

Norman Arnheim

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116
papers

22,984
citations

53
h-index

117
g-index

117
ext. papers

24,183
ext. citations

14.8
avg, IF

6.02
L-index

#	Paper	IF	Citations
116	Enzymatic amplification of beta-globin genomic sequences and restriction site analysis for diagnosis of sickle cell anemia. <i>Science</i> , 1985 , 230, 1350-4	33.3	7649
115	Most human carcinomas of the exocrine pancreas contain mutant c-K-ras genes. <i>Cell</i> , 1988 , 53, 549-54	56.2	1839
114	Whole genome amplification from a single cell: implications for genetic analysis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1992 , 89, 5847-51	11.5	783
113	Detection of a specific mitochondrial DNA deletion in tissues of older humans. <i>Nucleic Acids Research</i> , 1990 , 18, 6927-33	20.1	702
112	Involvement of mouse Mlh1 in DNA mismatch repair and meiotic crossing over. <i>Nature Genetics</i> , 1996 , 13, 336-42	36.3	677
111	Detection of human papilloma virus in paraffin-embedded tissue using the polymerase chain reaction. <i>Journal of Experimental Medicine</i> , 1988 , 167, 225-30	16.6	656
110	Amplification and analysis of DNA sequences in single human sperm and diploid cells. <i>Nature</i> , 1988 , 335, 414-7	50.4	622
109	A simple method for site-directed mutagenesis using the polymerase chain reaction. <i>Nucleic Acids Research</i> , 1989 , 17, 6545-51	20.1	582
108	Hydroxylated quantum dots as luminescent probes for in situ hybridization. <i>Journal of the American Chemical Society</i> , 2001 , 123, 4103-4	16.4	580
107	Venezuelan kindreds reveal that genetic and environmental factors modulate Huntington's disease age of onset. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004 , 101, 3498-503	11.5	572
106	A pattern of accumulation of a somatic deletion of mitochondrial DNA in aging human tissues. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1992 , 89, 7370-4	11.5	549
105	Male mice defective in the DNA mismatch repair gene PMS2 exhibit abnormal chromosome synapsis in meiosis. <i>Cell</i> , 1995 , 82, 309-19	56.2	466
104	Polyglutamine-expanded human huntingtin transgenes induce degeneration of Drosophila photoreceptor neurons. <i>Neuron</i> , 1998 , 21, 633-42	13.9	429
103	Molecular evidence for genetic exchanges among ribosomal genes on nonhomologous chromosomes in man and apes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1980 , 77, 7323-7	11.5	377
102	Mosaicism for a specific somatic mitochondrial DNA mutation in adult human brain. <i>Nature Genetics</i> , 1992 , 2, 318-23	36.3	376
101	Length mutations in human mitochondrial DNA: direct sequencing of enzymatically amplified DNA. <i>Nucleic Acids Research</i> , 1987 , 15, 529-42	20.1	336
100	Tumour susceptibility and spontaneous mutation in mice deficient in Mlh1, Pms1 and Pms2 DNA mismatch repair. <i>Nature Genetics</i> , 1998 , 18, 276-9	36.3	307

