

Nadia Chuzhanova

List of Publications by Year in descending order

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65
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156536

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#	ARTICLE	IF	CITATIONS
1	At Least 1 in 20 16S rRNA Sequence Records Currently Held in Public Repositories Is Estimated To Contain Substantial Anomalies. <i>Applied and Environmental Microbiology</i> , 2005, 71, 7724-7736.	1.4	716
2	New Screening Software Shows that Most Recent Large 16S rRNA Gene Clone Libraries Contain Chimeras. <i>Applied and Environmental Microbiology</i> , 2006, 72, 5734-5741.	1.4	621
3	Gene conversion: mechanisms, evolution and human disease. <i>Nature Reviews Genetics</i> , 2007, 8, 762-775.	7.7	576
4	A meta-analysis of nonsense mutations causing human genetic disease. <i>Human Mutation</i> , 2008, 29, 1037-1047.	1.1	348
5	An Absence of Cutaneous Neurofibromas Associated with a 3-bp Inframe Deletion in Exon 17 of the NF1 Gene (c.2970-2972 delAAT): Evidence of a Clinically Significant NF1 Genotype-Phenotype Correlation. <i>American Journal of Human Genetics</i> , 2007, 80, 140-151.	2.6	335
6	Translocation and gross deletion breakpoints in human inherited disease and cancer I: Nucleotide composition and recombination-associated motifs. <i>Human Mutation</i> , 2003, 22, 229-244.	1.1	214
7	Gains of glycosylation comprise an unexpectedly large group of pathogenic mutations. <i>Nature Genetics</i> , 2005, 37, 692-700.	9.4	198
8	Breakpoints of gross deletions coincide with non-B DNA conformations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 14162-14167.	3.3	184
9	Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. <i>Human Mutation</i> , 2010, 31, 631-655.	1.1	161
10	Meta-analysis of gross insertions causing human genetic disease: Novel mutational mechanisms and the role of replication slippage. <i>Human Mutation</i> , 2005, 25, 207-221.	1.1	148
11	Microdeletions and microinsertions causing human genetic disease: common mechanisms of mutagenesis and the role of local DNA sequence complexity. <i>Human Mutation</i> , 2005, 26, 205-213.	1.1	136
12	Comparative Analysis of Genome Sequences Covering the Seven <i>Cronobacter</i> Species. <i>PLoS ONE</i> , 2012, 7, e49455.	1.1	130
13	High-Resolution DNA Copy Number Profiling of Malignant Peripheral Nerve Sheath Tumors Using Targeted Microarray-Based Comparative Genomic Hybridization. <i>Clinical Cancer Research</i> , 2008, 14, 1015-1024.	3.2	119
14	Genomic rearrangements in the CFTR gene: Extensive allelic heterogeneity and diverse mutational mechanisms. <i>Human Mutation</i> , 2004, 23, 343-357.	1.1	115
15	Meta-analysis of indels causing human genetic disease: mechanisms of mutagenesis and the role of local DNA sequence complexity. <i>Human Mutation</i> , 2003, 21, 28-44.	1.1	112
16	Germline and somatic NF1 gene mutation spectrum in NF1-associated malignant peripheral nerve sheath tumors (MPNSTs). <i>Human Mutation</i> , 2008, 29, 74-82.	1.1	106
17	Translocation and gross deletion breakpoints in human inherited disease and cancer II: Potential involvement of repetitive sequence elements in secondary structure formation between DNA ends. <i>Human Mutation</i> , 2003, 22, 245-251.	1.1	98
18	Complex gene rearrangements caused by serial replication slippage. <i>Human Mutation</i> , 2005, 26, 125-134.	1.1	88

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19	SPRED1 mutations (Legius syndrome): another clinically useful genotype for dissecting the neurofibromatosis type 1 phenotype. <i>Journal of Medical Genetics</i> , 2009, 46, 431-437.	1.5	83
20	Gross genomic rearrangements involving deletions in the CFTR gene: characterization of six new events from a large cohort of hitherto unidentified cystic fibrosis chromosomes and meta-analysis of the underlying mechanisms. <i>European Journal of Human Genetics</i> , 2006, 14, 567-576.	1.4	77
21	Gain-of-glycosylation mutations. <i>Current Opinion in Genetics and Development</i> , 2007, 17, 245-251.	1.5	65
22	Gene conversion causing human inherited disease: Evidence for involvement of non-B-DNA-forming sequences and recombination-promoting motifs in DNA breakage and repair. <i>Human Mutation</i> , 2009, 30, 1189-1198.	1.1	63
23	Intrachromosomal serial replication slippage intrinsically gives rise to diverse genomic rearrangements involving inversions. <i>Human Mutation</i> , 2005, 26, 362-373.	1.1	62
24	The spectrum of somatic and germline NF1 mutations in NF1 patients with spinal neurofibromas. <i>Neurogenetics</i> , 2009, 10, 251-263.	0.7	61
25	Long homopurine*homopyrimidine sequences are characteristic of genes expressed in brain and the pseudoautosomal region. <i>Nucleic Acids Research</i> , 2006, 34, 2663-2675.	6.5	60
26	Type 2 NF1 Deletions Are Highly Unusual by Virtue of the Absence of Nonallelic Homologous Recombination Hotspots and an Apparent Preference for Female Mitotic Recombination. <i>American Journal of Human Genetics</i> , 2007, 81, 1201-1220.	2.6	60
27	Genome-wide high-resolution analysis of DNA copy number alterations in NF1-associated malignant peripheral nerve sheath tumors using 32K BAC array. <i>Genes Chromosomes and Cancer</i> , 2009, 48, 897-907.	1.5	50
28	Breakpoint analysis of the pericentric inversion distinguishing human chromosome 4 from the homologous chromosome in the chimpanzee (<i>Pan troglodytes</i>). <i>Human Mutation</i> , 2005, 25, 45-55.	1.1	47
29	Independent intrachromosomal recombination events underlie the pericentric inversions of chimpanzee and gorilla chromosomes homologous to human chromosome 16. <i>Genome Research</i> , 2005, 15, 1232-1242.	2.4	42
30	A novel third type of recurrent NF1 microdeletion mediated by nonallelic homologous recombination between LRRC37B-containing low-copy repeats in 17q11.2. <i>Human Mutation</i> , 2010, 31, 742-751.	1.1	42
31	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase α - and β -subunit (<i>GNPTAB</i>) gene mutations causing mucopolidiosis types II α and III α in 46 patients. <i>Human Mutation</i> , 2009, 30, E956-E973.	1.1	38
32	Intrachromosomal mitotic nonallelic homologous recombination is the major molecular mechanism underlying type-2 NF1 deletions. <i>Human Mutation</i> , 2010, 31, 1163-1173.	1.1	36
33	Evolution of the proximal promoter region of the mammalian growth hormone gene. <i>Gene</i> , 1999, 237, 143-151.	1.0	34
34	Cruciform-forming inverted repeats appear to have mediated many of the microinversions that distinguish the human and chimpanzee genomes. <i>Chromosome Research</i> , 2009, 17, 469-483.	1.0	31
35	Complete ascertainment of intragenic copy number mutations (CNMs) in the CFTR gene and its implications for CNM formation at other autosomal loci. <i>Human Mutation</i> , 2010, 31, 421-428.	1.1	31
36	Non-B DNA-forming Sequences and WRN Deficiency Independently Increase the Frequency of Base Substitution in Human Cells. <i>Journal of Biological Chemistry</i> , 2011, 286, 10017-10026.	1.6	31

