage analyses of event-related potential slow wave phenotypes recorded in a working memory task. *Behavior Genetics*, **2006**, 36, 29-44	3.2	7

111	Genome-wide scan of IQ finds significant linkage to a quantitative trait locus on 2q. *Behavior Genetics*, **2006**, 36, 45-55	3.2	35
110	An integrated genetic map for linkage analysis. *Behavior Genetics*, **2006**, 36, 4-6	3.2	28
109	Handedness in twins: joint analysis of data from 35 samples. *Twin Research and Human Genetics*, **2006**, 9, 46-53	2.2	35
108	Genomewide linkage study in 1,176 affected sister pair families identifies a significant susceptibility locus for endometriosis on chromosome 10q26. *American Journal of Human Genetics*, **2005**, 77, 365-76	11	173
107	Robust estimation of experimentwise P values applied to a genome scan of multiple asthma traits identifies a new region of significant linkage on chromosome 20q13. *American Journal of Human Genetics*, **2005**, 77, 1075-85	11	38
106	Systematic review and meta-analysis of the association between {beta}2-adrenoceptor polymorphisms and asthma: a HuGE review. *American Journal of Epidemiology*, **2005**, 162, 201-11	3.8	319
105	The androgen receptor CAG repeat polymorphism and modification of breast cancer risk in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research*, **2005**, 7, R176-83	8.3	39
104	Familial hyperaldosteronism type II is linked to the chromosome 7p22 region but also shows predicted heterogeneity. *Journal of Hypertension*, **2005**, 23, 1477-84	1.9	72
103	New Concepts for Distinguishing the Hidden Patterns of Linkage Disequilibrium Which Underlie Association Between Genotypes and Complex Phenotypes. *Twin Research and Human Genetics*, **2005**, 8, 95-100	2.2	
102	Sex-Limited Genome-Wide Linkage Scan for Body Mass Index in an Unselected Sample of 933 Australian Twin Families. *Twin Research and Human Genetics*, **2005**, 8, 616-632	2.2	37
101	Polymorphisms in the 5-lipoxygenase activating protein (ALOX5AP) gene are not associated with asthma in an Australian population. *Clinical and Experimental Allergy*, **2005**, 35, 332-8	4.1	11
100	A polymorphism in the promoter region of the human interleukin-16 gene is not associated with asthma or atopy in an Australian population. *Clinical and Experimental Allergy*, **2005**, 35, 327-31	4.1	15
99	BRAF polymorphisms and risk of melanocytic neoplasia. *Journal of Investigative Dermatology*, **2005**, 125, 1252-8	4.3	21
98	Genetic and environmental influences on skin pattern deterioration. *Journal of Investigative Dermatology*, **2005**, 125, 1119-29	4.3	37
97	BRAF polymorphisms and the risk of ovarian cancer of low malignant potential. *Gynecologic Oncology*, **2005**, 97, 807-12	4.9	9
96	Genetic time-series analysis identifies a major QTL for in vivo alcohol metabolism not predicted by in vitro studies of structural protein polymorphism at the ADH1B or ADH1C loci. *Behavior Genetics*, **2005**, 35, 509-24	3.2	21
95	Opposite effects of androgen receptor CAG repeat length on increased risk of left-handedness in males and females. *Behavior Genetics*, **2005**, 35, 735-44	3.2	83
94	The EPAS1 gene influences the aerobic-anaerobic contribution in elite endurance athletes. *Human Genetics*, **2005**, 118, 416-23	6.3	45

93	A method for meta-analysis of molecular association studies. *Statistics in Medicine*, **2005**, 24, 1291-306	2.3	519
92	A novel tissue inhibitor of metalloproteinase-1 (TIMP-1) polymorphism associated with asthma in Australian women. *Thorax*, **2005**, 60, 623-8	7.3	41
