

# Nicholas Lench

## List of Publications by Year in descending order

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81  
papers

7,213  
citations

101543

36  
h-index

64796

79  
g-index

81  
all docs

81  
docs citations

81  
times ranked

7511  
citing authors

#	ARTICLE	IF	CITATIONS
1	Diagnostic implications of genetic copy number variation in epilepsy plus. <i>Epilepsia</i> , 2019, 60, 689-706.	5.1	61
2	A targeted sequencing panel identifies rare damaging variants in multiple genes in the cranial neural tube defect, anencephaly. <i>Clinical Genetics</i> , 2018, 93, 870-879.	2.0	29
3	Exome Sequencing and Rare Variant Analysis Reveals Multiple Filaggrin Mutations in Bangladeshi Families with Atopic Eczema and Additional Risk Genes. <i>Journal of Investigative Dermatology</i> , 2018, 138, 2674-2677.	0.7	37
4	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. <i>Brain</i> , 2016, 139, 2844-2854.	7.6	35
5	Favourable response to ketogenic dietary therapies: undiagnosed glucose 1 transporter deficiency syndrome is only one factor. <i>Developmental Medicine and Child Neurology</i> , 2015, 57, 969-976.	2.1	8
6	Exome sequencing for prenatal diagnosis of fetuses with sonographic abnormalities. <i>Prenatal Diagnosis</i> , 2015, 35, 1010-1017.	2.3	189
7	<i>CHD2</i> variants are a risk factor for photosensitivity in epilepsy. <i>Brain</i> , 2015, 138, 1198-1208.	7.6	112
8	Non-invasive prenatal diagnosis of achondroplasia and thanatophoric dysplasia: next-generation sequencing allows for a safer, more accurate, and comprehensive approach. <i>Prenatal Diagnosis</i> , 2015, 35, 656-662.	2.3	156
9	Functional analysis of four LDLR 5'UTR and promoter variants in patients with familial hypercholesterolaemia. <i>European Journal of Human Genetics</i> , 2015, 23, 790-795.	2.8	18
10	Confined placental mosaicism: implications for fetal chromosomal analysis using microarray comparative genomic hybridization. <i>Prenatal Diagnosis</i> , 2014, 34, 98-101.	2.3	14
11	A novel homozygous <i>ERCC5</i> truncating mutation in a family with prenatal arthrogyposis—Further evidence of genotype-phenotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1777-1783.	1.2	31
12	A novel heterozygous SOX2 mutation causing congenital bilateral anophthalmia, hypogonadotropic hypogonadism and growth hormone deficiency. <i>Gene</i> , 2014, 534, 282-285.	2.2	25
13	Targeted gene panel sequencing in children with very early onset inflammatory bowel disease—evaluation and prospective analysis. <i>Journal of Medical Genetics</i> , 2014, 51, 748-755.	3.2	91
14	Evaluation of non-invasive prenatal testing (NIPT) for aneuploidy in an NHS setting: a reliable accurate prenatal non-invasive diagnosis (RAPID) protocol. <i>BMC Pregnancy and Childbirth</i> , 2014, 14, 229.	2.4	72
15	Clinico-pathological correlations of congenital and infantile nephrotic syndrome over twenty years. <i>Pediatric Nephrology</i> , 2014, 29, 2173-2180.	1.7	24
16	Whole exome sequencing of familial hypercholesterolaemia patients negative for <i>LDLR</i> , <i>APOB</i> , <i>PCSK9</i> mutations. <i>Journal of Medical Genetics</i> , 2014, 51, 537-544.	3.2	104
17	The clinical implementation of non-invasive prenatal diagnosis for single gene disorders: challenges and progress made. <i>Prenatal Diagnosis</i> , 2013, 33, 555-562.	2.3	121
18	The future role of genetic screening to detect newborns at risk of childhood-onset hearing loss. <i>International Journal of Audiology</i> , 2013, 52, 124-133.	1.7	23

