

Michael Nothnagel

List of Publications by Year in descending order

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125
papers

8,707
citations

46918

47
h-index

45213

90
g-index

134
all docs

134
docs citations

134
times ranked

15607
citing authors

#	ARTICLE	IF	CITATIONS
1	Sequence variants in IL10, ARPC2 and multiple other loci contribute to ulcerative colitis susceptibility. <i>Nature Genetics</i> , 2008, 40, 1319-1323.	9.4	534
2	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. <i>Nature Genetics</i> , 2009, 41, 160-162.	9.4	511
3	Correlation between Genetic and Geographic Structure in Europe. <i>Current Biology</i> , 2008, 18, 1241-1248.	1.8	449
4	A genome-wide association study confirms PNPLA3 and identifies TM6SF2 and MBOAT7 as risk loci for alcohol-related cirrhosis. <i>Nature Genetics</i> , 2015, 47, 1443-1448.	9.4	435
5	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. <i>Nature Genetics</i> , 2013, 45, 1067-1072.	9.4	391
6	Genome-Wide Association Analysis in Primary Sclerosing Cholangitis. <i>Gastroenterology</i> , 2010, 138, 1102-1111.	0.6	325
7	Combined Analysis of Genome-wide Association Studies for Crohn Disease and Psoriasis Identifies Seven Shared Susceptibility Loci. <i>American Journal of Human Genetics</i> , 2012, 90, 636-647.	2.6	290
8	Power and Sample Size Calculations for Case-Control Genetic Association Tests when Errors Are Present: Application to Single Nucleotide Polymorphisms. <i>Human Heredity</i> , 2002, 54, 22-33.	0.4	269
9	Genome-wide association study identifies ANXA11 as a new susceptibility locus for sarcoidosis. <i>Nature Genetics</i> , 2008, 40, 1103-1106.	9.4	239
10	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. <i>Nature Genetics</i> , 2015, 47, 393-399.	9.4	224
11	A global analysis of Y-chromosomal haplotype diversity for 23 STR loci. <i>Forensic Science International: Genetics</i> , 2014, 12, 12-23.	1.6	214
12	Wnt Signaling and Dupuytren's Disease. <i>New England Journal of Medicine</i> , 2011, 365, 307-317.	13.9	201
13	Genome-wide search for novel human uORFs and N-terminal protein extensions using ribosomal footprinting. <i>Genome Research</i> , 2012, 22, 2208-2218.	2.4	198
14	Identification of a Shared Genetic Susceptibility Locus for Coronary Heart Disease and Periodontitis. <i>PLoS Genetics</i> , 2009, 5, e1000378.	1.5	189
15	A genome-wide association study confirms APOE as the major gene influencing survival in long-lived individuals. <i>Mechanisms of Ageing and Development</i> , 2011, 132, 324-330.	2.2	184
16	A genome-wide association study identifies GLT6D1 as a susceptibility locus for periodontitis. <i>Human Molecular Genetics</i> , 2010, 19, 553-562.	1.4	176
17	Benign infantile seizures and paroxysmal dyskinesia caused by an <i>SCN8A</i> mutation. <i>Annals of Neurology</i> , 2016, 79, 428-436.	2.8	159
18	Association Between Variants of PRDM1 and NDP52 and Crohn's Disease, Based on Exome Sequencing and Functional Studies. <i>Gastroenterology</i> , 2013, 145, 339-347.	0.6	149