91	Cyclooxygenase-1 gene polymorphisms in patients with different asthma phenotypes and atopy. *European Respiratory Journal*, **2005**, 26, 249-56	13.6	22
90	The CD14 C-159T polymorphism is not associated with asthma or asthma severity in an Australian adult population. *Thorax*, **2005**, 60, 211-4	7.3	45
89	Association between polymorphisms in the progesterone receptor gene and endometriosis. *Molecular Human Reproduction*, **2005**, 11, 641-7	4.4	30
88	Sex-limited genome-wide linkage scan for body mass index in an unselected sample of 933 Australian twin families. *Twin Research and Human Genetics*, **2005**, 8, 616-32	2.2	23
87	New concepts for distinguishing the hidden patterns of linkage disequilibrium which underlie association between genotypes and complex phenotypes. *Twin Research and Human Genetics*, **2005**, 8, 95-100	2.2	
86	Interactive effects of MC1R and OCA2 on melanoma risk phenotypes. *Human Molecular Genetics*, **2004**, 13, 447-61	5.6	199
85	A genome scan for eye color in 502 twin families: most variation is due to a QTL on chromosome 15q. *Twin Research and Human Genetics*, **2004**, 7, 197-210		88
84	A functional polymorphism in the promoter region of the cyclooxygenase-2 gene is not associated with asthma and atopy in an Australian population. *Clinical and Experimental Allergy*, **2004**, 34, 1714-8	4.1	26
83	Epidermal growth factor gene (EGF) polymorphism and risk of melanocytic neoplasia. *Journal of Investigative Dermatology*, **2004**, 123, 760-2	4.3	39
82	Multivariate QTL linkage analysis suggests a QTL for platelet count on chromosome 19q. *European Journal of Human Genetics*, **2004**, 12, 835-42	5.3	17
81	CCR5-Delta32 mutation is strongly associated with primary sclerosing cholangitis. *Genes and Immunity*, **2004**, 5, 444-50	4.4	51
80	A major quantitative trait locus for CD4-CD8 ratio is located on chromosome 11. *Genes and Immunity*, **2004**, 5, 548-52	4.4	22
79	A simulation study concerning the effect of varying the residual phenotypic correlation on the power of bivariate quantitative trait loci linkage analysis. *Behavior Genetics*, **2004**, 34, 135-41	3.2	14
78	Identification of families with cortical Lewy body disease. *American Journal of Medical Genetics Part A*, **2004**, 128B, 118-22		13
77	Latent class and genetic analysis does not support migraine with aura and migraine without aura as separate entities. *Genetic Epidemiology*, **2004**, 26, 231-44	2.6	99
76	Characterization of two polymorphisms in the leukotriene C4 synthase gene in an Australian population of subjects with mild, moderate, and severe asthma. *Journal of Allergy and Clinical Immunology*, **2004**, 113, 889-95	11.5	56

75	Major quantitative trait locus for eosinophil count is located on chromosome 2q. *Journal of Allergy and Clinical Immunology*, **2004**, 114, 826-30	11.5	25
74	Identification of SQSTM1 mutations in familial Paget© disease in Australian pedigrees. *Bone*, **2004**, 35, 277-82	4.7	54
73	A deletion mutation in GDF9 in sisters with spontaneous DZ twins. *Twin Research and Human Genetics*, **2004**, 7, 548-55		64
72	Dizygotic twinning is not associated with methylenetetrahydrofolate reductase haplotypes. *Human Reproduction*, **2003**, 18, 2460-4	5.7	15
71	Ocular melanoma is not associated with CDKN2A or MC1R variants--a population-based study. *Melanoma Research*, **2003**, 13, 409-13	3.3	9
70	Special twin environments, genetic influences and their effects on the handedness of twins and their siblings. *Twin Research and Human Genetics*, **2003**, 6, 119-30		27
69	Linkage and association analysis of radiation damage repair genes XRCC3 and XRCC5 with nevus density in adolescent twins. *Twin Research and Human Genetics*, **2003**, 6, 315-21		10