99	Advancing age has differential effects on DNA damage, chromatin integrity, gene mutations, and aneuploidies in sperm. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006 , 103, 9601-6	11.5	307
98	Extension of base mispairs by Taq DNA polymerase: implications for single nucleotide discrimination in PCR. <i>Nucleic Acids Research</i> , 1992 , 20, 4567-73	20.1	299
97	5Sflanking region of immunoglobulin heavy chain constant region genes displays length heterogeneity in germlines of inbred mouse strains. <i>Cell</i> , 1980 , 22, 187-96	56.2	241
96	The polymerase chain reaction. <i>Trends in Genetics</i> , 1989 , 5, 185-9	8.5	222
95	Taq DNA polymerase slippage mutation rates measured by PCR and quasi-likelihood analysis: (CA/GT) _n and (A/T) _n microsatellites. <i>Nucleic Acids Research</i> , 2003 , 31, 974-80	20.1	182
94	Detection of human papillomavirus in formalin-fixed, invasive squamous carcinomas using the polymerase chain reaction. <i>American Journal of Surgical Pathology</i> , 1989 , 13, 221-4	6.7	180
93	Single sperm analysis of the trinucleotide repeats in the Huntington's disease gene: quantification of the mutation frequency spectrum. <i>Human Molecular Genetics</i> , 1995 , 4, 1519-26	5.6	171
92	Polymerase chain reaction strategy. <i>Annual Review of Biochemistry</i> , 1992 , 61, 131-56	29.1	161
91	Deleterious mitochondrial DNA mutations accumulate in aging human tissues. <i>Mutation Research - DNAging</i> , 1992 , 275, 157-67		157
90	The observed human sperm mutation frequency cannot explain the achondroplasia paternal age effect. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002 , 99, 14952-7	11.5	151
89	Triplet repeat mutation length gains correlate with cell-type specific vulnerability in Huntington disease brain. <i>Human Molecular Genetics</i> , 2007 , 16, 1133-42	5.6	139
88	Heterogeneity of the ribosomal genes in mice and men. <i>Cell</i> , 1977 , 11, 363-70	56.2	131
87	Characterization of mouse ribosomal gene fragments purified by molecular cloning. <i>Gene</i> , 1979 , 7, 83-96	3.8	128
86	New HLA-DPB1 alleles generated by interallelic gene conversion detected by analysis of sperm. <i>Nature Genetics</i> , 1995 , 10, 407-14	36.3	119
85	Rapid prenatal diagnosis of sickle cell anemia by a new method of DNA analysis. <i>New England Journal of Medicine</i> , 1987 , 316, 656-61	59.2	114
84	Differential contributions of mammalian Rad54 paralogs to recombination, DNA damage repair, and meiosis. <i>Molecular and Cellular Biology</i> , 2006 , 26, 976-89	4.8	113
83	Nup50, a nucleoplasmically oriented nucleoporin with a role in nuclear protein export. <i>Molecular and Cellular Biology</i> , 2000 , 20, 5619-30	4.8	102
82	Studying human mutations by sperm typing: instability of CAG trinucleotide repeats in the human androgen receptor gene. <i>Nature Genetics</i> , 1994 , 7, 531-5	36.3	99

81	Stability of intrastrand hairpin structures formed by the CAG/CTG class of DNA triplet repeats associated with neurological diseases. <i>Nucleic Acids Research</i> , 1996 , 24, 1992-8	20.1	98
80	Understanding what determines the frequency and pattern of human germline mutations. <i>Nature Reviews Genetics</i> , 2009 , 10, 478-88	30.1	90
79	Huntington disease expansion mutations in humans can occur before meiosis is completed. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2003 , 100, 8834-8	11.5	87
78	Identification of the in vivo and in vitro origin of transcription in human rDNA. <i>Nucleic Acids Research</i> , 1982 , 10, 3933-49	20.1	86
77	Mammalian meiotic recombination hot spots. <i>Annual Review of Genetics</i> , 2007 , 41, 369-99	14.5	82
76	Contributions by MutL homologues Mlh3 and Pms2 to DNA mismatch repair and tumor suppression in the mouse. <i>Cancer Research</i> , 2005 , 65, 8662-70	10.1	80
75	Quantitative Immunological Comparison of Bird Lysozymes. <i>Journal of Biological Chemistry</i> , 1967 , 242, 3951-3956	5.4	80
74	Hot and cold spots of recombination in the human genome: the reason we should find them and how this can be achieved. <i>American Journal of Human Genetics</i> , 2003 , 73, 5-16	11	78
73	Analysis of germline mutation spectra at the Huntington's disease locus supports a mitotic mutation mechanism. <i>Human Molecular Genetics</i> , 1999 , 8, 173-83	5.6	78
72	High resolution localization of recombination hot spots using sperm typing. <i>Nature Genetics</i> , 1994 , 7, 420-4	36.3	74
71	Mouse rDNA nontranscribed spacer sequences are found flanking immunoglobulin CH genes and elsewhere throughout the genome. <i>Cell</i> , 1980 , 22, 179-85	56.2	70
70	Structure and organization of the highly repeated and interspersed 1.3 kb EcoRI-BglII sequence family in mice. <i>Nucleic Acids Research</i> , 1980 , 8, 5031-42	20.1	67
69	High-resolution recombination patterns in a region of human chromosome 21 measured by sperm typing. <i>PLoS Genetics</i> , 2006 , 2, e70	6	65
68	Application of PCR: Organismal and Population Biology. <i>BioScience</i> , 1990 , 40, 174-182	5.7	65
67	Widespread Distribution of Lysozyme g in Egg White of Birds. <i>Journal of Biological Chemistry</i> , 1974 , 249, 7295-7297	5.4	64
66	A germ-line-selective advantage rather than an increased mutation rate can explain some unexpectedly common human disease mutations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008 , 105, 10143-8	11.5	61
65	Close linkage between bovine prolactin and BoLA-DRB3 genes: genetic mapping in cattle by single sperm typing. <i>Genomics</i> , 1992 , 13, 44-8	4.3	55
64	Nucleotide sequence of the genetically labile repeated elements 5S to the origin of mouse rRNA transcription. <i>Nucleic Acids Research</i> , 1983 , 11, 211-24	20.1	54