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19	Genomic and geographic distribution of SNP-defined runs of homozygosity in Europeans. <i>Human Molecular Genetics</i> , 2010, 19, 2927-2935.	1.4	146
20	A comprehensive evaluation of SNP genotype imputation. <i>Human Genetics</i> , 2009, 125, 163-171.	1.8	139
21	Polymorphisms in the glial glutamate transporter <i>SLC1A2</i> are associated with essential tremor. <i>Neurology</i> , 2012, 79, 243-248.	1.5	111
22	Polymorphisms in NACHT-LRR (<i>NLR</i>) genes in atopic dermatitis. <i>Experimental Dermatology</i> , 2007, 16, 692-698.	1.4	108
23	Heterozygous carriage of the alpha1-antitrypsin Pi*Z variant increases the risk to develop liver cirrhosis. <i>Gut</i> , 2019, 68, 1099-1107.	6.1	100
24	Loci From a Genome-Wide Analysis of Bilirubin Levels Are Associated With Gallstone Risk and Composition. <i>Gastroenterology</i> , 2010, 139, 1942-1951.e2.	0.6	96
25	Continent-Wide Decoupling of Y-Chromosomal Genetic Variation from Language and Geography in Native South Americans. <i>PLoS Genetics</i> , 2013, 9, e1003460.	1.5	89
26	Genome-Wide Association Analysis in Sarcoidosis and Crohn's Disease Unravels a Common Susceptibility Locus on 10p12.2. <i>Gastroenterology</i> , 2008, 135, 1207-1215.	0.6	85
27	Entropy as a Measure for Linkage Disequilibrium over Multilocus Haplotype Blocks. <i>Human Heredity</i> , 2002, 54, 186-198.	0.4	81
28	Expanding the genetic architecture of nicotine dependence and its shared genetics with multiple traits. <i>Nature Communications</i> , 2020, 11, 5562.	5.8	80
29	The Wegener's granulomatosis quantitative trait locus on chromosome 6p21.3 as characterised by tagSNP genotyping. <i>Annals of the Rheumatic Diseases</i> , 2008, 67, 972-979.	0.5	79
30	Performance of in silico prediction tools for the classification of rare BRCA1/2 missense variants in clinical diagnostics. <i>BMC Medical Genomics</i> , 2018, 11, 35.	0.7	78
31	Validation of reported genetic risk factors for periodontitis in a large-scale replication study. <i>Journal of Clinical Periodontology</i> , 2013, 40, 563-572.	2.3	74
32	Genetic and functional identification of the likely causative variant for cholesterol gallstone disease at the <i>ABCG5/8</i> lithogenic locus. <i>Hepatology</i> , 2013, 57, 2407-2417.	3.6	74
33	Identification and characterization of two functional variants in the human longevity gene <i>FOXO3</i> . <i>Nature Communications</i> , 2017, 8, 2063.	5.8	69
34	A Genome-wide Association Study of Dupuytren Disease Reveals 17 Additional Variants Implicated in Fibrosis. <i>American Journal of Human Genetics</i> , 2017, 101, 417-427.	2.6	67
35	Ancient DNA study reveals HLA susceptibility locus for leprosy in medieval Europeans. <i>Nature Communications</i> , 2018, 9, 1569.	5.8	67
36	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. <i>Lancet Neurology</i> , The, 2018, 17, 699-708.	4.9	67

