Hywel Williams

List of Publications by Year in Descending Order

114 papers	16,776 citations	55 h-index	116 g-index
116 ext. papers	18,863 ext. citations	10.5 avg, IF	6.76 L-index

#	Paper	IF	Citations
114	A recessive PRDM13 mutation results in congenital hypogonadotropic hypogonadism and cerebellar hypoplasia. *Journal of Clinical Investigation*, **2021**,	15.9	4
113	Low-frequency variation in TP53 has large effects on head circumference and intracranial volume. *Nature Communications*, **2019**, 10, 357	17.4	12
112	Impaired EIF2S3 function associated with a novel phenotype of X-linked hypopituitarism with glucose dysregulation. *EBioMedicine*, **2019**, 42, 470-480	8.8	24
111	Sotos Syndrome Presenting with Neonatal Hyperinsulinaemic Hypoglycaemia, Extensive Thrombosis, and Multisystem Involvement. *Hormone Research in Paediatrics*, **2019**, 92, 64-70	3.3	0
110	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17
109	Mutations in MAGEL2 and L1CAM Are Associated With Congenital Hypopituitarism and Arthrogryposis. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 5737-5750	5.6	5
108	Tumour compartment transcriptomics demonstrates the activation of inflammatory and odontogenic programmes in human adamantinomatous craniopharyngioma and identifies the MAPK/ERK pathway as a novel therapeutic target. *Acta Neuropathologica*, **2018**, 135, 757-777	14.3	64
107	Mutations in TOP3A Cause a Bloom Syndrome-like Disorder. *American Journal of Human Genetics*, **2018**, 103, 221-231	11	35
106	Rapid Paediatric Sequencing (RaPS): comprehensive real-life workflow for rapid diagnosis of critically ill children. *Journal of Medical Genetics*, **2018**, 55, 721-728	5.8	62
105	High prevalence of p.His154Pro mutation causing primary ciliary dyskinesia disrupts protein oligomerisation and is associated with normal diagnostic investigations. *Thorax*, **2018**, 73, 157-166	7.3	37
104	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. *American Journal of Human Genetics*, **2017**, 100, 281-296	11	43
103	X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3. *Nature Communications*, **2017**, 8, 14279	17.4	87
102	An example of the utility of genomic analysis for fast and accurate clinical diagnosis of complex rare phenotypes. *Orphanet Journal of Rare Diseases*, **2017**, 12, 24	4.2	8
101	Mutations in SLC25A22: hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. *Journal of Inherited Metabolic Disease*, **2017**, 40, 385-394	5.4	12
100	Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity. *Scientific Reports*, **2017**, 7, 4394	4.9	31
99	STAG3 truncating variant as the cause of primary ovarian insufficiency. *European Journal of Human Genetics*, **2016**, 24, 135-8	5.3	41
98	The use of whole-exome sequencing to disentangle complex phenotypes. *European Journal of Human Genetics*, **2016**, 24, 298-301	5.3	14

97	Opportunities and technical challenges in next-generation sequencing for diagnosis of rare pediatric diseases. *Expert Review of Molecular Diagnostics*, **2016**, 16, 1073-1082	3.8	11
96	Mutation screening of SCN2A in schizophrenia and identification of a novel loss-of-function mutation. *Psychiatric Genetics*, **2016**, 26, 60-5	2.9	33
95	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. *Nature Neuroscience*, **2016**, 19, 571-7	25.5	284
94	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. *American Journal of Human Genetics*, **2015**, 96, 1008-1009	11	78
93	Analysis of exome sequence in 604 trios for recessive genotypes in schizophrenia. *Translational Psychiatry*, **2015**, 5, e607	8.6	25
92	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. *Nature Communications*, **2015**, 6, 7074	17.4	41
91	The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90	50.4	776
90	Improved imputation of low-frequency and rare variants using the UK10K haplotype reference panel. *Nature Communications*, **2015**, 6, 8111	17.4	186
89	Exome sequencing for prenatal diagnosis of fetuses with sonographic abnormalities. *Prenatal Diagnosis*, **2015**, 35, 1010-7	3.2	140
88	Whole-genome sequence-based analysis of thyroid function. *Nature Communications*, **2015**, 6, 5681	17.4	56
87	Variability in working memory performance explained by epistasis vs polygenic scores in the ZNF804A pathway. *JAMA Psychiatry*, **2014**, 71, 778-785	14.5	24
86	De novo mutations in schizophrenia implicate synaptic networks. *Nature*, **2014**, 506, 179-84	50.4	1163
85	A rare variant in APOC3 is associated with plasma triglyceride and VLDL levels in Europeans. *Nature Communications*, **2014**, 5, 4871	17.4	46
84	Mutations in SNX14 cause a distinctive autosomal-recessive cerebellar ataxia and intellectual disability syndrome. *American Journal of Human Genetics*, **2014**, 95, 611-21	11	73
83	Meningococcal encephalitis associated with cerebellar tonsillar herniation and acute cervicomedullary injury. *British Journal of Neurosurgery*, **2013**, 27, 513-5	1	3
82	Schizophrenia two-hit hypothesis in velo-cardio facial syndrome. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2013**, 162B, 177-82	3.5	19
81	Genetic schizophrenia risk variants jointly modulate total brain and white matter volume. *Biological Psychiatry*, **2013**, 73, 525-31	7.9	87
80	Schizophrenia genetic variants are not associated with intelligence. *Psychological Medicine*, **2013**, 43, 2563-70	6.9	34

