

# Michael Nothnagel

## 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.

126  
papers

6,824  
citations

41  
h-index

81  
g-index

134  
ext. papers

7,962  
ext. citations

8.1  
avg. IF

4.83  
L-index

#	Paper	IF	Citations
126	Sequence variants in IL10, ARPC2 and multiple other loci contribute to ulcerative colitis susceptibility. <i>Nature Genetics</i> , <b>2008</b> , 40, 1319-23	36.3	468
125	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. <i>Nature Genetics</i> , <b>2009</b> , 41, 160-236.3	36.3	454
124	Correlation between genetic and geographic structure in Europe. <i>Current Biology</i> , <b>2008</b> , 18, 1241-8	6.3	382
123	A genome-wide association study confirms PNPLA3 and identifies TM6SF2 and MBOAT7 as risk loci for alcohol-related cirrhosis. <i>Nature Genetics</i> , <b>2015</b> , 47, 1443-8	36.3	303
122	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. <i>Nature Genetics</i> , <b>2013</b> , 45, 1067-72	36.3	301
121	Genome-wide association analysis in primary sclerosing cholangitis. <i>Gastroenterology</i> , <b>2010</b> , 138, 1102-1113.3	13.3	255
120	Power and sample size calculations for case-control genetic association tests when errors are present: application to single nucleotide polymorphisms. <i>Human Heredity</i> , <b>2002</b> , 54, 22-33	1.1	242
119	Combined analysis of genome-wide association studies for Crohn disease and psoriasis identifies seven shared susceptibility loci. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 636-47	11	224
118	Genome-wide association study identifies ANXA11 as a new susceptibility locus for sarcoidosis. <i>Nature Genetics</i> , <b>2008</b> , 40, 1103-6	36.3	175
117	A global analysis of Y-chromosomal haplotype diversity for 23 STR loci. <i>Forensic Science International: Genetics</i> , <b>2014</b> , 12, 12-23	4.3	171
116	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. <i>Nature Genetics</i> , <b>2015</b> , 47, 393-399	36.3	162
115	A genome-wide association study confirms APOE as the major gene influencing survival in long-lived individuals. <i>Mechanisms of Ageing and Development</i> , <b>2011</b> , 132, 324-30	5.6	162
114	Identification of a shared genetic susceptibility locus for coronary heart disease and periodontitis. <i>PLoS Genetics</i> , <b>2009</b> , 5, e1000378	6	162
113	Wnt signaling and Dupuytren's disease. <i>New England Journal of Medicine</i> , <b>2011</b> , 365, 307-17	59.2	153
112	Genome-wide search for novel human uORFs and N-terminal protein extensions using ribosomal footprinting. <i>Genome Research</i> , <b>2012</b> , 22, 2208-18	9.7	150
111	A genome-wide association study identifies GLT6D1 as a susceptibility locus for periodontitis. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 553-62	5.6	150
110	Association between variants of PRDM1 and NDP52 and Crohn's disease, based on exome sequencing and functional studies. <i>Gastroenterology</i> , <b>2013</b> , 145, 339-47	13.3	125

