John F Robinson

List of Publications by Year in Descending Order

22 papers	418 citations	10 h-index	20 g-index
22 ext. papers	537 ext. citations	4.8 avg, IF	2.92 L-index

#	Paper	IF	Citations
22	Enrichment of loss-of-function and copy number variants in ventricular cardiomyopathy genes in lone atrial fibrillation. *Europace*, **2021**, 23, 844-850	3.9	2
21	Simplifying Detection of Copy-Number Variations in Maturity-Onset Diabetes of the Young. *Canadian Journal of Diabetes*, **2021**, 45, 71-77	2.1	
20	Association of apolipoprotein E variation with cognitive impairment across multiple neurodegenerative diagnoses. *Neurobiology of Aging*, **2021**, 105, 378.e1-378.e9	5.6	1
19	Six years experience with LipidSeq: clinical and research learnings from a hybrid, targeted sequencing panel for dyslipidemias. *BMC Medical Genomics*, **2020**, 13, 23	3.7	23
18	Partial deletions: rare copy-number variants contributing towards severe hypertriglyceridemia. *Journal of Lipid Research*, **2019**, 60, 1953-1958	6.3	10
17	Genetic and epigenetic study of an Alzheimer disease family with monozygotic triplets. *Brain*, **2019**, 142, 3375-3381	11.2	6
16	Genetic Variation in the Ontario Neurodegenerative Disease Research Initiative. *Canadian Journal of Neurological Sciences*, **2019**, 46, 491-498	1	5
15	Clinical Utility and Practical Considerations of a Coronary Artery Disease Genetic Risk Score. *CJC Open*, **2019**, 1, 69-75	2	3
14	Targeted sequencing reveals expanded genetic diversity of human transfer RNAs. *RNA Biology*, **2019**, 16, 1574-1585	4.8	9
13	Bioinformatic detection of copy number variation in HNF4A causing maturity onset diabetes of the young. *Clinical Genetics*, **2019**, 96, 376-377	4	2
12	Copy Number Variation in GCK in Patients With Maturity-Onset Diabetes of the Young. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 3428-3436	5.6	7
11	Ankyrin-B dysfunction predisposes to arrhythmogenic cardiomyopathy and is amenable to therapy. *Journal of Clinical Investigation*, **2019**, 129, 3171-3184	15.9	23
10	Targeted Next-generation Sequencing and Bioinformatics Pipeline to Evaluate Genetic Determinants of Constitutional Disease. *Journal of Visualized Experiments*, **2018**,	1.6	14
9	Whole-Gene Duplication of PCSK9 as a Novel Genetic Mechanism for Severe Familial Hypercholesterolemia. *Canadian Journal of Cardiology*, **2018**, 34, 1316-1324	3.8	23
8	Polygenic determinants in extremes of high-density lipoprotein cholesterol. *Journal of Lipid Research*, **2017**, 58, 2162-2170	6.3	33
7	Polygenic Versus Monogenic Causes of Hypercholesterolemia Ascertained Clinically. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2016**, 36, 2439-2445	9.4	130
6	Proprietary Considerations in the Use of Cardiovascular Genetic Data. *Canadian Journal of Cardiology*, **2016**, 32, 1297-1299	3.8	0

5	Whole-genome sequencing in French Canadians from Quebec. *Human Genetics*, **2016**, 135, 1213-1221	6.3	13
4	The ONDRISeq panel: custom-designed next-generation sequencing of genes related to neurodegeneration. *Npj Genomic Medicine*, **2016**, 1, 16032	6.2	17
3	Exome sequencing identifies NFS1 deficiency in a novel Fe-S cluster disease, infantile mitochondrial complex II/III deficiency. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 73-80	2.3	41
2	A novel LIPE nonsense mutation found using exome sequencing in siblings with late-onset familial partial lipodystrophy. *Canadian Journal of Cardiology*, **2014**, 30, 1649-54	3.8	50
1	The Ontario Neurodegenerative Disease Research Initiative		6