

# Nathan Oliver Stitzel

## List of Publications by Year in descending order

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Version: 2024-02-01

62  
papers

9,683  
citations

87888

38  
h-index

106344

65  
g-index

71  
all docs

71  
docs citations

71  
times ranked

16694  
citing authors

#	ARTICLE	IF	CITATIONS
1	Loss-of-Function Mutations in <i>APOC3</i> , Triglycerides, and Coronary Disease. <i>New England Journal of Medicine</i> , 2014, 371, 22-31.	27.0	936
2	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. <i>Nature</i> , 2015, 518, 102-106.	27.8	581
3	Genetic risk, coronary heart disease events, and the clinical benefit of statin therapy: an analysis of primary and secondary prevention trials. <i>Lancet</i> , The, 2015, 385, 2264-2271.	13.7	564
4	A general approach to single-nucleotide polymorphism discovery. <i>Nature Genetics</i> , 1999, 23, 452-456.	21.4	550
5	Exome-wide association study of plasma lipids in >300,000 individuals. <i>Nature Genetics</i> , 2017, 49, 1758-1766.	21.4	470
6	Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. <i>Science</i> , 2016, 351, 1166-1171.	12.6	438
7	Coding Variation in <i>ANGPTL4</i> , <i>LPL</i> and <i>SVEP1</i> and the Risk of Coronary Disease. <i>New England Journal of Medicine</i> , 2016, 374, 1134-1144.	27.0	427
8	Polygenic Risk Score Identifies Subgroup With Higher Burden of Atherosclerosis and Greater Relative Benefit From Statin Therapy in the Primary Prevention Setting. <i>Circulation</i> , 2017, 135, 2091-2101.	1.6	403
9	Clinical Genetic Testing for Familial Hypercholesterolemia. <i>Journal of the American College of Cardiology</i> , 2018, 72, 662-680.	2.8	387
10	Inactivating Mutations in <i>NPC1L1</i> and Protection from Coronary Heart Disease. <i>New England Journal of Medicine</i> , 2014, 371, 2072-2082.	27.0	386
11	Distribution and Medical Impact of Loss-of-Function Variants in the Finnish Founder Population. <i>PLoS Genetics</i> , 2014, 10, e1004494.	3.5	351
12	ANGPTL3 Deficiency and Protection Against Coronary Artery Disease. <i>Journal of the American College of Cardiology</i> , 2017, 69, 2054-2063.	2.8	348
13	Exome sequencing and the genetic basis of complex traits. <i>Nature Genetics</i> , 2012, 44, 623-630.	21.4	340
14	Association of Low-Frequency and Rare Coding-Sequence Variants with Blood Lipids and Coronary Heart Disease in 56,000 Whites and Blacks. <i>American Journal of Human Genetics</i> , 2014, 94, 223-232.	6.2	287
15	Meta-analysis identifies common and rare variants influencing blood pressure and overlapping with metabolic trait loci. <i>Nature Genetics</i> , 2016, 48, 1162-1170.	21.4	223
16	Systematic Evaluation of Pleiotropy Identifies 6 Further Loci Associated With Coronary Artery Disease. <i>Journal of the American College of Cardiology</i> , 2017, 69, 823-836.	2.8	214
17	Mapping and characterization of structural variation in 17,795 human genomes. <i>Nature</i> , 2020, 583, 83-89.	27.8	194
18	Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause a subtype of Distal Arthrogyriposis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 4667-4672.	7.1	193

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19	Phenotypic Characterization of Genetically Lowered Human Lipoprotein(a) Levels. <i>Journal of the American College of Cardiology</i> , 2016, 68, 2761-2772.	2.8	186
20	Exome sequencing of Finnish isolates enhances rare-variant association power. <i>Nature</i> , 2019, 572, 323-328.	27.8	161
21	Association of Rare and Common Variation in the Lipoprotein Lipase Gene With Coronary Artery Disease. <i>JAMA - Journal of the American Medical Association</i> , 2017, 317, 937.	7.4	148
22	Loss of function mutation in <i>LOX</i> causes thoracic aortic aneurysm and dissection in humans. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 8759-8764.	7.1	144
23	Genetic architecture of human plasma lipidome and its link to cardiovascular disease. <i>Nature Communications</i> , 2019, 10, 4329.	12.8	120
24	Computational and statistical approaches to analyzing variants identified by exome sequencing. <i>Genome Biology</i> , 2011, 12, 227.	9.6	116
25	Description of a Large Family with Autosomal Dominant Hypercholesterolemia Associated with the <i>APOE</i> p.Leu167del Mutation. <i>Human Mutation</i> , 2013, 34, 83-87.	2.5	103
26	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. <i>Genome Biology</i> , 2014, 15, R53.	9.6	101
27	Exome Sequencing and Directed Clinical Phenotyping Diagnose Cholesterol Ester Storage Disease Presenting as Autosomal Recessive Hypercholesterolemia. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2013, 33, 2909-2914.	2.4	87
28	Phenotypic Consequences of a Genetic Predisposition to Enhanced Nitric Oxide Signaling. <i>Circulation</i> , 2018, 137, 222-232.	1.6	87
29	High-protein diets increase cardiovascular risk by activating macrophage mTOR to suppress mitophagy. <i>Nature Metabolism</i> , 2020, 2, 110-125.	11.9	85
30	<i>APOE</i> p.Leu167del mutation in familial hypercholesterolemia. <i>Atherosclerosis</i> , 2013, 231, 218-222.	0.8	84
31	topoSNP: a topographic database of non-synonymous single nucleotide polymorphisms with and without known disease association. <i>Nucleic Acids Research</i> , 2004, 32, 520D-522.	14.5	81
32	Clinical characteristics and plasma lipids in subjects with familial combined hypolipidemia: a pooled analysis. <i>Journal of Lipid Research</i> , 2013, 54, 3481-3490.	4.2	76
33	Structural Location of Disease-associated Single-nucleotide Polymorphisms. <i>Journal of Molecular Biology</i> , 2003, 327, 1021-1030.	4.2	74
34	Identification of Medically Actionable Secondary Findings in the 1000 Genomes. <i>PLoS ONE</i> , 2015, 10, e0135193.	2.5	74
35	Genome-wide association studies of metabolites in Finnish men identify disease-relevant loci. <i>Nature Communications</i> , 2022, 13, 1644.	12.8	63
36	Multiethnic Exome-Wide Association Study of Subclinical Atherosclerosis. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 511-520.	5.1	54