79	A bias-reducing pathway enrichment analysis of genome-wide association data confirmed association of the MHC region with schizophrenia. *Journal of Medical Genetics*, **2012**, 49, 96-103	5.8	64
78	Absence of de novo point mutations in exons of GRIN2B in a large schizophrenia trio sample. *Schizophrenia Research*, **2012**, 141, 274-6	3.6	4
77	Genome-wide supported psychosis risk variant in ZNF804A gene and impact on cortico-limbic WM integrity in schizophrenia. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 255-62	3.5	29
76	Runs of homozygosity implicate autozygosity as a schizophrenia risk factor. *PLoS Genetics*, **2012**, 8, e1002656	8	91
75	Investigation of the genetic association between quantitative measures of psychosis and schizophrenia: a polygenic risk score analysis. *PLoS ONE*, **2012**, 7, e37852	3.7	53
74	ARVCF genetic influences on neurocognitive and neuroanatomical intermediate phenotypes in Chinese patients with schizophrenia. *Journal of Clinical Psychiatry*, **2012**, 73, 320-6	4.6	13
73	Genome-wide association study of schizophrenia in a Japanese population. *Biological Psychiatry*, **2011**, 69, 472-8	7.9	145
72	Genome-wide association study identifies five new schizophrenia loci. *Nature Genetics*, **2011**, 43, 969-76	36.3	1508
71	Two non-synonymous markers in PTPN21, identified by genome-wide association study data-mining and replication, are associated with schizophrenia. *Schizophrenia Research*, **2011**, 131, 43-51	3.6	19
70	Genetic classification of populations using supervised learning. *PLoS ONE*, **2011**, 6, e14802	3.7	14
69	Steroid sulfatase is a potential modifier of cognition in attention deficit hyperactivity disorder. *Genes, Brain and Behavior*, **2011**, 10, 334-44	3.6	37
68	Fine mapping of ZNF804A and genome-wide significant evidence for its involvement in schizophrenia and bipolar disorder. *Molecular Psychiatry*, **2011**, 16, 429-41	15.1	221
67	Molecular pathways involved in neuronal cell adhesion and membrane scaffolding contribute to schizophrenia and bipolar disorder susceptibility. *Molecular Psychiatry*, **2011**, 16, 286-92	15.1	175
66	GWA study data mining and independent replication identify cardiomyopathy-associated 5 (CMYA5) as a risk gene for schizophrenia. *Molecular Psychiatry*, **2011**, 16, 1117-29	15.1	58
65	Investigation of rare non-synonymous variants at ABCA13 in schizophrenia and bipolar disorder. *Molecular Psychiatry*, **2011**, 16, 790-1	15.1	14
64	Analysis of neurogranin (NRGN) in schizophrenia. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156B, 532-5	3.5	10
63	Association between TCF4 and schizophrenia does not exert its effect by common nonsynonymous variation or by influencing cis-acting regulation of mRNA expression in adult human brain. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156B, 781-4	3.5	15
62	Mutation screening of the 3q29 microdeletion syndrome candidate genes DLG1 and PAK2 in schizophrenia. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156B, 844-9	3.5	22

