Norman Arnheim

List of Publications by Year in Descending Order

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	Frequency of Human Disease Mutations and Spermatogonial Stem Cell Function **2017**, 181-204		
115	Estimating Exceptionally Rare Germline and Somatic Mutation Frequencies via Next Generation Sequencing. *PLoS ONE*, **2016**, 11, e0158340	3.7	7
114	Germline Stem Cell Competition, Mutation Hot Spots, Genetic Disorders, and Older Fathers. *Annual Review of Genomics and Human Genetics*, **2016**, 17, 219-43	9.7	23
113	Age-dependent germline mosaicism of the most common noonan syndrome mutation shows the signature of germline selection. *American Journal of Human Genetics*, **2013**, 92, 917-26	11	39
112	New evidence for positive selection helps explain the paternal age effect observed in achondroplasia. *Human Molecular Genetics*, **2013**, 22, 4117-26	5.6	37
111	Positive selection for new disease mutations in the human germline: evidence from the heritable cancer syndrome multiple endocrine neoplasia type 2B. *PLoS Genetics*, **2012**, 8, e1002420	6	45
110	The ups and downs of mutation frequencies during aging can account for the Apert syndrome paternal age effect. *PLoS Genetics*, **2009**, 5, e1000558	6	41
109	Understanding what determines the frequency and pattern of human germline mutations. *Nature Reviews Genetics*, **2009**, 10, 478-88	30.1	90
108	Product length, dye choice, and detection chemistry in the bead-emulsion amplification of millions of single DNA molecules in parallel. *Analytical Chemistry*, **2009**, 81, 5770-6	7.8	12
107	Detection of meiotic DNA breaks in mouse testicular germ cells. *Methods in Molecular Biology*, **2009** , 557, 165-81	1.4	
106	A germ-line-selective advantage rather than an increased mutation rate can explain some unexpectedly common human disease mutations. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 10143-8	11.5	61
105	Combining sperm typing and linkage disequilibrium analyses reveals differences in selective pressures or recombination rates across human populations. *Genetics*, **2007**, 175, 795-804	4	9
104	The molecular anatomy of spontaneous germline mutations in human testes. *PLoS Biology*, **2007**, 5, e224	9.7	53
103	Triplet repeat mutation length gains correlate with cell-type specific vulnerability in Huntington disease brain. *Human Molecular Genetics*, **2007**, 16, 1133-42	5.6	139
102	Mammalian meiotic recombination hot spots. *Annual Review of Genetics*, **2007**, 41, 369-99	14.5	82
101	High-resolution recombination patterns in a region of human chromosome 21 measured by sperm typing. *PLoS Genetics*, **2006**, 2, e70	6	65
100	Advancing age has differential effects on DNA damage, chromatin integrity, gene mutations, and aneuploidies in sperm. *Proceedings of the National Academy of Sciences of the United States of America*, **2006**, 103, 9601-6	11.5	307

99	Differential contributions of mammalian Rad54 paralogs to recombination, DNA damage repair, and meiosis. *Molecular and Cellular Biology*, **2006**, 26, 976-89	4.8	113
98	Genetic instability induced by overexpression of DNA ligase I in budding yeast. *Genetics*, **2005**, 171, 427-41	4	31
97	Contributions by MutL homologues Mlh3 and Pms2 to DNA mismatch repair and tumor suppression in the mouse. *Cancer Research*, **2005**, 65, 8662-70	10.1	80
96	Mouse strains with an active H2-Ea meiotic recombination hot spot exhibit increased levels of H2-Ea-specific DNA breaks in testicular germ cells. *Molecular and Cellular Biology*, **2004**, 24, 1655-66	4.8	30
95	Venezuelan kindreds reveal that genetic and environmental factors modulate Huntington's disease age of onset. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 3498-503	11.5	572
94	Single-molecule analysis of the hypermutable tetranucleotide repeat locus D21S1245 through sperm genotyping: a heterogeneous pattern of mutation but no clear male age effect. *Molecular Biology and Evolution*, **2004**, 21, 58-64	8.3	9
93	Taq DNA polymerase slippage mutation rates measured by PCR and quasi-likelihood analysis: (CA/GT)n and (A/T)n microsatellites. *Nucleic Acids Research*, **2003**, 31, 974-80	20.1	182
92	The mutation process of microsatellites during the polymerase chain reaction. *Journal of Computational Biology*, **2003**, 10, 143-55	1.7	13
91	Hot and cold spots of recombination in the human genome: the reason we should find them and how this can be achieved. *American Journal of Human Genetics*, **2003**, 73, 5-16	11	78
90	Candidate DNA replication initiation regions at human trinucleotide repeat disease loci. *Human Molecular Genetics*, **2003**, 12, 1021-8	5.6	27
89	Huntington disease expansion mutations in humans can occur before meiosis is completed. *Proceedings of the National Academy of Sciences of the United States of America*, **2003**, 100, 8834-8	11.5	87
88	The observed human sperm mutation frequency cannot explain the achondroplasia paternal age effect. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 14952-7	11.5	151
87	Single-sperm typing. *Current Protocols in Human Genetics*, **2002**, Chapter 1, Unit 1.6	3.2	3
86	Hydroxylated quantum dots as luminescent probes for in situ hybridization. *Journal of the American Chemical Society*, **2001**, 123, 4103-4	16.4	580
85	Nup50, a nucleoplasmically oriented nucleoporin with a role in nuclear protein export. *Molecular and Cellular Biology*, **2000**, 20, 5619-30	4.8	102
84	Meiotic segregation analysis of RB1 alleles in retinoblastoma pedigrees by use of single-sperm typing. *American Journal of Human Genetics*, **2000**, 66, 167-75	11	9
83	Studying Germline Genetic Events in Single Individuals. *Stadler Genetics Symposia Series*, **2000**, 71-80		
82	Analysis of germline mutation spectra at the Huntington's disease locus supports a mitotic mutation mechanism. *Human Molecular Genetics*, **1999**, 8, 173-83	5.6	78

