Michael Nothnagel

List of Publications by Year
in descending order

125 papers	8,707 citations	47006 47 h-index	45317 90 g-index
134 all docs	134 docs citations	134 times ranked	15607 citing authors

#	Article	IF	Citations
1	Benchmarking of univariate pleiotropy detection methods applied to epilepsy. Human Mutation, 2022, 43, 1314-1332.	2.5	0
2	The impact of correlations between pigmentation phenotypes and underlying genotypes on genetic prediction of pigmentation traits. Forensic Science International: Genetics, 2021, 50, 102395.	3.1	7
3	A Y-chromosomal survey of Ecuadorâ€™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. Forensic Science International: Genetics, 2021, 51, 102427.	3.1	1
4	Testing the impact of trait prevalence priors in Bayesian-based genetic prediction modeling of human appearance traits. Forensic Science International: Genetics, 2021, 50, 102412.	3.1	3
5	Exome sequencing utility in defining the genetic landscape of hearing loss and novelâ€gene discovery in Iran. Clinical Genetics, 2021, 100, 59-78.	2.0	4
6	Analysis of single nucleotide polymorphisms in chronic beryllium disease. Respiratory Research, 2021, 22, 107.	3.6	1
7	What Makes a Hot-Spring Habitat â€œHotâ€•for the Hot-Spring Snake: Distributional Data and Niche Modelling for the Genus Thermophis (Serpentes, Colubridae). Diversity, 2021, 13, 325.	1.7	1
8	Evaluation of supervised machine-learning methods for predicting appearance traits from DNA. Forensic Science International: Genetics, 2021, 53, 102507.	3.1	11
9	Distinct gene-set burden patterns underlie common generalized and focal epilepsies. EBioMedicine, 2021, 72, 103588.	6.1	7
10	Special issue on â€˜Genetic epidemiology of complex diseases: impact of population history and modelling assumptionsâ€™. Human Genetics, 2020, 139, 1-3.	3.8	1
11	Expanding the genetic architecture of nicotine dependence and its shared genetics with multiple traits. Nature Communications, 2020, 11, 5562.	12.8	80
12	The exhaustive genomic scan approach, with an application to rare-variant association analysis. European Journal of Human Genetics, 2020, 28, 1283-1291.	2.8	3
13	Towards a fine-scale picture of European genetic diversity. European Journal of Human Genetics, 2020, 28, 851-852.	2.8	0
14	Heterozygous carriage of the alpha1-antitrypsin Pi*Z variant increases the risk to develop liver cirrhosis. Gut, 2019, 68, 1099-1107.	12.1	100
15	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.	6.2	16
16	Distinct genetic variation and heterogeneity of the Iranian population. PLoS Genetics, 2019, 15, e1008385.	3.5	34
17	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.	1.2	13
18	True colors: A literature review on the spatial distribution of eye and hair pigmentation. Forensic Science International: Genetics, 2019, 39, 109-118.	3.1	24

