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	Connexin 26 mutations in hereditary non-syndromic sensorineural deafness. Nature, 1997, 387, 80-83.	27.8	1,363
2	Prelingual Deafness: High Prevalence of a 30delG Mutation in the Connexin 26 Gene. Human Molecular Genetics, 1997, 6, 2173-2177.	2.9	601
3	Use of low-density lipoprotein cholesterol gene score to distinguish patients with polygenic and monogenic familial hypercholesterolaemia: a case-control study. Lancet, The, 2013, 381, 1293-1301.	13.7	485
4	Loss-of-function mutations in the cathepsin C gene result in periodontal disease and palmoplantar keratosis. Nature Genetics, 1999, 23, 421-424.	21.4	442
5	A candidate for the cystic fibrosis locus isolated by selection for methylation-free islands. Nature, 1987, 326, 840-845.	27.8	364
6	Positional cloning of a novel gene influencing asthma from Chromosome 2q14. Nature Genetics, 2003, 35, 258-263.	21.4	326
7	Extent and Distribution of Linkage Disequilibrium in Three Genomic Regions. American Journal of Human Genetics, 2001, 68, 191-197.	6.2	325
8	Exome sequencing for prenatal diagnosis of fetuses with sonographic abnormalities. Prenatal Diagnosis, 2015, 35, 1010-1017.	2.3	189
9	Lowâ€Ðensity Lipoprotein Receptor Gene Familial Hypercholesterolemia Variant Database: Update and Pathological Assessment. Annals of Human Genetics, 2012, 76, 387-401.	0.8	173
10	Nonâ€invasive prenatal diagnosis of achondroplasia and thanatophoric dysplasia: nextâ€generation sequencing allows for a safer, more accurate, and comprehensive approach. Prenatal Diagnosis, 2015, 35, 656-662.	2.3	156
11	Primary Autosomal Recessive Microcephaly (MCPH1) Maps to Chromosome 8p22-pter. American Journal of Human Genetics, 1998, 63, 541-546.	6.2	151
12	The clinical implementation of nonâ€invasive prenatal diagnosis for singleâ€gene disorders: challenges and progress made. Prenatal Diagnosis, 2013, 33, 555-562.	2.3	121
13	Characterisation of molecular defects in X-linked amelogenesis imperfecta (AIH1). Human Mutation, 1995, 5, 251-259.	2.5	117
14	CHD2variants are a risk factor for photosensitivity in epilepsy. Brain, 2015, 138, 1198-1208.	7.6	112
15	Connexin-26 mutations in sporadic non-syndromal sensorineural deafness. Lancet, The, 1998, 351, 415.	13.7	109
16	A Third Novel Locus for Primary Autosomal Recessive Microcephaly Maps to Chromosome 9q34. American Journal of Human Genetics, 2000, 66, 724-727.	6.2	105
17	Whole exome sequencing of familial hypercholesterolaemia patients negative forLDLR/APOB/PCSK9mutations. Journal of Medical Genetics, 2014, 51, 537-544.	3.2	104
18	The second locus for autosomal recessive primary microcephaly (MCPH2) maps to chromosome 19q13.1â€“13.2. European Journal of Human Genetics, 1999, 7, 815-820.	2.8	103

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19	Cloning the mouse homolog of the human cystic fibrosis transmembrane conductance regulator gene. Genomics, 1991, 10, 301-307.	2.9	100
20	Analysis of the IBD5 locus and potential gene-gene interactions in Crohn's disease. Gut, 2003, 52, 541-546.	12.1	96
21	Targeted gene panel sequencing in children with very early onset inflammatory bowel diseaseâ€"evaluation and prospective analysis. Journal of Medical Genetics, 2014, 51, 748-755.	3.2	91
22	Characterisation of human patched germ line mutations in naevoid basal cell carcinoma syndrome. Human Genetics, 1997, 100, 497-502.	3.8	88
23	Prostate-specific membrane antigen: evidence for the existence of a second related human gene. British Journal of Cancer, 1995, 72, 583-588.	6.4	74
24	Evaluation of non-invasive prenatal testing (NIPT) for aneuploidy in an NHS setting: a reliable accurate prenatal non-invasive diagnosis (RAPID) protocol. BMC Pregnancy and Childbirth, 2014, 14, 229.	2.4	72
25	SSCP detection of a nonsense mutation in exon 5 of the amelogenin gene (AMGX) causing X-linked amelogenesis imperfecta (AIH1). Human Molecular Genetics, 1994, 3, 827-828.	2.9	71
26	Replication and extension studies of inflammatory bowel disease susceptibility regions confirm linkage to chromosome 6p (IBD3). European Journal of Human Genetics, 2001, 9, 627-633.	2.8	70
27	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	5.1	61
28	Molecular prenatal diagnosis: the impact of modern technologies. Prenatal Diagnosis, 2010, 30, 674-681.	2.3	58