81	Direct estimation of the recombination frequency between the RB1 gene and two closely linked microsatellites using sperm typing. *European Journal of Human Genetics*, **1999**, 7, 239-42	5.3	4
80	Combining Data from Polymerase Chain Reaction DNA Typing Experiments: Applications to Sperm Typing Data. *Journal of the American Statistical Association*, **1999**, 94, 726-733	2.8	2
79	Polymorphisms in the human DNA repair gene XPF. *Mutation Research - Mutation Research Genomics*, **1999**, 406, 115-20		14
78	Tumour susceptibility and spontaneous mutation in mice deficient in Mlh1, Pms1 and Pms2 DNA mismatch repair. *Nature Genetics*, **1998**, 18, 276-9	36.3	307
77	Polyglutamine-expanded human huntingtin transgenes induce degeneration of Drosophila photoreceptor neurons. *Neuron*, **1998**, 21, 633-42	13.9	429
76	Gene hunting without sequencing genomic clones: finding exon boundaries in cDNAs. *Genomics*, **1998**, 47, 171-9	4.3	22
75	Patchy fur, a mouse coat mutation associated with X-Y nondisjunction, maps to the pseudoautosomal boundary region. *Genomics*, **1998**, 54, 556-9	4.3	9
74	cDNA cloning and characterization of Npap60: a novel rat nuclear pore-associated protein with an unusual subcellular localization during male germ cell differentiation. *Genomics*, **1997**, 40, 444-53	4.3	35
73	DNA mismatch repair in mammals: role in disease and meiosis. *Current Opinion in Genetics and Development*, **1997**, 7, 364-70	4.9	34
72	Detection and quantification of mitochondrial DNA deletions. *Methods in Enzymology*, **1996**, 264, 421-31	1.7	23
71	Stability of intrastrand hairpin structures formed by the CAG/CTG class of DNA triplet repeats associated with neurological diseases. *Nucleic Acids Research*, **1996**, 24, 1992-8	20.1	98
70	Involvement of mouse Mlh1 in DNA mismatch repair and meiotic crossing over. *Nature Genetics*, **1996**, 13, 336-42	36.3	677
69	New HLA-DPB1 alleles generated by interallelic gene conversion detected by analysis of sperm. *Nature Genetics*, **1995**, 10, 407-14	36.3	119
68	Whole genome amplification of single cells: mathematical analysis of PEP and tagged PCR. *Nucleic Acids Research*, **1995**, 23, 3034-40	20.1	26
67	Male mice defective in the DNA mismatch repair gene PMS2 exhibit abnormal chromosome synapsis in meiosis. *Cell*, **1995**, 82, 309-19	56.2	466
66	Single sperm analysis of the trinucleotide repeats in the Huntington's disease gene: quantification of the mutation frequency spectrum. *Human Molecular Genetics*, **1995**, 4, 1519-26	5.6	171
65	[8] Quantitative PCR: Analysis of rare mitochondrial DNA mutations in central nervous system tissues. *Methods in Neurosciences*, **1995**, 105-128		6
64	Analysis of mutational changes at the HLA locus in single human sperm. *Human Mutation*, **1995**, 6, 303-10	4.7	21