63	The molecular anatomy of spontaneous germline mutations in human testes. <i>PLoS Biology</i> , 2007 , 5, e2249.7	5.7	53
62	Genetic analysis using the polymerase chain reaction. <i>Annual Review of Genetics</i> , 1992 , 26, 479-506	14.5	53
61	PCR analysis of DNA sequences in single cells: single sperm gene mapping and genetic disease diagnosis. <i>Genomics</i> , 1990 , 8, 415-9	4.3	51
60	Positive selection for new disease mutations in the human germline: evidence from the heritable cancer syndrome multiple endocrine neoplasia type 2B. <i>PLoS Genetics</i> , 2012 , 8, e1002420	6	45
59	Length heterogeneity in a region of the human ribosomal gene spacer is not accompanied by extensive population polymorphism. <i>Journal of Molecular Biology</i> , 1978 , 126, 91-104	6.5	42
58	Multiple genes for lysozyme in birds. <i>Archives of Biochemistry and Biophysics</i> , 1970 , 141, 656-61	4.1	42
57	Immunochemical resemblance between human leukemia and hen egg-white lysozyme and their reduced carboxymethyl derivatives. <i>Journal of Molecular Biology</i> , 1971 , 61, 237-50	6.5	42
56	The ups and downs of mutation frequencies during aging can account for the Apert syndrome paternal age effect. <i>PLoS Genetics</i> , 2009 , 5, e1000558	6	41
55	Biochemical characterization of "LAP," a polymorphic aminopeptidase from the blue mussel, <i>Mytilus edulis</i> . <i>Biochemical Genetics</i> , 1979 , 17, 305-23	2.4	40
54	Age-dependent germline mosaicism of the most common noonan syndrome mutation shows the signature of germline selection. <i>American Journal of Human Genetics</i> , 2013 , 92, 917-26	11	39
53	Ordering three DNA polymorphisms on human chromosome 3 by sperm typing. <i>Genomics</i> , 1991 , 10, 748-55	4.5	39
52	New evidence for positive selection helps explain the paternal age effect observed in achondroplasia. <i>Human Molecular Genetics</i> , 2013 , 22, 4117-26	5.6	37
51	cDNA cloning and characterization of Npap60: a novel rat nuclear pore-associated protein with an unusual subcellular localization during male germ cell differentiation. <i>Genomics</i> , 1997 , 40, 444-53	4.3	35
50	DNA mismatch repair in mammals: role in disease and meiosis. <i>Current Opinion in Genetics and Development</i> , 1997 , 7, 364-70	4.9	34
49	Non-Darwinian evolution: consequences for neutral allelic variation. <i>Nature</i> , 1969 , 223, 900-3	50.4	34
48	Chemical Studies on the Enzymatic Specificity of Goose Egg White Lysozyme. <i>Journal of Biological Chemistry</i> , 1973 , 248, 233-236	5.4	34
47	Genetic instability induced by overexpression of DNA ligase I in budding yeast. <i>Genetics</i> , 2005 , 171, 427-41	4	31
46	Mouse strains with an active H2-Ea meiotic recombination hot spot exhibit increased levels of H2-Ea-specific DNA breaks in testicular germ cells. <i>Molecular and Cellular Biology</i> , 2004 , 24, 1655-66	4.8	30