61	Phenotype evaluation and genomewide linkage study of clinical variables in schizophrenia. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2011**, 156B, 929-40	3.5	13
60	The Machado-Joseph disease-associated mutant form of ataxin-3 regulates parkin ubiquitination and stability. *Human Molecular Genetics*, **2011**, 20, 141-54	5.6	217
59	Evidence for rare and common genetic risk variants for schizophrenia at protein kinase C, alpha. *Molecular Psychiatry*, **2010**, 15, 1101-11	15.1	28
58	Psychosis susceptibility gene ZNF804A and cognitive performance in schizophrenia. *Archives of General Psychiatry*, **2010**, 67, 692-700		120
57	Identification of novel candidate genes for treatment response to risperidone and susceptibility for schizophrenia: integrated analysis among pharmacogenomics, mouse expression, and genetic case-control association approaches. *Biological Psychiatry*, **2010**, 67, 263-9	7.9	78
56	Failure to confirm association between PIK4CA and psychosis in 22q11.2 deletion syndrome. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 980-2	3.5	6
55	No evidence that rare coding variants in ZNF804A confer risk of schizophrenia. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1411-6	3.5	18
54	Accurately assessing the risk of schizophrenia conferred by rare copy-number variation affecting genes with brain function. *PLoS Genetics*, **2010**, 6, e1001097	6	118
53	New findings from genetic association studies of schizophrenia. *Journal of Human Genetics*, **2009**, 54, 9-14	4.3	27
52	Influence of NOS1 on verbal intelligence and working memory in both patients with schizophrenia and healthy control subjects. *Archives of General Psychiatry*, **2009**, 66, 1045-54		42
51	Identifying relationships among genomic disease regions: predicting genes at pathogenic SNP associations and rare deletions. *PLoS Genetics*, **2009**, 5, e1000534	6	337
50	Schizophrenia genetics: new insights from new approaches. *British Medical Bulletin*, **2009**, 91, 61-74	5.4	58
49	Common polygenic variation contributes to risk of schizophrenia and bipolar disorder. *Nature*, **2009** , 460, 748-52	50.4	3568
48	Analysis of 10 independent samples provides evidence for association between schizophrenia and a SNP flanking fibroblast growth factor receptor 2. *Molecular Psychiatry*, **2009**, 14, 30-6	15.1	62
47	Schizophrenia genetics: advancing on two fronts. *Current Opinion in Genetics and Development*, **2009**, 19, 266-70	4.9	64
46	An examination of MUTED as a schizophrenia susceptibility gene. *Schizophrenia Research*, **2009**, 107, 110-1	3.6	5
45	Rare chromosomal deletions and duplications increase risk of schizophrenia. *Nature*, **2008**, 455, 237-41	50.4	1251
44	Identification of loci associated with schizophrenia by genome-wide association and follow-up. *Nature Genetics*, **2008**, 40, 1053-5	36.3	877

43	Molecular dissection of NRG1-ERBB4 signaling implicates PTPRZ1 as a potential schizophrenia susceptibility gene. *Molecular Psychiatry*, **2008**, 13, 162-72	15.1	63
42	Strong evidence that GNB1L is associated with schizophrenia. *Human Molecular Genetics*, **2008**, 17, 555-66	5.6	58
41	Analysis of copy number variation using quantitative interspecies competitive PCR. *Nucleic Acids Research*, **2008**, 36, e112	20.1	9
40	Association analysis of AKT1 and schizophrenia in a UK case control sample. *Schizophrenia Research*, **2007**, 93, 58-65	3.6	51
39	Association analysis of the glial cell line-derived neurotrophic factor (GDNF) gene in schizophrenia. *Schizophrenia Research*, **2007**, 97, 271-6	3.6	15
38	Genotype effects of CHRNA7, CNR1 and COMT in schizophrenia: interactions with tobacco and cannabis use. *British Journal of Psychiatry*, **2007**, 191, 402-7	5.4	144
37	Is COMT a susceptibility gene for schizophrenia?. *Schizophrenia Bulletin*, **2007**, 33, 635-41	1.3	143
36	Evidence that interaction between neuregulin 1 and its receptor erbB4 increases susceptibility to schizophrenia. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2006**, 141B, 96-101	3.5	143
35	Convergent evidence that oligodendrocyte lineage transcription factor 2 (OLIG2) and interacting genes influence susceptibility to schizophrenia. *Proceedings of the National Academy of Sciences of the United States of America*, **2006**, 103, 12469-74	11.5	101
34	Analysis of ProDH, COMT and ZDHHC8 risk variants does not support individual or interactive effects on schizophrenia susceptibility. *Schizophrenia Research*, **2006**, 87, 21-7	3.6	36
33	An update on the genetics of schizophrenia. *Current Opinion in Psychiatry*, **2006**, 19, 158-64	4.9	113
32	Variation at the DAOA/G30 locus influences susceptibility to major mood episodes but not psychosis in schizophrenia and bipolar disorder. *Archives of General Psychiatry*, **2006**, 63, 366-73		124
31	Genome wide significant linkage in schizophrenia conditioning on occurrence of depressive episodes. *Journal of Medical Genetics*, **2006**, 43, 563-7	5.8	15
30	No association between the putative functional ZDHHC8 single nucleotide polymorphism rs175174 and schizophrenia in large European samples. *Biological Psychiatry*, **2005**, 58, 78-80	7.9	38
29	A family based study implicates solute carrier family 1-member 3 (SLC1A3) gene in attention-deficit/hyperactivity disorder. *Biological Psychiatry*, **2005**, 57, 1461-6	7.9	23
28	No evidence for association between polymorphisms in GRM3 and schizophrenia. *BMC Psychiatry*, **2005**, 5, 23	4.2	41
27	Genome screen for loci influencing age at onset and rate of decline in late onset Alzheimer's disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2005**, 135B, 24-32	3.5	56
26	A family based study of catechol-O-methyltransferase (COMT) and attention deficit hyperactivity disorder (ADHD). *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2005**, 133B, 64-7	3.5	19