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19	Unsupported claim of significant discrimination between monozygotic twins from multiple pairs based on three age-related DNA methylation markers. Forensic Science International: Genetics, 2019, 39, e1-e2.	3.1	1
20	Pathway-induced allelic spectra of diseases in the presence of strong genetic effects. Human Genetics, 2018, 137, 215-230.	3.8	1
21	Exome-wide analysis of mutational burden in patients with typical and atypical Rolandic epilepsy. European Journal of Human Genetics, 2018, 26, 258-264.	2.8	22
22	Clinical and experimental evidence suggest a link between KIF7 and C5orf42-related ciliopathies through Sonic Hedgehog signaling. European Journal of Human Genetics, 2018, 26, 197-209.	2.8	23
23	Ancient DNA study reveals HLA susceptibility locus for leprosy in medieval Europeans. Nature Communications, 2018, 9, 1569.	12.8	67
24	Guidelineâ€based and bioinformatic reassessment of lesionâ€associated gene and variant pathogenicity in focal human epilepsies. Epilepsia, 2018, 59, 2145-2152.	5.1	8
25	Evaluation of potential effects of Plastin 3 overexpression and low-dose SMN-antisense oligonucleotides on putative biomarkers in spinal muscular atrophy mice. PLoS ONE, 2018, 13, e0203398.	2.5	11
26	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	10.2	67
27	Performance of in silico prediction tools for the classification of rare BRCA1/2 missense variants in clinical diagnostics. BMC Medical Genomics, 2018, 11, 35.	1.5	78
28	Securing the use of existing sample collections for future human genetic research. European Journal of Human Genetics, 2017, 25, 522-529.	2.8	0
29	Meta-analysis identifies novel risk loci and yields systematic insights into the biology of male-pattern baldness. Nature Communications, 2017, 8, 14694.	12.8	58
30	Serum metabolomic profiling highlights pathways associated with liver fat content in a general population sample. European Journal of Clinical Nutrition, 2017, 71, 995-1001.	2.9	20
31	A Genome-wide Association Study of Dupuytren Disease Reveals 17 Additional Variants Implicated in Fibrosis. American Journal of Human Genetics, 2017, 101, 417-427.	6.2	67
32	Identification and characterization of two functional variants in the human longevity gene FOXO3. Nature Communications, 2017, 8, 2063.	12.8	69
33	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 Dupuytrenâ€™s Disease. PLoS ONE, 2016, 11, e0158101.	2.5	26
34	The Slavic NBN Founder Mutation: A Role for Reproductive Fitness?. PLoS ONE, 2016, 11, e0167984.	2.5	21
35	Benign infantile seizures and paroxysmal dyskinesia caused by an SCN8A mutation. Annals of Neurology, 2016, 79, 428-436.	5.3	159
36	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.	3.1	21

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37	Increased Probability of Co-Occurrence of Two Rare Diseases in Consanguineous Families and Resolution of a Complex Phenotype by Next Generation Sequencing. PLoS ONE, 2016, 11, e0146040.	2.5	32
38	Evaluation of Presumably Disease Causing SCN1A Variants in a Cohort of Common Epilepsy Syndromes. PLoS ONE, 2016, 11, e0150426.	2.5	22
39	A Critical Evaluation of Analytic Aspects of Gene Expression Profiling in Lymphoid Leukemias with Broad Applications to Cancer Genomics. AIMS Medical Science, 2016, 3, 248-271.	0.4	1
40	Rare variants in Î³â€aminobutyric acid type <scp>A</scp> receptor genes in rolandic epilepsy and related syndromes. Annals of Neurology, 2015, 77, 972-986.	5.3	51
41	Family-Based Benchmarking of Copy Number Variation Detection Software. PLoS ONE, 2015, 10, e0133465.	2.5	9
42	CoNCoS: Copy number estimation in cancer with controlled support. Journal of Bioinformatics and Computational Biology, 2015, 13, 1550027.	0.8	1
43	Shannon's equivocation for forensic Y-STR marker selection. Forensic Science International: Genetics, 2015, 16, 216-225.	3.1	18
44	De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Nature Genetics, 2015, 47, 393-399.	21.4	224
45	A genome-wide association study confirms PNPLA3 and identifies TM6SF2 and MBOAT7 as risk loci for alcohol-related cirrhosis. Nature Genetics, 2015, 47, 1443-1448.	21.4	435
46	Mutations Causing Complex Disease May under Certain Circumstances Be Protective in an Epidemiological Sense. PLoS ONE, 2015, 10, e0132150.	2.5	4
47	Evaluation of genome-wide loci of iron metabolism in hereditary hemochromatosis identifies PCSK7 as a host risk factor of liver cirrhosis. Human Molecular Genetics, 2014, 23, 3883-3890.	2.9	50
48	A global analysis of Y-chromosomal haplotype diversity for 23 STR loci. Forensic Science International: Genetics, 2014, 12, 12-23.	3.1	214
49	Effects of <scp>P</scp>leistocene climatic fluctuations on the phylogeography, demography and population structure of a highâ€elevation snake species, <scp>T</scp>hermophisÂbaileyi, on the Tibetan Plateau. Journal of Biogeography, 2014, 41, 2162-2172.	3.0	14
50	Prognostic relevance of gastric cancer staging by endoscopic ultrasound. Surgical Endoscopy and Other Interventional Techniques, 2013, 27, 1124-1129.	2.4	23
51	Association Between Variants of PRDM1 and NDP52 and Crohn's Disease, Based on Exome Sequencing and Functional Studies. Gastroenterology, 2013, 145, 339-347.	1.3	149
52	Mutations in GRIN2A cause idiopathic focal epilepsy with rolandic spikes. Nature Genetics, 2013, 45, 1067-1072.	21.4	391
53	Genome-wide investigation of geneâ€“environment interactions in colorectal cancer. Human Genetics, 2013, 132, 219-231.	3.8	38
54	Haplotypes of IL-12RÎ²1 impact on the clinical phenotype of hidradenitis suppurativa. Cytokine, 2013, 62, 297-301.	3.2	26