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19	Use of low-density lipoprotein cholesterol gene score to distinguish patients with polygenic and monogenic familial hypercholesterolaemia: a case-control study. <i>Lancet, The</i> , 2013, 381, 1293-1301.	13.7	485
20	KCNJ10 Mutations Display Differential Sensitivity to Heteromerisation with KCNJ16. <i>Nephron Physiology</i> , 2013, 123, 7-14.	1.2	34
21	Neurological features of epilepsy, ataxia, sensorineural deafness, tubulopathy syndrome. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 846-856.	2.1	53
22	Evaluation of Real-Time Quantitative PCR as a Standard Cytogenetic Diagnostic Tool for Confirmation of Microarray (aCGH) Results and Determination of Inheritance. <i>Genetic Testing and Molecular Biomarkers</i> , 2013, 17, 821-825.	0.7	5
23	Low-Density Lipoprotein Receptor Gene Familial Hypercholesterolemia Variant Database: Update and Pathological Assessment. <i>Annals of Human Genetics</i> , 2012, 76, 387-401.	0.8	173
24	Array comparative genomic hybridization: Results from an adult population with drug-resistant epilepsy and co-morbidities. <i>European Journal of Medical Genetics</i> , 2012, 55, 342-348.	1.3	28
25	Genetic testing in renal disease. <i>Pediatric Nephrology</i> , 2012, 27, 873-883.	1.7	45
26	Sotos syndrome, infantile hypercalcemia, and nephrocalcinosis: a contiguous gene syndrome. <i>Pediatric Nephrology</i> , 2011, 26, 1331-1334.	1.7	29
27	Uncovering Genomic Causes of Co-Morbidity in Epilepsy: Gene-Driven Phenotypic Characterization of Rare Microdeletions. <i>PLoS ONE</i> , 2011, 6, e23182.	2.5	24
28	Molecular prenatal diagnosis: the impact of modern technologies. <i>Prenatal Diagnosis</i> , 2010, 30, 674-681.	2.3	58
29	Single-Point Haplotype Scores Telomeric to Human Leukocyte Antigen-C Give a High Susceptibility Major Histocompatibility Complex Haplotype for Psoriasis in a Caucasian Population. <i>Journal of Investigative Dermatology</i> , 2005, 124, 545-552.	0.7	11
30	Inflammatory Bowel Disease Is Linked to 19p13 and Associated with ICAM-1. <i>Inflammatory Bowel Diseases</i> , 2004, 10, 173-181.	1.9	52
31	Positional cloning of a novel gene influencing asthma from Chromosome 2q14. <i>Nature Genetics</i> , 2003, 35, 258-263.	21.4	326
32	Analysis of the IBD5 locus and potential gene-gene interactions in Crohn's disease. <i>Gut</i> , 2003, 52, 541-546.	12.1	96
33	The IBD6 Crohn's disease locus demonstrates complex interactions with CARD15 and IBD5 disease-associated variants. <i>Human Molecular Genetics</i> , 2003, 12, 2569-2575.	2.9	57
34	Molecular analysis for genetic counselling in amelogenesis imperfecta. <i>Oral Diseases</i> , 2002, 8, 249-253.	3.0	5
35	Extent and Distribution of Linkage Disequilibrium in Three Genomic Regions. <i>American Journal of Human Genetics</i> , 2001, 68, 191-197.	6.2	325
36	Replication and extension studies of inflammatory bowel disease susceptibility regions confirm linkage to chromosome 6p (IBD3). <i>European Journal of Human Genetics</i> , 2001, 9, 627-633.	2.8	70