68	The role of melanocortin-1 receptor polymorphism in skin cancer risk phenotypes. *Pigment Cell & Melanoma Research*, **2003**, 16, 266-72		90
67	Genetic association and cellular function of MC1R variant alleles in human pigmentation. *Annals of the New York Academy of Sciences*, **2003**, 994, 348-58	6.5	104
66	Evidence for the genetic control of immunoglobulin E reactivity to the allergens of Alternaria alternata. *Clinical and Experimental Allergy*, **2002**, 32, 1316-22	4.1	8
65	The Korean Twin Registry--methods, current stage, and interim results. *Twin Research and Human Genetics*, **2002**, 5, 394-400		2
64	A genomewide search for type 2 diabetes-susceptibility genes in indigenous Australians. *American Journal of Human Genetics*, **2002**, 70, 349-57	11	78
63	Linkage of Paget disease of bone to a novel region on human chromosome 18q23. *American Journal of Human Genetics*, **2002**, 70, 517-25	11	68
62	Melanocortin-1 receptor genotype is a risk factor for basal and squamous cell carcinoma. *Journal of Investigative Dermatology*, **2001**, 116, 224-9	4.3	137
61	Human twinning is not linked to the region of chromosome 4 syntenic with the sheep twinning gene FecB. *American Journal of Medical Genetics Part A*, **2001**, 100, 182-6		17
60	Familial Paget© disease of bone: nonlinkage to the PDB1 and PDB2 loci on chromosomes 6p and 18q in a large pedigree. *Journal of Bone and Mineral Research*, **2001**, 16, 33-8	6.3	26
59	Natural selection and quantitative genetics of life-history traits in Western women: a twin study. *Evolution; International Journal of Organic Evolution*, **2001**, 55, 423-35	3.8	186
58	IBD sharing around the PPARG locus is not increased in dizygotic twins or their mothers. *Nature Genetics*, **2001**, 28, 315	36.3	12

57	Variation in Alcohol Pharmacokinetics as a Risk Factor for Alcohol Dependence. *Alcoholism: Clinical and Experimental Research*, **2001**, 25, 1257-1263	3.7	24
56	NATURAL SELECTION AND QUANTITATIVE GENETICS OF LIFE-HISTORY TRAITS IN WESTERN WOMEN: A TWIN STUDY. *Evolution; International Journal of Organic Evolution*, **2001**, 55, 423	3.8	13
55	MC1R genotype modifies risk of melanoma in families segregating CDKN2A mutations. *American Journal of Human Genetics*, **2001**, 69, 765-73	11	247
54	Mutations in the follicle-stimulating hormone receptor and familial dizygotic twinning. *Lancet, The*, **2001**, 357, 773-4	40	36
53	Applying statistical approaches in the dissection of genes versus environment for asthma and allergic disease. *Current Opinion in Allergy and Clinical Immunology*, **2001**, 1, 431-434	3.3	3
52	Increasing the response rate to a mailed questionnaire by including more stamps on the return envelope: a cotwin control study. *Twin Research and Human Genetics*, **2001**, 4, 71-2		4
51	Dizygotic twinning is not linked to variation at the alpha-inhibin locus on human chromosome 2. *Journal of Clinical Endocrinology and Metabolism*, **2000**, 85, 3391-5	5.6	19
50	Melanocortin-1 receptor polymorphisms and risk of melanoma: is the association explained solely by pigmentation phenotype?. *American Journal of Human Genetics*, **2000**, 66, 176-86	11	424
49	Effects of HFE C282Y and H63D polymorphisms and polygenic background on iron stores in a large community sample of twins. *American Journal of Human Genetics*, **2000**, 66, 1246-58	11	160
48	Dizygotic Twinning Is Not Linked to Variation at the Inhibin Locus on Human Chromosome 2. *Journal of Clinical Endocrinology and Metabolism*, **2000**, 85, 3391-3395	5.6	19