109	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. <i>Annals of Neurology</i> , <b>2016</b> , 79, 428-36	9.4	124
108	A comprehensive evaluation of SNP genotype imputation. <i>Human Genetics</i> , <b>2009</b> , 125, 163-71	6.3	123
107	Genomic and geographic distribution of SNP-defined runs of homozygosity in Europeans. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 2927-35	5.6	99
106	Polymorphisms in the glial glutamate transporter SLC1A2 are associated with essential tremor. <i>Neurology</i> , <b>2012</b> , 79, 243-8	6.5	94
105	Polymorphisms in NACHT-LRR (NLR) genes in atopic dermatitis. <i>Experimental Dermatology</i> , <b>2007</b> , 16, 692-8	4	94
104	Continent-wide decoupling of Y-chromosomal genetic variation from language and geography in native South Americans. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003460	6	75
103	Loci from a genome-wide analysis of bilirubin levels are associated with gallstone risk and composition. <i>Gastroenterology</i> , <b>2010</b> , 139, 1942-1951.e2	13.3	74
102	Validation of reported genetic risk factors for periodontitis in a large-scale replication study. <i>Journal of Clinical Periodontology</i> , <b>2013</b> , 40, 563-72	7.7	68
101	Entropy as a measure for linkage disequilibrium over multilocus haplotype blocks. <i>Human Heredity</i> , <b>2002</b> , 54, 186-98	1.1	68
100	Genome-wide association analysis in sarcoidosis and Crohn's disease unravels a common susceptibility locus on 10p12.2. <i>Gastroenterology</i> , <b>2008</b> , 135, 1207-15	13.3	66
99	Heterozygous carriage of the alpha1-antitrypsin Pi*Z variant increases the risk to develop liver cirrhosis. <i>Gut</i> , <b>2019</b> , 68, 1099-1107	19.2	62
98	Genetic and functional identification of the likely causative variant for cholesterol gallstone disease at the ABCG5/8 lithogenic locus. <i>Hepatology</i> , <b>2013</b> , 57, 2407-17	11.2	61
97	The Wegener's granulomatosis quantitative trait locus on chromosome 6p21.3 as characterised by tagSNP genotyping. <i>Annals of the Rheumatic Diseases</i> , <b>2008</b> , 67, 972-9	2.4	59
96	CDKN2BAS is associated with periodontitis in different European populations and is activated by bacterial infection. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 38-47	5.8	57
95	A 3' UTR transition within DEFB1 is associated with chronic and aggressive periodontitis. <i>Genes and Immunity</i> , <b>2010</b> , 11, 45-54	4.4	56
94	Revisiting the male genetic landscape of China: a multi-center study of almost 38,000 Y-STR haplotypes. <i>Human Genetics</i> , <b>2017</b> , 136, 485-497	6.3	51
93	The effect of single-nucleotide polymorphism marker selection on patterns of haplotype blocks and haplotype frequency estimates. <i>American Journal of Human Genetics</i> , <b>2005</b> , 77, 988-98	11	51
92	Collaborative genetic mapping of 12 forensic short tandem repeat (STR) loci on the human X chromosome. <i>Forensic Science International: Genetics</i> , <b>2012</b> , 6, 778-84	4.3	49

91	Identification and characterization of two functional variants in the human longevity gene FOXO3. <i>Nature Communications</i> , <b>2017</b> , 8, 2063	17.4	46
90	Schizophrenia risk polymorphisms in the TCF4 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> , <b>2012</b> , 109, 6271-6	11.5	45
89	Rare coding variants in genes encoding GABA receptors in genetic generalised epilepsies: an exome-based case-control study. <i>Lancet Neurology</i> , <b>2018</b> , 17, 699-708	24.1	44
88	Performance of in silico prediction tools for the classification of rare BRCA1/2 missense variants in clinical diagnostics. <i>BMC Medical Genomics</i> , <b>2018</b> , 11, 35	3.7	44
87	LINGO1 polymorphisms are associated with essential tremor in Europeans. <i>Movement Disorders</i> , <b>2010</b> , 25, 717-23	7	44
86	Ancient DNA study reveals HLA susceptibility locus for leprosy in medieval Europeans. <i>Nature Communications</i> , <b>2018</b> , 9, 1569	17.4	41
85	Potentials and limits of pairwise kinship analysis using autosomal short tandem repeat loci. <i>International Journal of Legal Medicine</i> , <b>2010</b> , 124, 205-15	3.1	41
84	Rare variants in GABA <sub>A</sub> receptor genes in rolandic epilepsy and related syndromes. <i>Annals of Neurology</i> , <b>2015</b> , 77, 972-86	9.4	40
83	A genome-wide association study reveals evidence of association with sarcoidosis at 6p12.1. <i>European Respiratory Journal</i> , <b>2011</b> , 38, 1127-35	13.6	40
82	Association of inflammatory bowel disease risk loci with sarcoidosis, and its acute and chronic subphenotypes. <i>European Respiratory Journal</i> , <b>2011</b> , 37, 610-6	13.6	40
81	Polymorphisms in the interleukin-1 (IL1) gene cluster are not associated with aggressive periodontitis in a large Caucasian population. <i>Genomics</i> , <b>2008</b> , 92, 309-15	4.3	40
80	X chromosomal variation is associated with slow progression to AIDS in HIV-1-infected women. <i>American Journal of Human Genetics</i> , <b>2009</b> , 85, 228-39	11	39
79	COX-2 is associated with periodontitis in Europeans. <i>Journal of Dental Research</i> , <b>2010</b> , 89, 384-8	8.1	38
78	Meta-analysis identifies novel risk loci and yields systematic insights into the biology of male-pattern baldness. <i>Nature Communications</i> , <b>2017</b> , 8, 14694	17.4	36
77	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> , <b>2014</b> , 23, 3883-90	5.6	36
76	Technology-specific error signatures in the 1000 Genomes Project data. <i>Human Genetics</i> , <b>2011</b> , 130, 505-16	4.6	36
75	A Genome-wide Association Study of Dupuytren Disease Reveals 17 Additional Variants Implicated in Fibrosis. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 417-427	11	35
74	A novel sarcoidosis risk locus for Europeans on chromosome 11q13.1. <i>American Journal of Respiratory and Critical Care Medicine</i> , <b>2012</b> , 186, 877-85	10.2	34