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55	Genome-wide association analysis reveals 12q13.3â€“q14.1 as new risk locus for sarcoidosis. European Respiratory Journal, 2013, 41, 888-900.	6.7	43
56	Continent-Wide Decoupling of Y-Chromosomal Genetic Variation from Language and Geography in Native South Americans. PLoS Genetics, 2013, 9, e1003460.	3.5	89
57	Validation of reported genetic risk factors for periodontitis in a largeâ€scale replication study. Journal of Clinical Periodontology, 2013, 40, 563-572.	4.9	74
58	Genetic and functional identification of the likely causative variant for cholesterol gallstone disease at theABCG5/8lithogenic locus. Hepatology, 2013, 57, 2407-2417.	7.3	74
59	Metabolic Signature of Electrosurgical Liver Dissection. PLoS ONE, 2013, 8, e72022.	2.5	2
60	Diagnosing Fatty Liver Disease: A Comparative Evaluation of Metabolic Markers, Phenotypes, Genotypes and Established Biomarkers. PLoS ONE, 2013, 8, e76813.	2.5	8
61	Schizophrenia risk polymorphisms in the TCF4 gene interact with smoking in the modulation of auditory sensory gating. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 6271-6276.	7.1	60
62	A Novel Sarcoidosis Risk Locus for Europeans on Chromosome 11q13.1. American Journal of Respiratory and Critical Care Medicine, 2012, 186, 877-885.	5.6	51
63	Collaborative genetic mapping of 12 forensic short tandem repeat (STR) loci on the human X chromosome. Forensic Science International: Genetics, 2012, 6, 778-784.	3.1	60
64	Genome-wide search for novel human uORFs and N-terminal protein extensions using ribosomal footprinting. Genome Research, 2012, 22, 2208-2218.	5.5	198
65	SFRS10â€”A Splicing Factor Gene Reduced in Human Obesity?. Cell Metabolism, 2012, 15, 265-266.	16.2	11
66	Association studies of the copy-number variable ÃŸ-defensin cluster on 8p23.1 in adenocarcinoma and chronic pancreatitis. BMC Research Notes, 2012, 5, 629.	1.4	12
67	Combined Analysis of Genome-wide Association Studies for Crohn Disease and Psoriasis Identifies Seven Shared Susceptibility Loci. American Journal of Human Genetics, 2012, 90, 636-647.	6.2	290
68	Polymorphisms in the glial glutamate transporter SLC1A2 are associated with essential tremor. Neurology, 2012, 79, 243-248.	1.1	111
69	The effect of FABP2 promoter haplotype on response to a diet with medium-chain triacylglycerols. Genes and Nutrition, 2012, 7, 437-445.	2.5	3
70	Common genetic risk variants of <scp>TLR</scp>2 are not associated with periodontitis in large <scp>E</scp>uropean caseâ€control populations. Journal of Clinical Periodontology, 2012, 39, 315-322.	4.9	8
71	A case-only study of gene-environment interaction between genetic susceptibility variants in NOD2 and cigarette smoking in Crohn's disease aetiology. BMC Medical Genetics, 2012, 13, 14.	2.1	22
72	CDKN2BAS is associated with periodontitis in different European populations and is activated by bacterial infection. Journal of Medical Genetics, 2011, 48, 38-47.	3.2	61