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37	The Effect of Single-Nucleotide Polymorphism Marker Selection on Patterns of Haplotype Blocks and Haplotype Frequency Estimates. <i>American Journal of Human Genetics</i> , 2005, 77, 988-998.	2.6	66
38	A 3â€² UTR transition within DEFEB1 is associated with chronic and aggressive periodontitis. <i>Genes and Immunity</i> , 2010, 11, 45-54.	2.2	62
39	CDKN2BAS is associated with periodontitis in different European populations and is activated by bacterial infection. <i>Journal of Medical Genetics</i> , 2011, 48, 38-47.	1.5	61
40	Schizophrenia risk polymorphisms in the <i>TCF4</i> gene interact with smoking in the modulation of auditory sensory gating. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 6271-6276.	3.3	60
41	Collaborative genetic mapping of 12 forensic short tandem repeat (STR) loci on the human X chromosome. <i>Forensic Science International: Genetics</i> , 2012, 6, 778-784.	1.6	60
42	A genome-wide association study reveals evidence of association with sarcoidosis at 6p12.1. <i>European Respiratory Journal</i> , 2011, 38, 1127-1135.	3.1	58
43	Meta-analysis identifies novel risk loci and yields systematic insights into the biology of male-pattern baldness. <i>Nature Communications</i> , 2017, 8, 14694.	5.8	58
44	Association of inflammatory bowel disease risk loci with sarcoidosis, and its acute and chronic subphenotypes. <i>European Respiratory Journal</i> , 2011, 37, 610-616.	3.1	53
45	A Novel Sarcoidosis Risk Locus for Europeans on Chromosome 11q13.1. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2012, 186, 877-885.	2.5	51
46	Rare variants in Î³-aminobutyric acid type A receptor genes in rolandic epilepsy and related syndromes. <i>Annals of Neurology</i> , 2015, 77, 972-986.	2.8	51
47	Evaluation of genome-wide loci of iron metabolism in hereditary hemochromatosis identifies PCSK7 as a host risk factor of liver cirrhosis. <i>Human Molecular Genetics</i> , 2014, 23, 3883-3890.	1.4	50
48	Potentials and limits of pairwise kinship analysis using autosomal short tandem repeat loci. <i>International Journal of Legal Medicine</i> , 2010, 124, 205-215.	1.2	48
49	<i>LINGO1</i> polymorphisms are associated with essential tremor in Europeans. <i>Movement Disorders</i> , 2010, 25, 717-723.	2.2	47
50	Polymorphisms in the interleukin-1 (IL1) gene cluster are not associated with aggressive periodontitis in a large Caucasian population. <i>Genomics</i> , 2008, 92, 309-315.	1.3	45
51	<i>COX-2</i> Is Associated with Periodontitis in Europeans. <i>Journal of Dental Research</i> , 2010, 89, 384-388.	2.5	43
52	Genome-wide association analysis reveals 12q13.3â€“q14.1 as new risk locus for sarcoidosis. <i>European Respiratory Journal</i> , 2013, 41, 888-900.	3.1	43
53	X Chromosomal Variation Is Associated with Slow Progression to AIDS in HIV-1-Infected Women. <i>American Journal of Human Genetics</i> , 2009, 85, 228-239.	2.6	41
54	Genome-wide investigation of geneâ€“environment interactions in colorectal cancer. <i>Human Genetics</i> , 2013, 132, 219-231.	1.8	38