73	Genome-wide investigation of gene-environment interactions in colorectal cancer. <i>Human Genetics</i> , <b>2013</b> , 132, 219-31	6.3	32
72	Association of toll-interacting protein gene polymorphisms with atopic dermatitis. <i>BMC Dermatology</i> , <b>2007</b> , 7, 3	2.1	30
71	Role of the toll-like receptor 4 polymorphism Asp299Gly in longevity and myocardial infarction in German men. <i>Mechanisms of Ageing and Development</i> , <b>2007</b> , 128, 409-11	5.6	28
70	Genome-wide association analysis reveals 12q13.3-q14.1 as new risk locus for sarcoidosis. <i>European Respiratory Journal</i> , <b>2013</b> , 41, 888-900	13.6	26
69	Expanding the genetic architecture of nicotine dependence and its shared genetics with multiple traits. <i>Nature Communications</i> , <b>2020</b> , 11, 5562	17.4	25
68	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> , <b>2008</b> , 7, 32	4.4	25
67	GABA(A) receptor- and GABA transporter polymorphisms and risk for essential tremor. <i>European Journal of Neurology</i> , <b>2011</b> , 18, 1098-100	6	24
66	Postprandial plasma adiponectin decreases after glucose and high fat meal and is independently associated with postprandial triacylglycerols but not with -- 11388 promoter polymorphism. <i>British Journal of Nutrition</i> , <b>2008</b> , 99, 76-82	3.6	22
65	The association of fatty acid-binding protein 2 A54T polymorphism with postprandial lipemia depends on promoter variability. <i>Metabolism: Clinical and Experimental</i> , <b>2007</b> , 56, 723-31	12.7	22
64	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> , <b>2016</b> , 11, e0146040	3.7	22
63	Haplotypes of IL-12R $\beta$ 1 impact on the clinical phenotype of hidradenitis suppurativa. <i>Cytokine</i> , <b>2013</b> , 62, 297-301	4	20
62	Distinct genetic variation and heterogeneity of the Iranian population. <i>PLoS Genetics</i> , <b>2019</b> , 15, e1008385		18
61	Statistical inference of allelic imbalance from transcriptome data. <i>Human Mutation</i> , <b>2011</b> , 32, 98-106	4.7	18
60	Current software for genotype imputation. <i>Human Genomics</i> , <b>2009</b> , 3, 371-80	6.8	18
59	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> , <b>2012</b> , 13, 14	2.1	17
58	Prognostic relevance of gastric cancer staging by endoscopic ultrasound. <i>Surgical Endoscopy and Other Interventional Techniques</i> , <b>2013</b> , 27, 1124-9	5.2	17
57	A genome-wide linkage analysis in 181 German sarcoidosis families using clustered biallelic markers. <i>Chest</i> , <b>2010</b> , 138, 151-7	5.3	17
56	The minor allele of the PPARgamma2 pro12Ala polymorphism is associated with lower postprandial TAG and insulin levels in non-obese healthy men. <i>British Journal of Nutrition</i> , <b>2007</b> , 97, 847-54	3.6	17