63	High resolution localization of recombination hot spots using sperm typing. *Nature Genetics*, **1994**, 7, 420-4	36.3	74
62	Studying human mutations by sperm typing: instability of CAG trinucleotide repeats in the human androgen receptor gene. *Nature Genetics*, **1994**, 7, 531-5	36.3	99
61	Cloning and identification of the pig ribosomal gene promoter. *Gene*, **1994**, 150, 375-9	3.8	3
60	The mouse Eb meiotic recombination hotspot contains a tissue-specific transcriptional enhancer. *Immunogenetics*, **1993**, 37, 331-6	3.2	9
59	Genetic happing and preimplantation diagnosis in agriculturally important species using single cell PCR. *Animal Biotechnology*, **1992**, 3, 55-65	1.4	3
58	A pattern of accumulation of a somatic deletion of mitochondrial DNA in aging human tissues. *Proceedings of the National Academy of Sciences of the United States of America*, **1992**, 89, 7370-4	11.5	549
57	Whole genome amplification from a single cell: implications for genetic analysis. *Proceedings of the National Academy of Sciences of the United States of America*, **1992**, 89, 5847-51	11.5	783
56	Deleterious mitochondrial DNA mutations accumulate in aging human tissues. *Mutation Research - DNAging*, **1992**, 275, 157-67		157
55	Genetic analysis using the polymerase chain reaction. *Annual Review of Genetics*, **1992**, 26, 479-506	14.5	53
54	Polymerase chain reaction strategy. *Annual Review of Biochemistry*, **1992**, 61, 131-56	29.1	161
53	Sperm typing allows accurate measurement of the recombination fraction between D3S2 and D3S3 on the short arm of human chromosome 3. *Genomics*, **1992**, 12, 683-7	4.3	20
52	Close linkage between bovine prolactin and BoLA-DRB3 genes: genetic mapping in cattle by single sperm typing. *Genomics*, **1992**, 13, 44-8	4.3	55
51	Extension of base mispairs by Taq DNA polymerase: implications for single nucleotide discrimination in PCR. *Nucleic Acids Research*, **1992**, 20, 4567-73	20.1	299
50	Gene-centromere linkage mapping by PCR analysis of individual oocytes. *Genomics*, **1992**, 13, 713-7	4.3	24
49	Using PCR in preimplantation genetic disease diagnosis. *Human Reproduction*, **1992**, 7, 288	5.7	2
48	Preimplantation genetic diagnosis--a rolling stone gathers no moss!. *Human Reproduction*, **1992**, 7, 1481	5.7	6
47	Mosaicism for a specific somatic mitochondrial DNA mutation in adult human brain. *Nature Genetics* , **1992**, 2, 318-23	36.3	376
46	Using the polymerase chain reaction to estimate mutation frequencies and rates in human cells. *Mutation Research - Reviews in Genetic Toxicology*, **1992**, 277, 239-49		19

45	Eliminating primers from completed polymerase chain reactions with exonuclease VII. *Nucleic Acids Research*, **1991**, 19, 3139-41	20.1	10
44	Ordering three DNA polymorphisms on human chromosome 3 by sperm typing. *Genomics*, **1991**, 10, 748-55	4.3	39
43	Single Sperm PCR Analysis Implications for Preimplantation Genetic Disease Diagnosis **1991**, 121-130		1
42	Dating PCR. *Nature Biotechnology*, **1990**, 8, 357	44.5	
41	Detection of a specific mitochondrial DNA deletion in tissues of older humans. *Nucleic Acids Research*, **1990**, 18, 6927-33	20.1	702
40	Application of PCR: Organismal and Population Biology. *BioScience*, **1990**, 40, 174-182	5.7	65
39	PCR analysis of DNA sequences in single cells: single sperm gene mapping and genetic disease diagnosis. *Genomics*, **1990**, 8, 415-9	4.3	51
38	The polymerase chain reaction. *Genetic Engineering*, **1990**, 12, 115-37		0
37	The polymerase chain reaction. *Trends in Genetics*, **1989**, 5, 185-9	8.5	222
36	Detection of human papillomavirus DNA in fine-needle aspirations of metastatic squamous-cell carcinoma of the uterine cervix using the polymerase chain reaction. *Diagnostic Cytopathology*, **1989**, 5, 40-3	1.4	27
35	A simple method for site-directed mutagenesis using the polymerase chain reaction. *Nucleic Acids Research*, **1989**, 17, 6545-51	20.1	582
34	Detection of human papillomavirus in formalin-fixed, invasive squamous carcinomas using the polymerase chain reaction. *American Journal of Surgical Pathology*, **1989**, 13, 221-4	6.7	180
33	A New Approach to Constructing Genetic Maps: PCR Analysis of DNA Sequences in Individual Gametes **1989**, 119-135		3
32	Amplification and analysis of DNA sequences in single human sperm and diploid cells. *Nature*, **1988**, 335, 414-7	50.4	622
31	Most human carcinomas of the exocrine pancreas contain mutant c-K-ras genes. *Cell*, **1988**, 53, 549-54	56.2	1839
30	Determination of RFLPs linked to multiple sclerosis susceptibility. *Annals of the New York Academy of Sciences*, **1988**, 540, 269-70	6.5	
29	Detection of human papilloma virus in paraffin-embedded tissue using the polymerase chain reaction. *Journal of Experimental Medicine*, **1988**, 167, 225-30	16.6	656
28	New technologies for studying human genetic variation. *Basic Life Sciences*, **1988**, 43, 37-44		1