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73	Pipeline for Large-Scale Microdroplet Bisulfite PCR-Based Sequencing Allows the Tracking of Hepitype Evolution in Tumors. PLoS ONE, 2011, 6, e21332.	2.5	8
74	GABAA receptor- and GABA transporter polymorphisms and risk for essential tremor. European Journal of Neurology, 2011, 18, 1098-1100.	3.3	28
75	A genome-wide association study confirms APOE as the major gene influencing survival in long-lived individuals. Mechanisms of Ageing and Development, 2011, 132, 324-330.	4.6	184
76	Technology-specific error signatures in the 1000 Genomes Project data. Human Genetics, 2011, 130, 505-516.	3.8	37
77	Statistical inference of allelic imbalance from transcriptome data. Human Mutation, 2011, 32, 98-106.	2.5	33
78	A genome-wide association study reveals evidence of association with sarcoidosis at 6p12.1. European Respiratory Journal, 2011, 38, 1127-1135.	6.7	58
79	Association of inflammatory bowel disease risk loci with sarcoidosis, and its acute and chronic subphenotypes. European Respiratory Journal, 2011, 37, 610-616.	6.7	53
80	Wnt Signaling and Dupuytren's Disease. New England Journal of Medicine, 2011, 365, 307-317.	27.0	201
81	Potentials and limits of pairwise kinship analysis using autosomal short tandem repeat loci. International Journal of Legal Medicine, 2010, 124, 205-215.	2.2	48
82	LINGO1 is not associated with Parkinson's disease in German patients. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 1173-1178.	1.7	11
83	LINGO1 polymorphisms are associated with essential tremor in Europeans. Movement Disorders, 2010, 25, 717-723.	3.9	47
84	Depletion of potential A2M risk haplotype for Alzheimer's disease in long-lived individuals. European Journal of Human Genetics, 2010, 18, 59-61.	2.8	11
85	A 3â€² UTR transition within DEFB1 is associated with chronic and aggressive periodontitis. Genes and Immunity, 2010, 11, 45-54.	4.1	62
86	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.	3.7	13
87	Genomic and geographic distribution of SNP-defined runs of homozygosity in Europeans. Human Molecular Genetics, 2010, 19, 2927-2935.	2.9	146
88	A genome-wide association study identifies GLT6D1 as a susceptibility locus for periodontitis. Human Molecular Genetics, 2010, 19, 553-562.	2.9	176
89	A Genome-Wide Linkage Analysis in 181 German Sarcoidosis Families Using Clustered Biallelic Markers. Chest, 2010, 138, 151-157.	0.8	20
90	Genome-Wide Association Analysis in Primary Sclerosing Cholangitis. Gastroenterology, 2010, 138, 1102-1111.	1.3	325

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91	Loci From a Genome-Wide Analysis of Bilirubin Levels Are Associated With Gallstone Risk and Composition. Gastroenterology, 2010, 139, 1942-1951.e2.	1.3	96
92	COX-2 Is Associated with Periodontitis in Europeans. Journal of Dental Research, 2010, 89, 384-388.	5.2	43
93	Identification of a Shared Genetic Susceptibility Locus for Coronary Heart Disease and Periodontitis. PLoS Genetics, 2009, 5, e1000378.	3.5	189
94	NOD1 gene polymorphisms in relation to aggressive periodontitis. Innate Immunity, 2009, 15, 225-232.	2.4	5
95	Variation in genes of the epidermal differentiation complex in German atopic dermatitis patients. International Journal of Immunogenetics, 2009, 36, 217-222.	1.8	15
96	A comprehensive evaluation of SNP genotype imputation. Human Genetics, 2009, 125, 163-171.	3.8	139
97	An evaluation of the genetic-matched pair study design using genome-wide SNP data from the European population. European Journal of Human Genetics, 2009, 17, 967-975.	2.8	8
98	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	21.4	511
99	X Chromosomal Variation Is Associated with Slow Progression to AIDS in HIV-1-Infected Women. American Journal of Human Genetics, 2009, 85, 228-239.	6.2	41
100	Current software for genotype imputation. Human Genomics, 2009, 3, 371-80.	2.9	19
101	Genome-wide association study identifies ANXA11 as a new susceptibility locus for sarcoidosis. Nature Genetics, 2008, 40, 1103-1106.	21.4	239
102	Hypotheses in genome-wide association scans. European Journal of Human Genetics, 2008, 16, 1174-1175.	2.8	1
103	Sequence variants in IL10, ARPC2 and multiple other loci contribute to ulcerative colitis susceptibility. Nature Genetics, 2008, 40, 1319-1323.	21.4	534
104	Correlation between Genetic and Geographic Structure in Europe. Current Biology, 2008, 18, 1241-1248.	3.9	449
105	Genome-Wide Association Analysis in Sarcoidosis and Crohn's Disease Unravels a Common Susceptibility Locus on 10p12.2. Gastroenterology, 2008, 135, 1207-1215.	1.3	85
106	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. Lipids in Health and Disease, 2008, 7, 32.	3.0	33
107	Polymorphisms in the interleukin-1 (IL1) gene cluster are not associated with aggressive periodontitis in a large Caucasian population. Genomics, 2008, 92, 309-315.	2.9	45
108	Female-specific association of C-C chemokine receptor 5 gene polymorphisms with LÃ¶fgrenâ€™s syndrome. Journal of Molecular Medicine, 2008, 86, 553-561.	3.9	15