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55	Technology-specific error signatures in the 1000 Genomes Project data. <i>Human Genetics</i> , 2011, 130, 505-516.	1.8	37
56	Association of toll-interacting protein gene polymorphisms with atopic dermatitis. <i>BMC Dermatology</i> , 2007, 7, 3.	2.1	34
57	Distinct genetic variation and heterogeneity of the Iranian population. <i>PLoS Genetics</i> , 2019, 15, e1008385.	1.5	34
58	s-ICAM-1 and s-VCAM-1 in healthy men are strongly associated with traits of the metabolic syndrome, becoming evident in the postprandial response to a lipid-rich meal. <i>Lipids in Health and Disease</i> , 2008, 7, 32.	1.2	33
59	Statistical inference of allelic imbalance from transcriptome data. <i>Human Mutation</i> , 2011, 32, 98-106.	1.1	33
60	Increased Probability of Co-Occurrence of Two Rare Diseases in Consanguineous Families and Resolution of a Complex Phenotype by Next Generation Sequencing. <i>PLoS ONE</i> , 2016, 11, e0146040.	1.1	32
61	Role of the toll-like receptor 4 polymorphism Asp299Gly in longevity and myocardial infarction in German men. <i>Mechanisms of Ageing and Development</i> , 2007, 128, 409-411.	2.2	30
62	GABAA receptor- and GABA transporter polymorphisms and risk for essential tremor. <i>European Journal of Neurology</i> , 2011, 18, 1098-1100.	1.7	28
63	Haplotypes of IL-12R β 1 impact on the clinical phenotype of hidradenitis suppurativa. <i>Cytokine</i> , 2013, 62, 297-301.	1.4	26
64	Meta-Analysis of Genome-Wide Association Studies and Network Analysis-Based Integration with Gene Expression Data Identify New Suggestive Loci and Unravel a Wnt-Centric Network Associated with Dupuytren's Disease. <i>PLoS ONE</i> , 2016, 11, e0158101.	1.1	26
65	The association of fatty acid-binding protein 2 A54T polymorphism with postprandial lipemia depends on promoter variability. <i>Metabolism: Clinical and Experimental</i> , 2007, 56, 723-731.	1.5	24
66	Postprandial plasma adiponectin decreases after glucose and high fat meal and is independently associated with postprandial triacylglycerols but not with Δ 11388 promoter polymorphism. <i>British Journal of Nutrition</i> , 2008, 99, 76-82.	1.2	24
67	True colors: A literature review on the spatial distribution of eye and hair pigmentation. <i>Forensic Science International: Genetics</i> , 2019, 39, 109-118.	1.6	24
68	Prognostic relevance of gastric cancer staging by endoscopic ultrasound. <i>Surgical Endoscopy and Other Interventional Techniques</i> , 2013, 27, 1124-1129.	1.3	23
69	Clinical and experimental evidence suggest a link between KIF7 and C5orf42-related ciliopathies through Sonic Hedgehog signaling. <i>European Journal of Human Genetics</i> , 2018, 26, 197-209.	1.4	23
70	A case-only study of gene-environment interaction between genetic susceptibility variants in NOD2 and cigarette smoking in Crohn's disease aetiology. <i>BMC Medical Genetics</i> , 2012, 13, 14.	2.1	22
71	Exome-wide analysis of mutational burden in patients with typical and atypical Rolandic epilepsy. <i>European Journal of Human Genetics</i> , 2018, 26, 258-264.	1.4	22
72	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. <i>PLoS ONE</i> , 2016, 11, e0150426.	1.1	22

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73	The Slavic NBN Founder Mutation: A Role for Reproductive Fitness?. PLoS ONE, 2016, 11, e0167984.	1.1	21
74	Genetic mapping of 15 human X chromosomal forensic short tandem repeat (STR) loci by means of multi-core parallelization. Forensic Science International: Genetics, 2016, 25, 39-44.	1.6	21
75	The minor allele of the PPAR β Pro12Ala polymorphism is associated with lower postprandial TAG and insulin levels in non-obese healthy men. British Journal of Nutrition, 2007, 97, 847-854.	1.2	20
76	A Genome-Wide Linkage Analysis in 181 German Sarcoidosis Families Using Clustered Biallelic Markers. Chest, 2010, 138, 151-157.	0.4	20
77	Serum metabolomic profiling highlights pathways associated with liver fat content in a general population sample. European Journal of Clinical Nutrition, 2017, 71, 995-1001.	1.3	20
78	Current software for genotype imputation. Human Genomics, 2009, 3, 371-80.	1.4	19
79	Shannon's equivocation for forensic Y-STR marker selection. Forensic Science International: Genetics, 2015, 16, 216-225.	1.6	18
80	A Rare Variant Nonparametric Linkage Method for Nuclear and Extended Pedigrees with Application to Late-Onset Alzheimer Disease via WGS Data. American Journal of Human Genetics, 2019, 105, 822-835.	2.6	16
81	Efficacy assessment of SNP sets for genome-wide disease association studies. Nucleic Acids Research, 2007, 35, e113-e113.	6.5	15
82	Female-specific association of C-C chemokine receptor 5 gene polymorphisms with LÃ¶fgrenÃ¶s syndrome. Journal of Molecular Medicine, 2008, 86, 553-561.	1.7	15
83	Variation in genes of the epidermal differentiation complex in German atopic dermatitis patients. International Journal of Immunogenetics, 2009, 36, 217-222.	0.8	15
84	Effects of Pleistocene climatic fluctuations on the phylogeography, demography and population structure of a high-elevation snake species, <i>Thermophilis baileyi</i> , on the Tibetan Plateau. Journal of Biogeography, 2014, 41, 2162-2172.	1.4	14
85	Association of postprandial and fasting triglycerides with traits of the metabolic syndrome in the Metabolic Intervention Cohort Kiel. European Journal of Endocrinology, 2010, 162, 719-727.	1.9	13
86	Missense variants in NOX1 and p22phox in a case of very-early-onset inflammatory bowel disease are functionally linked to NOD2. Journal of Physical Education and Sports Management, 2019, 5, a002428.	0.5	13
87	Role of NOD2/CARD15 in coronary heart disease. BMC Genetics, 2007, 8, 76.	2.7	12
88	Association studies of the copy-number variable γ -defensin cluster on 8p23.1 in adenocarcinoma and chronic pancreatitis. BMC Research Notes, 2012, 5, 629.	0.6	12
89	Association screen for atopic dermatitis candidate gene regions using microsatellite markers in pooled DNA samples. International Journal of Immunogenetics, 2006, 33, 401-409.	0.8	11
90	<i>LINGO1</i> is not associated with Parkinson's disease in German patients. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1173-1178.	1.1	11