47	CDKN2A variants in a population-based sample of Queensland families with melanoma. *Journal of the National Cancer Institute*, **1999**, 91, 446-52	9.7	165
46	Genetic studies on atopy and helminthiasis. *Allergy: European Journal of Allergy and Clinical Immunology*, **1999**, 54, 1120-1	9.3	
45	No common major gene for apolipoprotein A-I and HDL3-C levels: evidence from bivariate segregation analysis. *Genetic Epidemiology*, **1999**, 16, 54-68	2.6	7
44	A major quantitative-trait locus for mole density is linked to the familial melanoma gene CDKN2A: a maximum-likelihood combined linkage and association analysis in twins and their sibs. *American Journal of Human Genetics*, **1999**, 65, 483-92	11	193
43	Dense mapping of chromosome 12q13.12-q23.3 and linkage to asthma and atopy. *Journal of Allergy and Clinical Immunology*, **1999**, 104, 485-91	11.5	74
42	Dating the origin of the CCR5-Delta32 AIDS-resistance allele by the coalescence of haplotypes. *American Journal of Human Genetics*, **1998**, 62, 1507-15	11	428
41	Genetic regulation of Dermatophagoides pteronyssinus-specific IgE responsiveness: a genome-wide multipoint linkage analysis in families recruited through 2 asthmatic sibs. Collaborative Study on the Genetics of Asthma (CSGA). *Journal of Allergy and Clinical Immunology*, **1998**, 102, 436-42	11.5	79
40	Linkage analysis of Dermatophagoides pteronyssinus-specific IgE responsiveness with polymorphic markers on chromosome 6p21 (HLA-D region) in Caucasian families by the transmission/disequilibrium test. Collaborative Study on the Genetics of Asthma (CSGA). *Journal of Allergy and Clinical Immunology*, **1998**, 102, 443-8	11.5	27

39	Genetic influences of chromosomes 5q31-q33 and 11q13 on specific IgE responsiveness to common inhaled allergens among African American families. Collaborative Study on the Genetics of Asthma (CSGA). *Journal of Allergy and Clinical Immunology*, **1998**, 102, 449-53	11.5	62
38	Immunoblotting analysis of twin sera provides evidence for limited genetic control of specific IgE to house dust mite allergens. *Journal of Allergy and Clinical Immunology*, **1998**, 101, 491-7	11.5	18
37	Novel susceptibility gene for late-onset NIDDM is localized to human chromosome 12q. *Diabetes*, **1998**, 47, 1793-6	0.9	69
36	Hazard and density estimation from bivariate censored data. *Journal of Nonparametric Statistics*, **1998**, 10, 67-93	0.7	7
35	Genetic and environmental risk factors for asthma: a cotwin-control study. *American Journal of Respiratory and Critical Care Medicine*, **1998**, 157, 840-5	10.2	67
34	A factor analysis of associations among self-reported immune related symptoms in a large twin sample. *Twin Research and Human Genetics*, **1998**, 1, 71-77		
33	A factor analysis of associations among self-reported immune related symptoms in a large twin sample. *Twin Research and Human Genetics*, **1998**, 1, 71-7		
32	Maternal Cigarette Smoking and Oral Clefts: A Meta-analysis. *Cleft Palate-Craniofacial Journal*, **1997**, 34, 206-210	1.9	82
31	Evidence for linkage of chromosome 12q15-q24.1 markers to high total serum IgE concentrations in children of the German Multicenter Allergy Study. *Genomics*, **1997**, 46, 159-62	4.3	97
30	Is fragile X syndrome a risk factor for dizygotic twinning?. *American Journal of Medical Genetics Part A*, **1997**, 72, 245-6		11
29	Application of transmission disequilibrium tests to nonsyndromic oral clefts: including candidate genes and environmental exposures in the models. *American Journal of Medical Genetics Part A*, **1997**, 73, 337-44		108
