

# Ryan K C Yuen

## List of Publications by Citations

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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.

70 papers	7,768 citations	34 h-index	88 g-index
92 ext. papers	10,369 ext. citations	11.7 avg, IF	5.21 L-index

#	Paper	IF	Citations
70	Synaptic, transcriptional and chromatin genes disrupted in autism. <i>Nature</i> , <b>2014</b> , 515, 209-15	50.4	1581
69	The Database of Genomic Variants: a curated collection of structural variation in the human genome. <i>Nucleic Acids Research</i> , <b>2014</b> , 42, D986-92	20.1	764
68	RNA splicing. The human splicing code reveals new insights into the genetic determinants of disease. <i>Science</i> , <b>2015</b> , 347, 1254806	33.3	748
67	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , <b>2020</b> , 180, 568-584.e23	56.2	578
66	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. <i>Nature Neuroscience</i> , <b>2017</b> , 20, 602-611	25.5	427
65	Whole-genome sequencing of quartet families with autism spectrum disorder. <i>Nature Medicine</i> , <b>2015</b> , 21, 185-91	50.5	353
64	Detection of clinically relevant genetic variants in autism spectrum disorder by whole-genome sequencing. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 249-63	11	345
63	Molecular Diagnostic Yield of Chromosomal Microarray Analysis and Whole-Exome Sequencing in Children With Autism Spectrum Disorder. <i>JAMA - Journal of the American Medical Association</i> , <b>2015</b> , 314, 895-903	27.4	241
62	Whole Genome Sequencing Expands Diagnostic Utility and Improves Clinical Management in Pediatric Medicine. <i>Npj Genomic Medicine</i> , <b>2016</b> , 1,	6.2	208
61	DNA methylation profiling of human placentas reveals promoter hypomethylation of multiple genes in early-onset preeclampsia. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 1006-12	5.3	164
60	Widespread DNA hypomethylation at gene enhancer regions in placentas associated with early-onset pre-eclampsia. <i>Molecular Human Reproduction</i> , <b>2013</b> , 19, 697-708	4.4	150
59	Evidence for widespread changes in promoter methylation profile in human placenta in response to increasing gestational age and environmental/stochastic factors. <i>BMC Genomics</i> , <b>2011</b> , 12, 529	4.5	142
58	Genome-wide characteristics of mutations in autism. <i>Npj Genomic Medicine</i> , <b>2016</b> , 1, 160271-1602710	6.2	126
57	The autism sequencing consortium: large-scale, high-throughput sequencing in autism spectrum disorders. <i>Neuron</i> , <b>2012</b> , 76, 1052-6	13.9	124
56	Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. <i>Nature Genetics</i> , <b>2014</b> , 46, 742-7	36.3	121
55	Hypermethylation of RASSF1A in human and rhesus placentas. <i>American Journal of Pathology</i> , <b>2007</b> , 170, 941-50	5.8	118
54	Disruption of the ASTN2/TRIM32 locus at 9q33.1 is a risk factor in males for autism spectrum disorders, ADHD and other neurodevelopmental phenotypes. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 2752-68	5.6	104