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91	Depletion of potential A2M risk haplotype for Alzheimer's disease in long-lived individuals. <i>European Journal of Human Genetics</i> , 2010, 18, 59-61.	1.4	11
92	SFRS10â€”A Splicing Factor Gene Reduced in Human Obesity?. <i>Cell Metabolism</i> , 2012, 15, 265-266.	7.2	11
93	Evaluation of potential effects of Plastin 3 overexpression and low-dose SMN-antisense oligonucleotides on putative biomarkers in spinal muscular atrophy mice. <i>PLoS ONE</i> , 2018, 13, e0203398.	1.1	11
94	Evaluation of supervised machine-learning methods for predicting appearance traits from DNA. <i>Forensic Science International: Genetics</i> , 2021, 53, 102507.	1.6	11
95	Family-Based Benchmarking of Copy Number Variation Detection Software. <i>PLoS ONE</i> , 2015, 10, e0133465.	1.1	9
96	An evaluation of the genetic-matched pair study design using genome-wide SNP data from the European population. <i>European Journal of Human Genetics</i> , 2009, 17, 967-975.	1.4	8
97	Pipeline for Large-Scale Microdroplet Bisulfite PCR-Based Sequencing Allows the Tracking of Hepitype Evolution in Tumors. <i>PLoS ONE</i> , 2011, 6, e21332.	1.1	8
98	Common genetic risk variants of <i>TLR2</i> are not associated with periodontitis in large European case-control populations. <i>Journal of Clinical Periodontology</i> , 2012, 39, 315-322.	2.3	8
99	Guideline-based and bioinformatic reassessment of lesion-associated gene and variant pathogenicity in focal human epilepsies. <i>Epilepsia</i> , 2018, 59, 2145-2152.	2.6	8
100	Diagnosing Fatty Liver Disease: A Comparative Evaluation of Metabolic Markers, Phenotypes, Genotypes and Established Biomarkers. <i>PLoS ONE</i> , 2013, 8, e76813.	1.1	8
101	The impact of correlations between pigmentation phenotypes and underlying genotypes on genetic prediction of pigmentation traits. <i>Forensic Science International: Genetics</i> , 2021, 50, 102395.	1.6	7
102	Distinct gene-set burden patterns underlie common generalized and focal epilepsies. <i>EBioMedicine</i> , 2021, 72, 103588.	2.7	7
103	Approaches to the genetics of cardiovascular disease through genetic field work. <i>Kidney International</i> , 1998, 53, 1449-1454.	2.6	6
104	NOD1 gene polymorphisms in relation to aggressive periodontitis. <i>Innate Immunity</i> , 2009, 15, 225-232.	1.1	5
105	Exome sequencing utility in defining the genetic landscape of hearing loss and novel gene discovery in Iran. <i>Clinical Genetics</i> , 2021, 100, 59-78.	1.0	4
106	Mutations Causing Complex Disease May under Certain Circumstances Be Protective in an Epidemiological Sense. <i>PLoS ONE</i> , 2015, 10, e0132150.	1.1	4
107	Comparative Assessment of the Association Information Captured by SNP Tagging. <i>Human Heredity</i> , 2007, 64, 27-34.	0.4	3
108	The effect of FABP2 promoter haplotype on response to a diet with medium-chain triacylglycerols. <i>Genes and Nutrition</i> , 2012, 7, 437-445.	1.2	3