28	A comprehensive analysis of complex traits in problem 2A. *Genetic Epidemiology*, **1997**, 14, 815-20	2.6	3
27	Atopic disease and immunologic response. *Science*, **1997**, 276, 17; author reply 18-9	33.3	2
26	Linkage of asthma and total serum IgE concentration to markers on chromosome 12q: evidence from Afro-Caribbean and Caucasian populations. *Genomics*, **1996**, 37, 41-50	4.3	208
25	Atopy in Australia. *Nature Genetics*, **1995**, 10, 260	36.3	14
24	A psychometric evaluation of the Short Interpersonal Reactions Inventory (SIRI) in an Australian twin sample. *Personality and Individual Differences*, **1995**, 18, 307-320	3.3	5
23	Osteoporosis in rheumatoid arthritis. A monozygotic co-twin control study. *Arthritis and Rheumatism*, **1995**, 38, 806-9		36
22	Screening a 2 cM genetic map for allelic association: a simulated oligogenic trait. *Genetic Epidemiology*, **1995**, 12, 595-600	2.6	7

21	Biometrical genetic analysis of the cotwin control design. *Behavior Genetics*, **1994**, 24, 341-4	3.2	6
20	Type A personality in Australian twins. *Behavior Genetics*, **1994**, 24, 469-75	3.2	6
19	Direction of causation: Reply to commentaries. *Genetic Epidemiology*, **1994**, 11, 463-472	2.6	7
18	Inferring the direction of causation in cross-sectional twin data: theoretical and empirical considerations. *Genetic Epidemiology*, **1994**, 11, 483-502	2.6	88
17	Heterogeneity of melanoma risk in families of melanoma patients. *American Journal of Epidemiology*, **1994**, 140, 961-73	3.8	47
16	Psoriasis in Australian twins. *Journal of the American Academy of Dermatology*, **1993**, 29, 428-34	4.5	161
15	Twin studies in medical research. *Lancet, The*, **1993**, 341, 1418-1419	40	17
14	Lower respiratory tract symptoms in Queensland schoolchildren: risk factors for wheeze, cough and diminished ventilatory function. *Thorax*, **1993**, 48, 1021-4	7.3	11
13	Risk factors for atherosclerosis in twins. *Genetic Epidemiology*, **1993**, 10, 557-62	2.6	4
12	Genetic control of the renal clearance of urate: a study of twins. *Annals of the Rheumatic Diseases*, **1992**, 51, 375-7	2.4	63
11	Rheumatoid arthritis in twins: a study of aetiopathogenesis based on the Australian Twin Registry. *Annals of the Rheumatic Diseases*, **1992**, 51, 588-93	2.4	40
10	A population-based study of bronchial asthma in adult twin pairs. *Chest*, **1992**, 102, 654	5.3	10
9	Is the genetics of moliness simply the genetics of sun exposure? A path analysis of nevus counts and risk factors in British twins. *Cytogenetic and Genome Research*, **1992**, 59, 194-6	1.9	6
8	Respiratory symptoms in Queensland schoolchildren: an association between month of birth and respiratory illness. *Clinical and Experimental Allergy*, **1991**, 21, 231-3	4.1	7
7	Informativeness of twin-nuclear family and nuclear family designs for segregation analysis. *Genetic Epidemiology*, **1991**, 8, 231-5	2.6	1
6	Genetics of asthma and hay fever in Australian twins. *The American Review of Respiratory Disease*, **1990**, 142, 1351-8		389
5	Sex-Limited Genome-Wide Linkage Scan for Body Mass Index in an Unselected Sample of 933 Australian Twin Families		3
4	Special Twin Environments, Genetic Influences and their Effects on the Handedness of Twins and their Siblings		5

List of Publications

3	Linkage and Association Analysis of Radiation Damage Repair Genes XRCC3 and XRCC5 with Nevus Density in Adolescent Twins	3
2	A Genome Scan for Eye Color in 502 Twin Families: Most Variation is due to a QTL on Chromosome 15q	4
1	A Deletion Mutation in GDF9 in Sisters with Spontaneous DZ Twins	2