29	Congenital non-syndromal sensorineural hearing impairment due to connexin 26 gene mutations â€" molecular and audiological findings. International Journal of Pediatric Otorhinolaryngology, 1999, 50, 3-13.	1.0	57
30	The IBD6 Crohn's disease locus demonstrates complex interactions with CARD15 and IBD5 disease-associated variants. Human Molecular Genetics, 2003, 12, 2569-2575.	2.9	57
31	Neurological features of epilepsy, ataxia, sensorineural deafness, tubulopathy syndrome. Developmental Medicine and Child Neurology, 2013, 55, 846-856.	2.1	53
32	Inflammatory Bowel Disease Is Linked to 19p13 and Associated with ICAM-1. Inflammatory Bowel Diseases, 2004, 10, 173-181.	1.9	52
33	Detection of a Novel Mutation in X-linked Amelogenesis Imperfecta. Journal of Dental Research, 2000, 79, 1978-1982.	5.2	50
34	A Gene for Autosomal Recessive Symmetrical Spastic Cerebral Palsy Maps to Chromosome 2q24-25. American Journal of Human Genetics, 1999, 64, 526-532.	6.2	46
35	Genetic testing in renal disease. Pediatric Nephrology, 2012, 27, 873-883.	1.7	45
36	A gene for ataxic cerebral palsy maps to chromosome 9p12â€“q12. European Journal of Human Genetics, 2000, 8, 267-272.	2.8	42

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37	Exome Sequencing and Rare Variant Analysis RevealsÂMultiple Filaggrin Mutations in BangladeshiÂFamilies with Atopic Eczema andÂAdditional Risk Genes. Journal of Investigative Dermatology, 2018, 138, 2674-2677.	0.7	37
38	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. Brain, 2016, 139, 2844-2854.	7.6	35
39	KCNJ10 Mutations Display Differential Sensitivity to Heteromerisation with KCNJ16. Nephron Physiology, 2013, 123, 7-14.	1.2	34
40	Progress towards construction of a total restriction fragment map of a human chromosome. Nucleic Acids Research, 1987, 15, 1363-1375.	14.5	33
41	A novel homozygous ERCC5 truncating mutation in a family with prenatal arthrogryposisâ€"Further evidence of genotypeâ€"phenotype correlation. American Journal of Medical Genetics, Part A, 2014, 164, 1777-1783.	1.2	31
42	Vectorette PCR isolation of microsatellite repeat sequences using anchored dinucleotide repeat primers. Nucleic Acids Research, 1996, 24, 2190-2191.	14.5	30
43	Autozygosity Mapping, to Chromosome 11q25, of a Rare Autosomal Recessive Syndrome Causing Histiocytosis, Joint Contractures, and Sensorineural Deafness. American Journal of Human Genetics, 1998, 62, 1123-1128.	6.2	29
44	Sotos syndrome, infantile hypercalcemia, and nephrocalcinosis: a contiguous gene syndrome. Pediatric Nephrology, 2011, 26, 1331-1334.	1.7	29
45	A targeted sequencing panel identifies rare damaging variants in multiple genes in the cranial neural tube defect, anencephaly. Clinical Genetics, 2018, 93, 870-879.	2.0	29
46	Array comparative genomic hybridization: Results from an adult population with drug-resistant epilepsy and co-morbidities. European Journal of Medical Genetics, 2012, 55, 342-348.	1.3	28
47	A novel heterozygous SOX2 mutation causing congenital bilateral anophthalmia, hypogonadotropic hypogonadism and growth hormone deficiency. Gene, 2014, 534, 282-285.	2.2	25
48	Clinico-pathological correlations of congenital and infantile nephrotic syndrome over twenty years. Pediatric Nephrology, 2014, 29, 2173-2180.	1.7	24
49	Uncovering Genomic Causes of Co-Morbidity in Epilepsy: Gene-Driven Phenotypic Characterization of Rare Microdeletions. PLoS ONE, 2011, 6, e23182.	2.5	24
50	DFNB20: a novel locus for autosomal recessive, non-syndromal sensorineural hearing loss maps to chromosome 11q25â€"qter. European Journal of Human Genetics, 1999, 7, 243-246.	2.8	23
51	The future role of genetic screening to detect newborns at risk of childhood-onset hearing loss. International Journal of Audiology, 2013, 52, 124-133.	1.7	23
52	A new locus for autosomal recessive non-syndromal sensorineural hearing impairment (DFNB27) on chromosome 2q23â€"q31. European Journal of Human Genetics, 2000, 8, 991-993.	2.8	21
53	Physical and genetic analysis of cosmids from the vicinity of the cystic fibrosis locus. Nucleic Acids Research, 1987, 15, 3639-3652.	14.5	20
54	Functional analysis of four LDLR 5â€²UTR and promoter variants in patients with familial hypercholesterolaemia. European Journal of Human Genetics, 2015, 23, 790-795.	2.8	18

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55	A human ubiquitin conjugating enzyme, L-UBC, maps in the Alzheimer's disease locus on Chromosome 14q24.3. Mammalian Genome, 1995, 6, 725-731.	2.2	17