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109	The exhaustive genomic scan approach, with an application to rare-variant association analysis. <i>European Journal of Human Genetics</i> , 2020, 28, 1283-1291.	1.4	3
110	Testing the impact of trait prevalence priors in Bayesian-based genetic prediction modeling of human appearance traits. <i>Forensic Science International: Genetics</i> , 2021, 50, 102412.	1.6	3
111	Statistical gene mapping of traits in humans—hypertension as a complex trait: Is it amenable to genetic analysis?. <i>Seminars in Nephrology</i> , 2002, 22, 105-114.	0.6	3
112	Metabolic Signature of Electrosurgical Liver Dissection. <i>PLoS ONE</i> , 2013, 8, e72022.	1.1	2
113	Hypotheses in genome-wide association scans. <i>European Journal of Human Genetics</i> , 2008, 16, 1174-1175.	1.4	1
114	CoNCoS: Copy number estimation in cancer with controlled support. <i>Journal of Bioinformatics and Computational Biology</i> , 2015, 13, 1550027.	0.3	1
115	Pathway-induced allelic spectra of diseases in the presence of strong genetic effects. <i>Human Genetics</i> , 2018, 137, 215-230.	1.8	1
116	Unsupported claim of significant discrimination between monozygotic twins from multiple pairs based on three age-related DNA methylation markers. <i>Forensic Science International: Genetics</i> , 2019, 39, e1-e2.	1.6	1
117	Special issue on “Genetic epidemiology of complex diseases: impact of population history and modelling assumptions”. <i>Human Genetics</i> , 2020, 139, 1-3.	1.8	1
118	A Y-chromosomal survey of Ecuador’s multi-ethnic population reveals new insights into the tri-partite population structure and supports an early Holocene age of the rare Native American founder lineage C3-MPB373. <i>Forensic Science International: Genetics</i> , 2021, 51, 102427.	1.6	1
119	Analysis of single nucleotide polymorphisms in chronic beryllium disease. <i>Respiratory Research</i> , 2021, 22, 107.	1.4	1
120	What Makes a Hot-Spring Habitat “Hot” for the Hot-Spring Snake: Distributional Data and Niche Modelling for the Genus <i>Thermophilis</i> (Serpentes, Colubridae). <i>Diversity</i> , 2021, 13, 325.	0.7	1
121	A Critical Evaluation of Analytic Aspects of Gene Expression Profiling in Lymphoid Leukemias with Broad Applications to Cancer Genomics. <i>AIMS Medical Science</i> , 2016, 3, 248-271.	0.2	1
122	Modellvorstellungen zur Genetik multifaktorieller Krankheiten. <i>Medizinische Genetik</i> , 2007, 19, 295-299.	0.1	0
123	Securing the use of existing sample collections for future human genetic research. <i>European Journal of Human Genetics</i> , 2017, 25, 522-529.	1.4	0
124	Towards a fine-scale picture of European genetic diversity. <i>European Journal of Human Genetics</i> , 2020, 28, 851-852.	1.4	0
125	Benchmarking of univariate pleiotropy detection methods applied to epilepsy. <i>Human Mutation</i> , 2022, 43, 1314-1332.	1.1	0