53	A Comprehensive Workflow for Read Depth-Based Identification of Copy-Number Variation from Whole-Genome Sequence Data. <i>American Journal of Human Genetics</i> , <b>2018</b> , 102, 142-155	11	97
52	Compound heterozygous mutations in the noncoding RNU4ATAC cause Roifman Syndrome by disrupting minor intron splicing. <i>Nature Communications</i> , <b>2015</b> , 6, 8718	17.4	74
51	Altered TAOK2 activity causes autism-related neurodevelopmental and cognitive abnormalities through RhoA signaling. <i>Molecular Psychiatry</i> , <b>2019</b> , 24, 1329-1350	15.1	70
50	Hypoxia alters the epigenetic profile in cultured human placental trophoblasts. <i>Epigenetics</i> , <b>2013</b> , 8, 192-202	5.7	69
49	Evaluating DNA methylation and gene expression variability in the human term placenta. <i>Placenta</i> , <b>2010</b> , 31, 1070-7	3.4	68
48	Length of Uninterrupted CAG, Independent of Polyglutamine Size, Results in Increased Somatic Instability, Hastening Onset of Huntington Disease. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 1116-1126	11.1	67
47	Genome-wide mapping of imprinted differentially methylated regions by DNA methylation profiling of human placentas from triploidies. <i>Epigenetics and Chromatin</i> , <b>2011</b> , 4, 10	5.8	62
46	Genome-wide detection of tandem DNA repeats that are expanded in autism. <i>Nature</i> , <b>2020</b> , 586, 80-86	50.4	58
45	Complete Disruption of Autism-Susceptibility Genes by Gene Editing Predominantly Reduces Functional Connectivity of Isogenic Human Neurons. <i>Stem Cell Reports</i> , <b>2018</b> , 11, 1211-1225	8	58
44	A large data resource of genomic copy number variation across neurodevelopmental disorders. <i>Npj Genomic Medicine</i> , <b>2019</b> , 4, 26	6.2	54
43	Human placental-specific epipolymorphism and its association with adverse pregnancy outcomes. <i>PLoS ONE</i> , <b>2009</b> , 4, e7389	3.7	54
42	Extensive epigenetic reprogramming in human somatic tissues between fetus and adult. <i>Epigenetics and Chromatin</i> , <b>2011</b> , 4, 7	5.8	51
41	or human iPSC-derived neurons from individuals with autism develop hyperactive neuronal networks. <i>ELife</i> , <b>2019</b> , 8,	8.9	41
40	CNTN6 mutations are risk factors for abnormal auditory sensory perception in autism spectrum disorders. <i>Molecular Psychiatry</i> , <b>2017</b> , 22, 625-633	15.1	40
39	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. <i>Cmaj</i> , <b>2018</b> , 190, E126-E136	3.5	37
38	Review: A high capacity of the human placenta for genetic and epigenetic variation: implications for assessing pregnancy outcome. <i>Placenta</i> , <b>2011</b> , 32 Suppl 2, S136-41	3.4	37
37	Patterns of placental development evaluated by X chromosome inactivation profiling provide a basis to evaluate the origin of epigenetic variation. <i>Human Reproduction</i> , <b>2012</b> , 27, 1745-53	5.7	34
36	Synaptic Dysfunction in Human Neurons With Autism-Associated Deletions in PTCHD1-AS. <i>Biological Psychiatry</i> , <b>2020</b> , 87, 139-149	7.9	32

35	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion in. <i>New England Journal of Medicine</i> , <b>2019</b> , 380, 1433-1441	59.2	31
34	The utility of quantitative methylation assays at imprinted genes for the diagnosis of fetal and placental disorders. <i>Clinical Genetics</i> , <b>2011</b> , 79, 169-75	4	30
33	A framework for an evidence-based gene list relevant to autism spectrum disorder. <i>Nature Reviews Genetics</i> , <b>2020</b> , 21, 367-376	30.1	30
32	ExpansionHunter Denovo: a computational method for locating known and novel repeat expansions in short-read sequencing data. <i>Genome Biology</i> , <b>2020</b> , 21, 102	18.3	29
31	Germline and somatic mutations in with diverse neurodevelopmental phenotypes. <i>Neurology: Genetics</i> , <b>2017</b> , 3, e199	3.8	28
30	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. <i>Scientific Reports</i> , <b>2016</b> , 6, 28663	4.9	26
29	Performance of high-throughput sequencing for the discovery of genetic variation across the complete size spectrum. <i>G3: Genes, Genomes, Genetics</i> , <b>2014</b> , 4, 63-5	3.2	26
28	Development of a high-resolution Y-chromosome microarray for improved male infertility diagnosis. <i>Fertility and Sterility</i> , <b>2014</b> , 101, 1079-1085.e3	4.8	24
27	Mutations in in individuals with intellectual disability, autism spectrum disorder, and macrocephaly. <i>Molecular Autism</i> , <b>2017</b> , 8, 59	6.5	22
26	DIXDC1 Phosphorylation and Control of Dendritic Morphology Are Impaired by Rare Genetic Variants. <i>Cell Reports</i> , <b>2016</b> , 17, 1892-1904	10.6	21
25	Large-scale exome sequencing study implicates both developmental and functional changes in the neurobiology of autism		21
24	Variable phenotype expression in a family segregating microdeletions of the and autism spectrum disorder susceptibility genes. <i>Npj Genomic Medicine</i> , <b>2017</b> , 2,	6.2	17
23	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. <i>Brain</i> , <b>2019</b> , 142, 2617-2630	11.2	17
22	Using extended pedigrees to identify novel autism spectrum disorder (ASD) candidate genes. <i>Human Genetics</i> , <b>2015</b> , 134, 191-201	6.3	16
21	Genome and Transcriptome Assembly of the Canadian Beaver (). <i>G3: Genes, Genomes, Genetics</i> , <b>2017</b> , 7, 755-773	3.2	13
20	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. <i>JAMA Network Open</i> , <b>2020</b> , 3, e2018109	10.4	13
19	Predictive impact of rare genomic copy number variations in siblings of individuals with autism spectrum disorders. <i>Nature Communications</i> , <b>2019</b> , 10, 5519	17.4	13
18	Expanding the neurodevelopmental phenotypes of individuals with de novo variants. <i>Npj Genomic Medicine</i> , <b>2019</b> , 4, 9	6.2	10