45	Candidate DNA replication initiation regions at human trinucleotide repeat disease loci. <i>Human Molecular Genetics</i> , 2003 , 12, 1021-8	5.6	27
44	Detection of human papillomavirus DNA in fine-needle aspirations of metastatic squamous-cell carcinoma of the uterine cervix using the polymerase chain reaction. <i>Diagnostic Cytopathology</i> , 1989 , 5, 40-3	1.4	27
43	Whole genome amplification of single cells: mathematical analysis of PEP and tagged PCR. <i>Nucleic Acids Research</i> , 1995 , 23, 3034-40	20.1	26
42	Gene-centromere linkage mapping by PCR analysis of individual oocytes. <i>Genomics</i> , 1992 , 13, 713-7	4.3	24
41	Detection and quantification of mitochondrial DNA deletions. <i>Methods in Enzymology</i> , 1996 , 264, 421-31	1.7	23
40	Germline Stem Cell Competition, Mutation Hot Spots, Genetic Disorders, and Older Fathers. <i>Annual Review of Genomics and Human Genetics</i> , 2016 , 17, 219-43	9.7	23
39	Gene hunting without sequencing genomic clones: finding exon boundaries in cDNAs. <i>Genomics</i> , 1998 , 47, 171-9	4.3	22
38	Analysis of mutational changes at the HLA locus in single human sperm. <i>Human Mutation</i> , 1995 , 6, 303-10	1.7	21
37	Sperm typing allows accurate measurement of the recombination fraction between D3S2 and D3S3 on the short arm of human chromosome 3. <i>Genomics</i> , 1992 , 12, 683-7	4.3	20
36	Using the polymerase chain reaction to estimate mutation frequencies and rates in human cells. <i>Mutation Research - Reviews in Genetic Toxicology</i> , 1992 , 277, 239-49		19
35	A Novel Method for the Detection of Polymorphic Restriction Sites by Cleavage of Oligonucleotide Probes: Application to Sickle-Cell Anemia. <i>Nature Biotechnology</i> , 1985 , 3, 1008-1012	44.5	19
34	Polymorphisms in the human DNA repair gene XPF. <i>Mutation Research - Mutation Research Genomics</i> , 1999 , 406, 115-20		14
33	The Evolution of Proteins 1973 , 377-416		14
32	The mutation process of microsatellites during the polymerase chain reaction. <i>Journal of Computational Biology</i> , 2003 , 10, 143-55	1.7	13
31	Product length, dye choice, and detection chemistry in the bead-emulsion amplification of millions of single DNA molecules in parallel. <i>Analytical Chemistry</i> , 2009 , 81, 5770-6	7.8	12
30	Eliminating primers from completed polymerase chain reactions with exonuclease VII. <i>Nucleic Acids Research</i> , 1991 , 19, 3139-41	20.1	10
29	Combining sperm typing and linkage disequilibrium analyses reveals differences in selective pressures or recombination rates across human populations. <i>Genetics</i> , 2007 , 175, 795-804	4	9
28	Single-molecule analysis of the hypermutable tetranucleotide repeat locus D21S1245 through sperm genotyping: a heterogeneous pattern of mutation but no clear male age effect. <i>Molecular Biology and Evolution</i> , 2004 , 21, 58-64	8.3	9