55	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> , <b>2016</b> , 11, e0158101	3.7	16
54	True colors: A literature review on the spatial distribution of eye and hair pigmentation. <i>Forensic Science International: Genetics</i> , <b>2019</b> , 39, 109-118	4.3	15
53	Shannon's equivocation for forensic Y-STR marker selection. <i>Forensic Science International: Genetics</i> , <b>2015</b> , 16, 216-225	4.3	14
52	Serum metabolomic profiling highlights pathways associated with liver fat content in a general population sample. <i>European Journal of Clinical Nutrition</i> , <b>2017</b> , 71, 995-1001	5.2	13
51	Clinical and experimental evidence suggest a link between KIF7 and C5orf42-related ciliopathies through Sonic Hedgehog signaling. <i>European Journal of Human Genetics</i> , <b>2018</b> , 26, 197-209	5.3	13
50	Genetic mapping of 15 human X chromosomal forensic short tandem repeat (STR) loci by means of multi-core parallelization. <i>Forensic Science International: Genetics</i> , <b>2016</b> , 25, 39-44	4.3	13
49	Exome-wide analysis of mutational burden in patients with typical and atypical Rolandic epilepsy. <i>European Journal of Human Genetics</i> , <b>2018</b> , 26, 258-264	5.3	12
48	Association studies of the copy-number variable $\beta$ -defensin cluster on 8p23.1 in adenocarcinoma and chronic pancreatitis. <i>BMC Research Notes</i> , <b>2012</b> , 5, 629	2.3	12
47	Variation in genes of the epidermal differentiation complex in German atopic dermatitis patients. <i>International Journal of Immunogenetics</i> , <b>2009</b> , 36, 217-22	2.3	12
46	Female-specific association of C-C chemokine receptor 5 gene polymorphisms with Ljgren's syndrome. <i>Journal of Molecular Medicine</i> , <b>2008</b> , 86, 553-61	5.5	12
45	Missense variants in NOX1 and p22phox in a case of very-early-onset inflammatory bowel disease are functionally linked to NOD2. <i>Journal of Physical Education and Sports Management</i> , <b>2019</b> , 5,	2.8	11
44	Effects of Pleistocene climatic fluctuations on the phylogeography, demography and population structure of a high-elevation snake species, <i>Thermophilisbaileyi</i> , on the Tibetan Plateau. <i>Journal of Biogeography</i> , <b>2014</b> , 41, 2162-2172	4.1	11
43	Association of postprandial and fasting triglycerides with traits of the metabolic syndrome in the Metabolic Intervention Cohort Kiel. <i>European Journal of Endocrinology</i> , <b>2010</b> , 162, 719-27	6.5	11
42	Role of NOD2/CARD15 in coronary heart disease. <i>BMC Genetics</i> , <b>2007</b> , 8, 76	2.6	11
41	Association screen for atopic dermatitis candidate gene regions using microsatellite markers in pooled DNA samples. <i>International Journal of Immunogenetics</i> , <b>2006</b> , 33, 401-9	2.3	10
40	Efficacy assessment of SNP sets for genome-wide disease association studies. <i>Nucleic Acids Research</i> , <b>2007</b> , 35, e113	20.1	10
39	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. <i>PLoS ONE</i> , <b>2016</b> , 11, e0150426	3.7	10
38	The Slavic NBN Founder Mutation: A Role for Reproductive Fitness?. <i>PLoS ONE</i> , <b>2016</b> , 11, e0167984	3.7	10

37	SFRS10--a splicing factor gene reduced in human obesity?. <i>Cell Metabolism</i> , <b>2012</b> , 15, 265-6; author reply 267-9	24.6	9
36	A Rare Variant Nonparametric Linkage Method for Nuclear and Extended Pedigrees with Application to Late-Onset Alzheimer Disease via WGS Data. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 822-835	11	8
35	Common genetic risk variants of TLR2 are not associated with periodontitis in large European case-control populations. <i>Journal of Clinical Periodontology</i> , <b>2012</b> , 39, 315-22	7.7	8
34	Pipeline for large-scale microdroplet bisulfite PCR-based sequencing allows the tracking of hepitype evolution in tumors. <i>PLoS ONE</i> , <b>2011</b> , 6, e21332	3.7	8
33	Depletion of potential A2M risk haplotype for Alzheimer's disease in long-lived individuals. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 59-61	5.3	8
32	Diagnosing fatty liver disease: a comparative evaluation of metabolic markers, phenotypes, genotypes and established biomarkers. <i>PLoS ONE</i> , <b>2013</b> , 8, e76813	3.7	8
31	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> , <b>2018</b> , 13, e0203398	3.7	8
30	Family-Based Benchmarking of Copy Number Variation Detection Software. <i>PLoS ONE</i> , <b>2015</b> , 10, e0133465	5.7	7
29	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> , <b>2009</b> , 17, 967-75	5.3	7
28	LINGO1 is not associated with Parkinson's disease in German patients. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2010</b> , 153B, 1173-8	3.5	6
27	Guideline-based and bioinformatic reassessment of lesion-associated gene and variant pathogenicity in focal human epilepsies. <i>Epilepsia</i> , <b>2018</b> , 59, 2145-2152	6.4	6
26	Approaches to the genetics of cardiovascular disease through genetic field work. <i>Kidney International</i> , <b>1998</b> , 53, 1449-54	9.9	5
25	NOD1 gene polymorphisms in relation to aggressive periodontitis. <i>Innate Immunity</i> , <b>2009</b> , 15, 225-32	2.7	4
24	The effect of FABP2 promoter haplotype on response to a diet with medium-chain triacylglycerols. <i>Genes and Nutrition</i> , <b>2012</b> , 7, 437-45	4.3	3
23	Comparative assessment of the association information captured by SNP tagging. <i>Human Heredity</i> , <b>2007</b> , 64, 27-34	1.1	3
22	Mutations Causing Complex Disease May under Certain Circumstances Be Protective in an Epidemiological Sense. <i>PLoS ONE</i> , <b>2015</b> , 10, e0132150	3.7	3
21	Expanding the Genetic Architecture of Nicotine Dependence and its Shared Genetics with Multiple Traits: Findings from the Nicotine Dependence GenOmics (iNDiGO) Consortium		3
20	Evaluation of supervised machine-learning methods for predicting appearance traits from DNA. <i>Forensic Science International: Genetics</i> , <b>2021</b> , 53, 102507	4.3	3