27	Rapid prenatal diagnosis of sickle cell anemia by a new method of DNA analysis. *New England Journal of Medicine*, **1987**, 316, 656-61	59.2	114
26	Length mutations in human mitochondrial DNA: direct sequencing of enzymatically amplified DNA. *Nucleic Acids Research*, **1987**, 15, 529-42	20.1	336
25	THE EVOLUTION OF TRANSCRIPTIONAL CONTROL SIGNALS: COEVOLUTION OF RIBOSOMAL GENE PROMOTER SEQUENCES AND TRANSCRIPTION FACTORS **1986**, 37-51		
24	Enzymatic amplification of beta-globin genomic sequences and restriction site analysis for diagnosis of sickle cell anemia. *Science*, **1985**, 230, 1350-4	33.3	7649
23	A Novel Method for the Detection of Polymorphic Restriction Sites by Cleavage of Oligonucleotide Probes: Application to Sickle-Cell Anemia. *Nature Biotechnology*, **1985**, 3, 1008-1012	44.5	19
22	Nucleotide sequence of the genetically labile repeated elements 5Sto the origin of mouse rRNA transcription. *Nucleic Acids Research*, **1983**, 11, 211-24	20.1	54
21	Identification of the in vivo and in vitro origin of transcription in human rDNA. *Nucleic Acids Research*, **1982**, 10, 3933-49	20.1	86
20	Molecular evidence for genetic exchanges among ribosomal genes on nonhomologous chromosomes in man and apes. *Proceedings of the National Academy of Sciences of the United States of America*, **1980**, 77, 7323-7	11.5	377
19	Structure and organization of the highly repeated and interspersed 1.3 kb EcoRI-Bg1II sequence family in mice. *Nucleic Acids Research*, **1980**, 8, 5031-42	20.1	67
18	5Sflanking region of immunoglobulin heavy chain constant region genes displays length heterogeneity in germlines of inbred mouse strains. *Cell*, **1980**, 22, 187-96	56.2	241
17	Mouse rDNA nontranscribed spacer sequences are found flanking immunoglobulin CH genes and elsewhere throughout the genome. *Cell*, **1980**, 22, 179-85	56.2	70
16	Biochemical characterization of "LAP," a polymorphic aminopeptidase from the blue mussel, Mytilus edulis. *Biochemical Genetics*, **1979**, 17, 305-23	2.4	40
15	Characterization of mouse ribosomal gene fragments purified by molecular cloning. *Gene*, **1979**, 7, 83-96	3.8	128
14	Length heterogeneity in a region of the human ribosomal gene spacer is not accompanied by extensive population polymorphism. *Journal of Molecular Biology*, **1978**, 126, 91-104	6.5	42
13	Heterogeneity of the ribosomal genes in mice and men. *Cell*, **1977**, 11, 363-70	56.2	131
12	Preparation of specific antisera to Drosophila acid phosphatase without rigorous protein purification. *Biochemical Genetics*, **1976**, 14, 237-43	2.4	2
11	THE EVOLUTION OF REGULATORY MECHANISMS STUDIES ON THE MULTIPLE GENES FOR LYSOZYME **1975**, 623-632		2
10	Preliminary Biochemical Studies of the Lysozymes of the Black Swan, Cygnus atratus **1974**, 81-88		2

9	Multiple Genes for Lysozyme **1974**, 153-161		3
8	Widespread Distribution of Lysozyme g in Egg White of Birds. *Journal of Biological Chemistry*, **1974**, 249, 7295-7297	5.4	64
7	The Evolution of Proteins **1973**, 377-416		14
6	Chemical Studies on the Enzymatic Specificity of Goose Egg White Lysozyme. *Journal of Biological Chemistry*, **1973**, 248, 233-236	5.4	34
5	Immunochemical resemblance between human leukemia and hen egg-ehite lysozyme and their reduced carboxymethyl derivatives. *Journal of Molecular Biology*, **1971**, 61, 237-50	6.5	42
4	Multiple genes for lysozyme in birds. *Archives of Biochemistry and Biophysics*, **1970**, 141, 656-61	4.1	42
3	Non-Darwinian evolution: consequences for neutral allelic variation. *Nature*, **1969**, 223, 900-3	50.4	34
2	A small molecule in hagfish tissues, possibly related to the cardiac agent, eptatretin. *Comparative Biochemistry and Physiology*, **1968**, 25, 359-62		1
1	Quantitative Immunological Comparison of Bird Lysozymes. *Journal of Biological Chemistry*, **1967**, 242, 3951-3956	5.4	80