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37	Exome Sequencing in Suspected Monogenic Dyslipidemias. <i>Circulation: Cardiovascular Genetics</i> , 2015, 8, 343-350.	5.1	45
38	Prognostic value of diastolic filling parameters derived using a novel image processing technique in patients ≥70 years of age with congestive heart failure. <i>American Journal of Cardiology</i> , 1999, 84, 82-86.	1.6	39
39	Association of exome sequences with plasma C-reactive protein levels in >9000 participants. <i>Human Molecular Genetics</i> , 2015, 24, 559-571.	2.9	36
40	Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. <i>American Journal of Human Genetics</i> , 2020, 107, 46-59.	6.2	30
41	SVEP1 is a human coronary artery disease locus that promotes atherosclerosis. <i>Science Translational Medicine</i> , 2021, 13, .	12.4	28
42	Leveraging human genetics to guide drug target discovery. <i>Trends in Cardiovascular Medicine</i> , 2017, 27, 352-359.	4.9	26
43	New Sequencing technologies help revealing unexpected mutations in Autosomal Dominant Hypercholesterolemia. <i>Scientific Reports</i> , 2018, 8, 1943.	3.3	25
44	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. <i>Npj Genomic Medicine</i> , 2018, 3, 21.	3.8	24
45	Coronary Artery Disease Risk and Lipidomic Profiles Are Similar in Hyperlipidemias With Family History and Population-Ascertained Hyperlipidemias. <i>Journal of the American Heart Association</i> , 2019, 8, e012415.	3.7	24
46	Genetic invalidation of Lp-PLA2 as a therapeutic target: Large-scale study of five functional Lp-PLA2-lowering alleles. <i>European Journal of Preventive Cardiology</i> , 2017, 24, 492-504.	1.8	22
47	Roadmap for a precision-medicine initiative in the Nordic region. <i>Nature Genetics</i> , 2019, 51, 924-930.	21.4	22
48	Association of structural variation with cardiometabolic traits in Finns. <i>American Journal of Human Genetics</i> , 2021, 108, 583-596.	6.2	22
49	Common and Rare Genetic Variation in <i>CCR2</i> , <i>CCR5</i> , or <i>CX3CR1</i> and Risk of Atherosclerotic Coronary Heart Disease and Glucometabolic Traits. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 250-258.	5.1	20
50	Diagnosis and management of adult hereditary cardio-neuromuscular disorders: A model for the multidisciplinary care of complex genetic disorders. <i>Trends in Cardiovascular Medicine</i> , 2017, 27, 51-58.	4.9	19
51	Membrane-Associated and Secreted Genes in Breast Cancer. <i>Cancer Research</i> , 2004, 64, 8682-8687.	0.9	17
52	Genetic association studies in cardiovascular diseases: Do we have enough power?. <i>Trends in Cardiovascular Medicine</i> , 2017, 27, 397-404.	4.9	17
53	Genetics of the extracellular matrix in aortic aneurysmal diseases. <i>Matrix Biology</i> , 2018, 71-72, 128-143.	3.6	17
54	A Clinical Approach to Inherited Premature Coronary Artery Disease. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 558-564.	5.1	16

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55	Emerging Targets for Cardiovascular Disease Prevention in Diabetes. Trends in Molecular Medicine, 2020, 26, 744-757.	6.7	15
56	Functional Characterization of LIPA (Lysosomal Acid Lipase) Variants Associated With Coronary Artery Disease. Arteriosclerosis, Thrombosis, and Vascular Biology, 2019, 39, 2480-2491.	2.4	13
57	Human genetic insights into lipoproteins and risk of cardiometabolic disease. Current Opinion in Lipidology, 2017, 28, 113-119.	2.7	12
58	Intracellular retention of mutant lysyl oxidase leads to aortic dilation in response to increased hemodynamic stress. JCI Insight, 2019, 4, .	5.0	12
59	Inherited <i>CHST11/MIR3922</i> deletion is associated with a novel recessive syndrome presenting with skeletal malformation and malignant lymphoproliferative disease. Molecular Genetics & Genomic Medicine, 2015, 3, 413-423.	1.2	11
60	Mitochondrial genome copy number measured by DNA sequencing in human blood is strongly associated with metabolic traits via cell-type composition differences. Human Genomics, 2021, 15, 34.	2.9	7
61	Capitalizing on Insights from Human Genetics to Identify Novel Therapeutic Targets for Coronary Artery Disease. Annual Review of Medicine, 2019, 70, 19-32.	12.2	6
62	Reply. Journal of the American College of Cardiology, 2017, 70, 2099-2100.	2.8	1