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37	A gene for ataxic cerebral palsy maps to chromosome 9p12-q12. <i>European Journal of Human Genetics</i> , 2000, 8, 267-272.	2.8	42
38	A new locus for autosomal recessive non-syndromal sensorineural hearing impairment (DFNB27) on chromosome 2q23-q31. <i>European Journal of Human Genetics</i> , 2000, 8, 991-993.	2.8	21
39	Detection of a Novel Mutation in X-linked Amelogenesis Imperfecta. <i>Journal of Dental Research</i> , 2000, 79, 1978-1982.	5.2	50
40	A Third Novel Locus for Primary Autosomal Recessive Microcephaly Maps to Chromosome 9q34. <i>American Journal of Human Genetics</i> , 2000, 66, 724-727.	6.2	105
41	Loss-of-function mutations in the cathepsin C gene result in periodontal disease and palmoplantar keratosis. <i>Nature Genetics</i> , 1999, 23, 421-424.	21.4	442
42	DFNB20: a novel locus for autosomal recessive, non-syndromal sensorineural hearing loss maps to chromosome 11q25-qter. <i>European Journal of Human Genetics</i> , 1999, 7, 243-246.	2.8	23
43	The second locus for autosomal recessive primary microcephaly (MCPH2) maps to chromosome 19q13.1-q13.2. <i>European Journal of Human Genetics</i> , 1999, 7, 815-820.	2.8	103
44	A Gene for Autosomal Recessive Symmetrical Spastic Cerebral Palsy Maps to Chromosome 2q24-25. <i>American Journal of Human Genetics</i> , 1999, 64, 526-532.	6.2	46
45	Congenital non-syndromal sensorineural hearing impairment due to connexin 26 gene mutations - molecular and audiological findings. <i>International Journal of Pediatric Otorhinolaryngology</i> , 1999, 50, 3-13.	1.0	57
46	Connexin-26 mutations in sporadic non-syndromal sensorineural deafness. <i>Lancet</i> , The, 1998, 351, 415.	13.7	109
47	A Syndrome of Severe Mental Retardation, Spasticity, and Tapetoretinal Degeneration Linked to Chromosome 15q24. <i>American Journal of Human Genetics</i> , 1998, 62, 1070-1076.	6.2	15
48	Autozygosity Mapping, to Chromosome 11q25, of a Rare Autosomal Recessive Syndrome Causing Histiocytosis, Joint Contractures, and Sensorineural Deafness. <i>American Journal of Human Genetics</i> , 1998, 62, 1123-1128.	6.2	29
49	Primary Autosomal Recessive Microcephaly (MCPH1) Maps to Chromosome 8p22-pter. <i>American Journal of Human Genetics</i> , 1998, 63, 541-546.	6.2	151
50	cDNA Cloning, Genomic Organization, and Chromosomal Localization of a Novel Human Gene That Encodes a Kinesin-Related Protein Highly Similar to Mouse Kif3C. <i>Biochemical and Biophysical Research Communications</i> , 1998, 242, 407-412.	2.1	6
51	Prelingual Deafness: High Prevalence of a 30delG Mutation in the Connexin 26 Gene. <i>Human Molecular Genetics</i> , 1997, 6, 2173-2177.	2.9	601
52	Assignment of the STAT6 gene (STAT6) to human chromosome band 12q13 by in situ hybridization. <i>Cytogenetic and Genome Research</i> , 1997, 79, 208-209.	1.1	14
53	Assignment of the Rab13 gene (RAB13) to human chromosome band 12q13 by in situ hybridization. <i>Cytogenetic and Genome Research</i> , 1997, 79, 210-211.	1.1	0
54	DNA diagnosis of X-linked amelogenesis imperfecta (AIH1). <i>Journal of Oral Pathology and Medicine</i> , 1997, 26, 135-137.	2.7	12