27	Meiotic segregation analysis of RB1 alleles in retinoblastoma pedigrees by use of single-sperm typing. <i>American Journal of Human Genetics</i> , 2000 , 66, 167-75	11	9
26	Patchy fur, a mouse coat mutation associated with X-Y nondisjunction, maps to the pseudoautosomal boundary region. <i>Genomics</i> , 1998 , 54, 556-9	4.3	9
25	The mouse Eb meiotic recombination hotspot contains a tissue-specific transcriptional enhancer. <i>Immunogenetics</i> , 1993 , 37, 331-6	3.2	9
24	Estimating Exceptionally Rare Germline and Somatic Mutation Frequencies via Next Generation Sequencing. <i>PLoS ONE</i> , 2016 , 11, e0158340	3.7	7
23	[8] Quantitative PCR: Analysis of rare mitochondrial DNA mutations in central nervous system tissues. <i>Methods in Neurosciences</i> , 1995 , 105-128		6
22	Preimplantation genetic diagnosis--a rolling stone gathers no moss!. <i>Human Reproduction</i> , 1992 , 7, 1481-5	5.7	6
21	Direct estimation of the recombination frequency between the RB1 gene and two closely linked microsatellites using sperm typing. <i>European Journal of Human Genetics</i> , 1999 , 7, 239-42	5.3	4
20	Single-sperm typing. <i>Current Protocols in Human Genetics</i> , 2002 , Chapter 1, Unit 1.6	3.2	3
19	Cloning and identification of the pig ribosomal gene promoter. <i>Gene</i> , 1994 , 150, 375-9	3.8	3
18	Genetic mapping and preimplantation diagnosis in agriculturally important species using single cell PCR. <i>Animal Biotechnology</i> , 1992 , 3, 55-65	1.4	3
17	Multiple Genes for Lysozyme 1974 , 153-161		3
16	A New Approach to Constructing Genetic Maps: PCR Analysis of DNA Sequences in Individual Gametes 1989 , 119-135		3
15	Combining Data from Polymerase Chain Reaction DNA Typing Experiments: Applications to Sperm Typing Data. <i>Journal of the American Statistical Association</i> , 1999 , 94, 726-733	2.8	2
14	Using PCR in preimplantation genetic disease diagnosis. <i>Human Reproduction</i> , 1992 , 7, 288	5.7	2
13	Preparation of specific antisera to Drosophila acid phosphatase without rigorous protein purification. <i>Biochemical Genetics</i> , 1976 , 14, 237-43	2.4	2
12	Preliminary Biochemical Studies of the Lysozymes of the Black Swan, <i>Cygnus atratus</i> 1974 , 81-88		2
11	THE EVOLUTION OF REGULATORY MECHANISMS STUDIES ON THE MULTIPLE GENES FOR LYSOZYME 1975 , 623-632		2
10	A small molecule in hagfish tissues, possibly related to the cardiac agent, eptatretin. <i>Comparative Biochemistry and Physiology</i> , 1968 , 25, 359-62		1

- 9 Single Sperm PCR Analysis [Implications for Preimplantation Genetic Disease Diagnosis **1991**, 121-130 1
- 8 New technologies for studying human genetic variation. *Basic Life Sciences*, **1988**, 43, 37-44 1
- 7 The polymerase chain reaction. *Genetic Engineering*, **1990**, 12, 115-37 0
- 6 Dating PCR. *Nature Biotechnology*, **1990**, 8, 357 44.5
- 5 Determination of RFLPs linked to multiple sclerosis susceptibility. *Annals of the New York Academy of Sciences*, **1988**, 540, 269-70 6.5
- 4 Studying Germline Genetic Events in Single Individuals. *Stadler Genetics Symposia Series*, **2000**, 71-80
- 3 Frequency of Human Disease Mutations and Spermatogonial Stem Cell Function **2017**, 181-204
- 2 Detection of meiotic DNA breaks in mouse testicular germ cells. *Methods in Molecular Biology*, **2009**, 557, 165-81 1.4
- 1 THE EVOLUTION OF TRANSCRIPTIONAL CONTROL SIGNALS: COEVOLUTION OF RIBOSOMAL GENE PROMOTER SEQUENCES AND TRANSCRIPTION FACTORS **1986**, 37-51