17	A genome-wide linkage study of autism spectrum disorder and the broad autism phenotype in extended pedigrees. <i>Journal of Neurodevelopmental Disorders</i> , <b>2018</b> , 10, 20	4.6	10
16	Genome-wide DNA methylation identifies trophoblast invasion-related genes: Claudin-4 and Fucosyltransferase IV control mobility via altering matrix metalloproteinase activity. <i>Molecular Human Reproduction</i> , <b>2015</b> , 21, 452-65	4.4	9
15	Genes and Pathways Implicated in Tetralogy of Fallot Revealed by Ultra-Rare Variant Burden Analysis in 231 Genome Sequences. <i>Frontiers in Genetics</i> , <b>2020</b> , 11, 957	4.5	8
14	Genetics of Epileptic Networks: From Focal to Generalized Genetic Epilepsies. <i>Current Neurology and Neuroscience Reports</i> , <b>2020</b> , 20, 46	6.6	6
13	The Genetics of Autism Spectrum Disorders <b>2019</b> , 112-128		6
12	Genomic imbalances in the placenta are associated with poor fetal growth. <i>Molecular Medicine</i> , <b>2021</b> , 27, 3	6.2	5
11	Genome sequencing broadens the range of contributing variants with clinical implications in schizophrenia. <i>Translational Psychiatry</i> , <b>2021</b> , 11, 84	8.6	5
10	Length of uninterrupted CAG repeats, independent of polyglutamine size, results in increased somatic instability and hastened age of onset in Huntington disease		4
9	Segregating patterns of copy number variations in extended autism spectrum disorder (ASD) pedigrees. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2020</b> , 183, 268-276	3.5	3
8	Rare and low frequency genomic variants impacting neuronal functions modify the Dup7q11.23 phenotype. <i>Orphanet Journal of Rare Diseases</i> , <b>2021</b> , 16, 6	4.2	3
7	Genome sequencing identifies rare tandem repeat expansions and copy number variants in Lennox-Gastaut syndrome. <i>Brain Communications</i> , <b>2021</b> , 3, fcab207	4.5	2
6	ISDN2014_0253: High resolution genomic analyses of a clinically defined autism spectrum disorder cohort. <i>International Journal of Developmental Neuroscience</i> , <b>2015</b> , 47, 76-76	2.7	1
5	Sleep phenotype of individuals with autism spectrum disorder bearing mutations in the PER2 circadian rhythm gene. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 1120-1130	2.5	1
4	PP065. dNK and dNK-CM mediated alterations of DNA methylation in extravillous cytotrophoblasts (EVTS). <i>Pregnancy Hypertension</i> , <b>2012</b> , 2, 277	2.6	0
3	FAN1 exo- not endo-nuclease pausing on disease-associated slipped-DNA repeats: A mechanism of repeat instability. <i>Cell Reports</i> , <b>2021</b> , 37, 110078	10.6	0
2	Schizophrenia Risk Mediated by microRNA Target Genes Overlapped by Genome-Wide Rare Copy Number Variation in 22q11.2 Deletion Syndrome.. <i>Frontiers in Genetics</i> , <b>2022</b> , 13, 812183	4.5	0
1	Genome-Wide Mapping <b>2013</b> , 71-109		