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55	Connexin 26 mutations in hereditary non-syndromic sensorineural deafness. <i>Nature</i> , 1997, 387, 80-83.	27.8	1,363
56	The human gene encoding FKBP-rapamycin associated protein (FRAP) maps to chromosomal band 1p36.2. <i>Human Genetics</i> , 1997, 99, 547-549.	3.8	12
57	Characterisation of human patched germ line mutations in naevoid basal cell carcinoma syndrome. <i>Human Genetics</i> , 1997, 100, 497-502.	3.8	88
58	Human sequences homologous to the gene for the cochlear protein Ocp-II do not map to currently known non-syndromic hearing loss loci. <i>Annals of Human Genetics</i> , 1996, 60, 385-389.	0.8	2
59	An EST and STS-Based YAC Contig Map of Human Chromosome 9q22.3. <i>Genomics</i> , 1996, 38, 199-205.	2.9	14
60	Yeast artificial chromosome cloning and chromosomal localization of the abundant odontogenic keratocyst protein elafin. <i>Archives of Oral Biology</i> , 1996, 41, 445-452.	1.8	4
61	YAC clones that extend the human Chromosome 12cen-12q15 region contig map. <i>Mammalian Genome</i> , 1996, 7, 780-783.	2.2	2
62	Vectorette PCR isolation of microsatellite repeat sequences using anchored dinucleotide repeat primers. <i>Nucleic Acids Research</i> , 1996, 24, 2190-2191.	14.5	30
63	Characterisation of molecular defects in X-linked amelogenesis imperfecta (AIH1). <i>Human Mutation</i> , 1995, 5, 251-259.	2.5	117
64	(CGG) trinucleotide repeat polymorphism in the 5' region of the HHR6B gene: the human homolog of the yeast DNA repair gene RAD6. <i>Human Genetics</i> , 1995, 96, 369-70.	3.8	2
65	A human ubiquitin conjugating enzyme, L-UBC, maps in the Alzheimer's disease locus on Chromosome 14q24.3. <i>Mammalian Genome</i> , 1995, 6, 725-731.	2.2	17
66	Prostate-specific membrane antigen: evidence for the existence of a second related human gene. <i>British Journal of Cancer</i> , 1995, 72, 583-588.	6.4	74
67	Nance-Horan syndrome: a contiguous gene syndrome involving deletion of the amelogenin gene? A case report and molecular analysis. <i>Oral Diseases</i> , 1995, 1, 8-11.	3.0	6
68	Amelogenesis imperfecta in triplets: a unique family record. <i>British Dental Journal</i> , 1995, 178, 465-468.	0.6	4
69	SSCP detection of a nonsense mutation in exon 5 of the amelogenin gene (AMGX) causing X-linked amelogenesis imperfecta (AIH1). <i>Human Molecular Genetics</i> , 1994, 3, 827-828.	2.9	71
70	Comparative genetic mapping for the identification of novel diagnostic and therapeutic targets. <i>Current Opinion in Biotechnology</i> , 1994, 5, 643-647.	6.6	0
71	Cloning the mouse homolog of the human cystic fibrosis transmembrane conductance regulator gene. <i>Genomics</i> , 1991, 10, 301-307.	2.9	100
72	Aval RFLP detected by the anonymous DNA segment p 10E5.SC1 [D11S806] on chromosome 11q22 - 23. <i>Nucleic Acids Research</i> , 1991, 19, 5796-5796.	14.5	3

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73	ScrfI restriction fragment length polymorphism at the D7S23 locus (probe pKM.19), closely linked to cystic fibrosis. <i>Nucleic Acids Research</i> , 1990, 18, 1318-1318.	14.5	3
74	A new polymorphic locus, D7S411, isolated by cloning from preparative pulse-field gels is close to the mutation causing cystic fibrosis. <i>Genomics</i> , 1990, 6, 39-47.	2.9	15
75	Detection of a rare-cutter RFLP in a CpG-rich island near the cystic fibrosis locus. <i>Human Genetics</i> , 1988, 80, 309-310.	3.8	3
76	An anonymous DNA probe (NL32) recognises a MspI polymorphism on human chromosome 1 [D1s84]. <i>Nucleic Acids Research</i> , 1988, 16, 11854-11854.	14.5	4
77	RFLP for C2/11 (D7S374), a cosmid for chromosome seven. <i>Nucleic Acids Research</i> , 1987, 15, 8121-8121.	14.5	1
78	Progress towards construction of a total restriction fragment map of a human chromosome. <i>Nucleic Acids Research</i> , 1987, 15, 1363-1375.	14.5	33
79	Physical and genetic analysis of cosmids from the vicinity of the cystic fibrosis locus. <i>Nucleic Acids Research</i> , 1987, 15, 3639-3652.	14.5	20
80	A human regulatory subunit of type II cAMP-dependent protein kinase localized by its linkage relationship to several cloned chromosome 7q markers. <i>Cytogenetic and Genome Research</i> , 1987, 45, 237-239.	1.1	9
81	A candidate for the cystic fibrosis locus isolated by selection for methylation-free islands. <i>Nature</i> , 1987, 326, 840-845.	27.8	364