56	A new polymorphic locus, D7S411, isolated by cloning from preparative pulse-field gels is close to the mutation causing cystic fibrosis. Genomics, 1990, 6, 39-47.	2.9	15
57	A Syndrome of Severe Mental Retardation, Spasticity, and Tapetoretinal Degeneration Linked to Chromosome 15q24. American Journal of Human Genetics, 1998, 62, 1070-1076.	6.2	15
58	An EST and STS-Based YAC Contig Map of Human Chromosome 9q22.3. Genomics, 1996, 38, 199-205.	2.9	14
59	Assignment of the STAT6 gene (STAT6) to human chromosome band 12q13 by in situ hybridization. Cytogenetic and Genome Research, 1997, 79, 208-209.	1.1	14
60	Confined placental mosaicism: implications for fetal chromosomal analysis using microarray comparative genomic hybridization. Prenatal Diagnosis, 2014, 34, 98-101.	2.3	14
61	DNA diagnosis of X-linked amelogenesis imperfecta (AIH1). Journal of Oral Pathology and Medicine, 1997, 26, 135-137.	2.7	12
62	The human gene encoding FKBP-rapamycin associated protein (FRAP) maps to chromosomal band 1p36.2. Human Genetics, 1997, 99, 547-549.	3.8	12
63	Single-Point Haplotype Scores Telomeric to Human Leukocyte Antigen-C Give a High Susceptibility Major Histocompatability Complex Haplotype for Psoriasis in a Caucasian Population. Journal of Investigative Dermatology, 2005, 124, 545-552.	0.7	11
64	A human regulatory subunit of type II cAMP-dependent protein kinase localized by its linkage relationship to several cloned chromosome 7q markers. Cytogenetic and Genome Research, 1987, 45, 237-239.	1.1	9
65	Favourable response to ketogenic dietary therapies: undiagnosed glucose 1 transporter deficiency syndrome is only one factor. Developmental Medicine and Child Neurology, 2015, 57, 969-976.	2.1	8
66	cDNA Cloning, Genomic Organization, and Chromosomal Localization of a Novel Human Gene That Encodes a Kinesin-Related Protein Highly Similar to Mouse Kif3C. Biochemical and Biophysical Research Communications, 1998, 242, 407-412.	2.1	6
67	Nanceâ€Horan syndrome: a contiguous gene syndrome involving detetion of the ametogenin gene? A case report and molecular analysis. Oral Diseases, 1995, 1, 8-11.	3.0	6
68	Molecular analysis for genetic counselling in amelogenesis imperfecta. Oral Diseases, 2002, 8, 249-253.	3.0	5
69	Evaluation of Real-Time Quantitative PCR as a Standard Cytogenetic Diagnostic Tool for Confirmation of Microarray (aCGH) Results and Determination of Inheritance. Genetic Testing and Molecular Biomarkers, 2013, 17, 821-825.	0.7	5
70	An anonymous DNA probe (NL32) recognises a MspI polymorphism on human chromosome 1 [D1s84]. Nucleic Acids Research, 1988, 16, 11854-11854.	14.5	4
71	Yeast artificial chromosome cloning and chromosomal localization of the abundant odontogenic keratocyst protein elafin. Archives of Oral Biology, 1996, 41, 445-452.	1.8	4
72	Amelogenesis imperfecta in triplets: a unique family record. British Dental Journal, 1995, 178, 465-468.	0.6	4

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73	Detection of a rare-cutter RFLP in a CpG-rich island near the cystic fibrosis locus. Human Genetics, 1988, 80, 309-310.	3.8	3
74	Scrfl restriction fragment length polymorphism at the D7S23 locus (probe pKM.19), closely linked to cystic fibrosis. Nucleic Acids Research, 1990, 18, 1318-1318.	14.5	3
75	Aval RFLP detected by the anonymous DNA segement p 10E5.SC1 [D11S806] on chromosome 11q22 - 23. Nucleic Acids Research, 1991, 19, 5796-5796.	14.5	3
76	(CGG) trinucleotide repeat polymorphism in the 5? region of the HHR6B gene: the human homolog of the yeast DNA repair gene RAD6. Human Genetics, 1995, 96, 369-70.	3.8	2
77	Human sequences homologous to the gene for the cochlear protein Ocp-II do not map to currently known non-syndromic hearing loss loci. Annals of Human Genetics, 1996, 60, 385-389.	0.8	2
78	YAC clones that extend the human Chromosome 12cen-12q15 region contig map. Mammalian Genome, 1996, 7, 780-783.	2.2	2
79	RFLP for C2/11 (D7S374), a cosmid for chromosome seven. Nucleic Acids Research, 1987, 15, 8121-8121.	14.5	1
80	Comparative genetic mapping for the identification of novel diagnostic and therapeutic targets. Current Opinion in Biotechnology, 1994, 5, 643-647.	6.6	0
81	Assignment of the Rab13 gene (RAB13) to human chromosome band 12q13 by in situ hybridization. Cytogenetic and Genome Research, 1997, 79, 210-211.	1.1	0