19	Metabolic signature of electrosurgical liver dissection. <i>PLoS ONE</i> , <b>2013</b> , 8, e72022	3.7	2
18	Statistical gene mapping of traits in humans--hypertension as a complex trait: is it amenable to genetic analysis?. <i>Seminars in Nephrology</i> , <b>2002</b> , 22, 105-14	4.8	2
17	Pathway-induced allelic spectra of diseases in the presence of strong genetic effects. <i>Human Genetics</i> , <b>2018</b> , 137, 215-230	6.3	1
16	CoNCoS: copy number estimation in cancer with controlled support. <i>Journal of Bioinformatics and Computational Biology</i> , <b>2015</b> , 13, 1550027	1	1
15	Hypotheses in genome-wide association scans. <i>European Journal of Human Genetics</i> , <b>2008</b> , 16, 1174-5; author reply 1175	5.3	1
14	A Critical Evaluation of Analytic Aspects of Gene Expression Profiling in Lymphoid Leukemias with Broad Applications to Cancer Genomics. <i>AIMS Medical Science</i> , <b>2016</b> , 3, 248-271	0.4	1
13	Exome sequencing utility in defining the genetic landscape of hearing loss and novel-gene discovery in Iran. <i>Clinical Genetics</i> , <b>2021</b> , 100, 59-78	4	1
12	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> , <b>2019</b> , 39, e1-e2	4.3	1
11	The impact of correlations between pigmentation phenotypes and underlying genotypes on genetic prediction of pigmentation traits. <i>Forensic Science International: Genetics</i> , <b>2021</b> , 50, 102395	4.3	1
10	The exhaustive genomic scan approach, with an application to rare-variant association analysis. <i>European Journal of Human Genetics</i> , <b>2020</b> , 28, 1283-1291	5.3	0
9	Special issue on 'Genetic epidemiology of complex diseases: impact of population history and modelling assumptions'. <i>Human Genetics</i> , <b>2020</b> , 139, 1-3	6.3	0
8	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> , <b>2021</b> , 51, 102427	4.3	0
7	Testing the impact of trait prevalence priors in Bayesian-based genetic prediction modeling of human appearance traits. <i>Forensic Science International: Genetics</i> , <b>2021</b> , 50, 102412	4.3	0
6	Distinct gene-set burden patterns underlie common generalized and focal epilepsies. <i>EBioMedicine</i> , <b>2021</b> , 72, 103588	8.8	0
5	Securing the use of existing sample collections for future human genetic research. <i>European Journal of Human Genetics</i> , <b>2017</b> , 25, 522-529	5.3	
4	Towards a fine-scale picture of European genetic diversity. <i>European Journal of Human Genetics</i> , <b>2020</b> , 28, 851-852	5.3	
3	Modellvorstellungen zur Genetik multifaktorieller Krankheiten. <i>Medizinische Genetik</i> , <b>2007</b> , 19, 295-299	0.5	
2	Analysis of single nucleotide polymorphisms in chronic beryllium disease. <i>Respiratory Research</i> , <b>2021</b> , 22, 107	7.3	



- 1 What Makes a Hot-Spring Habitat Hot? For the Hot-Spring Snake: Distributional Data and Niche Modelling for the Genus *Thermophis* (Serpentes, Colubridae). *Diversity*, **2021**, 13, 325 2.5