25	No association between schizophrenia and polymorphisms in COMT in two large samples. *American Journal of Psychiatry*, **2005**, 162, 1736-8	11.9	70
24	Genomewide linkage scan in schizoaffective disorder: significant evidence for linkage at 1q42 close to DISC1, and suggestive evidence at 22q11 and 19p13. *Archives of General Psychiatry*, **2005**, 62, 1081-8		164
23	Identification in 2 independent samples of a novel schizophrenia risk haplotype of the dystrobrevin binding protein gene (DTNBP1). *Archives of General Psychiatry*, **2004**, 61, 336-44		155
22	Identification and analysis of the promoter region of the human hyaluronan synthase 2 gene. *Journal of Biological Chemistry*, **2004**, 279, 20576-81	5.4	28
21	Multicenter linkage study of schizophrenia loci on chromosome 22q. *Molecular Psychiatry*, **2004**, 9, 784-95	15.1	51
20	Support for RGS4 as a susceptibility gene for schizophrenia. *Biological Psychiatry*, **2004**, 55, 192-5	7.9	125
19	Chromosome 22q11 deletions, velo-cardio-facial syndrome and early-onset psychosis. Molecular genetic study. *British Journal of Psychiatry*, **2003**, 183, 409-13	5.4	58
18	Association between PRODH and schizophrenia is not confirmed. *Molecular Psychiatry*, **2003**, 8, 644-5	15.1	47
17	Support for genetic variation in neuregulin 1 and susceptibility to schizophrenia. *Molecular Psychiatry*, **2003**, 8, 485-7	15.1	206
16	A haplotype implicated in schizophrenia susceptibility is associated with reduced COMT expression in human brain. *American Journal of Human Genetics*, **2003**, 73, 152-61	11	301
15	A systematic genomewide linkage study in 353 sib pairs with schizophrenia. *American Journal of Human Genetics*, **2003**, 73, 1355-67	11	111
14	Full genome screen for Alzheimer disease: stage II analysis. *American Journal of Medical Genetics Part A*, **2002**, 114, 235-44		179
13	Universal, robust, highly quantitative SNP allele frequency measurement in DNA pools. *Human Genetics*, **2002**, 110, 471-8	6.3	152
12	Screening the human protocadherin 8 (PCDH8) gene in schizophrenia. *Genes, Brain and Behavior*, **2002**, 1, 187-91	3.6	22
11	Characterisation, mutation detection, and association analysis of alternative promoters and 5TUTRs of the human dopamine D3 receptor gene in schizophrenia. *Molecular Psychiatry*, **2002**, 7, 493-502	15.1	30
10	Determination of the genomic structure and mutation screening in schizophrenic individuals for five subunits of the N-methyl-D-aspartate glutamate receptor. *Molecular Psychiatry*, **2002**, 7, 508-14	15.1	94
9	Mutation screening and LD mapping in the VCFS deleted region of chromosome 22q11 in schizophrenia using a novel DNA pooling approach. *Molecular Psychiatry*, **2002**, 7, 1092-100	15.1	18
8	No major schizophrenia locus detected on chromosome 1q in a large multicenter sample. *Science*, **2002**, 296, 739-41	33.3	73

7	A genomewide linkage study of age at onset in schizophrenia. *American Journal of Medical Genetics Part A*, **2001**, 105, 439-45		55
6	Association analysis of NOTCH4 loci in schizophrenia using family and population-based controls. *Nature Genetics*, **2001**, 28, 126-8	36.3	57
5	Mutation screening of the KCNN3 gene reveals a rare frameshift mutation. *Molecular Psychiatry*, **2001**, 6, 259-60	15.1	30
4	Localization of the gene for distal hereditary motor neuronopathy VII (dHMN-VII) to chromosome 2q14. *American Journal of Human Genetics*, **2001**, 68, 1270-6	11	63
3	A full genome scan for late onset Alzheimer's disease. *Human Molecular Genetics*, **1999**, 8, 237-45	5.6	279
2	Variation in DCP1, encoding ACE, is associated with susceptibility to Alzheimer disease. *Nature Genetics*, **1999**, 21, 71-2	36.3	236
1	No evidence for allelic association between schizophrenia and a functional variant of the human dopamine beta-hydroxylase gene (DBH). *American Journal of Medical Genetics Part A*, **1999**, 88, 557-9		19