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109	The Wegener's granulomatosis quantitative trait locus on chromosome 6p21.3 as characterised by tagSNP genotyping. Annals of the Rheumatic Diseases, 2008, 67, 972-979.	0.9	79
110	Postprandial plasma adiponectin decreases after glucose and high fat meal and is independently associated with postprandial triacylglycerols but not withÂâˆ'Â11388 promoter polymorphism. British Journal of Nutrition, 2008, 99, 76-82.	2.3	24
111	Comparative Assessment of the Association Information Captured by SNP Tagging. Human Heredity, 2007, 64, 27-34.	0.8	3
112	Efficacy assessment of SNP sets for genome-wide disease association studies. Nucleic Acids Research, 2007, 35, e113-e113.	14.5	15
113	The minor allele of the PPARÎ³2 Pro12Ala polymorphism is associated with lower postprandial TAG and insulin levels in non-obese healthy men. British Journal of Nutrition, 2007, 97, 847-854.	2.3	20
114	The association of fatty acidâ€“binding protein 2 A54T polymorphism with postprandial lipemia depends on promoter variability. Metabolism: Clinical and Experimental, 2007, 56, 723-731.	3.4	24
115	Role of NOD2/CARD15 in coronary heart disease. BMC Genetics, 2007, 8, 76.	2.7	12
116	Association of toll-interacting protein gene polymorphisms with atopic dermatitis. BMC Dermatology, 2007, 7, 3.	2.1	34
117	Polymorphisms in NACHTâ€ŁRR (NLR) genes in atopic dermatitis. Experimental Dermatology, 2007, 16, 692-698.	2.9	108
118	Role of the toll-like receptor 4 polymorphism Asp299Gly in longevity and myocardial infarction in German men. Mechanisms of Ageing and Development, 2007, 128, 409-411.	4.6	30
119	Modellvorstellungen zur Genetik multifaktorieller Krankheiten. Medizinische Genetik, 2007, 19, 295-299.	0.2	0
120	Association screen for atopic dermatitis candidate gene regions using microsatellite markers in pooled DNA samples. International Journal of Immunogenetics, 2006, 33, 401-409.	1.8	11
121	The Effect of Single-Nucleotide Polymorphism Marker Selection on Patterns of Haplotype Blocks and Haplotype Frequency Estimates. American Journal of Human Genetics, 2005, 77, 988-998.	6.2	66
122	Entropy as a Measure for Linkage Disequilibrium over Multilocus Haplotype Blocks. Human Heredity, 2002, 54, 186-198.	0.8	81
123	Power and Sample Size Calculations for Case-Control Genetic Association Tests when Errors Are Present: Application to Single Nucleotide Polymorphisms. Human Heredity, 2002, 54, 22-33.	0.8	269
124	Statistical gene mapping of traits in humansâ€"hypertension as a complex trait: Is it amenable to genetic analysis?. Seminars in Nephrology, 2002, 22, 105-114.	1.6	3
125	Approaches to the genetics of cardiovascular disease through genetic field work. Kidney International, 1998, 53, 1449-1454.	5.2	6