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37	Characterization of the nonallelic homologous recombination hotspot PRS3 associated with type-3 <i>NF1</i> deletions. <i>Human Mutation</i> , 2012, 33, 372-383.	1.1	28
38	Molecular characterisation of the pericentric inversion that distinguishes human chromosome 5 from the homologous chimpanzee chromosome. <i>Human Genetics</i> , 2005, 117, 168-176.	1.8	27
39	Identification and molecular characterization of six novel mutations in the UDP-N-acetylglucosamine-1-phosphotransferase gamma subunit (GNPTG) gene in patients with mucopolipidosis III gamma. <i>Human Mutation</i> , 2009, 30, 978-984.	1.1	26
40	Identification of recurrent type-2 <i>NF1</i> microdeletions reveals a mitotic nonallelic homologous recombination hotspot underlying a human genomic disorder. <i>Human Mutation</i> , 2012, 33, 1599-1609.	1.1	26
41	Co-inheritance of a novel deletion of the entire SPINK1 gene with a CFTR missense mutation (L997F) in a family with chronic pancreatitis. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 168-175.	0.5	25
42	Analysis of NF1 somatic mutations in cutaneous neurofibromas from patients with high tumor burden. <i>Neurogenetics</i> , 2010, 11, 391-400.	0.7	25
43	Exploring the somatic NF1 mutational spectrum associated with NF1 cutaneous neurofibromas. <i>European Journal of Human Genetics</i> , 2012, 20, 411-419.	1.4	25
44	A Role for Non-B DNA Forming Sequences in Mediating Microlesions Causing Human Inherited Disease. <i>Human Mutation</i> , 2016, 37, 65-73.	1.1	22
45	Identification of an intronic regulatory element in the human protein C (PROC) gene. <i>Human Genetics</i> , 2000, 107, 458-465.	1.8	20
46	In Silico identification of pathogenic strains of Cronobacter from Biochemical data reveals association of inositol fermentation with pathogenicity. <i>BMC Microbiology</i> , 2011, 11, 204.	1.3	20
47	Promoter shuffling has occurred during the evolution of the vertebrate growth hormone gene. <i>Gene</i> , 2000, 254, 9-18.	1.0	19
48	A novel Alu-mediated 61-kb deletion of the von Willebrand factor (VWF) gene whose breakpoints co-locate with putative matrix attachment regions. <i>Blood Cells, Molecules, and Diseases</i> , 2006, 36, 385-391.	0.6	19
49	Changes in primary DNA sequence complexity influence the phenotypic consequences of mutations in human gene regulatory regions. <i>Human Genetics</i> , 2000, 107, 362-365.	1.8	16
50	Complexity and robustness in hypernetwork models of metabolism. <i>Journal of Theoretical Biology</i> , 2016, 406, 99-104.	0.8	16
51	A novel gross deletion caused by non-homologous recombination of the PDHX gene in a patient with pyruvate dehydrogenase deficiency†. <i>Molecular Genetics and Metabolism</i> , 2006, 89, 106-110.	0.5	13
52	A gene conversion hotspot in the human growth hormone (<i>GH1</i>) gene promoter. <i>Human Mutation</i> , 2009, 30, 239-247.	1.1	13
53	Comparative analysis of germline and somatic microlesion mutational spectra in 17 human tumor suppressor genes. <i>Human Mutation</i> , 2011, 32, 620-632.	1.1	13
54	An isolated case of lissencephaly caused by the insertion of a mitochondrial genome-derived DNA sequence into the 5' untranslated region of the PFAH1B1 (LIS1) gene. <i>Human Genomics</i> , 2010, 4, 384.	1.4	10

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55	Searching for potential microRNA-binding site mutations amongst known disease-associated 3' UTR variants. <i>Genomic Medicine</i> , 2007, 1, 29-33.	0.6	7
56	Network motif frequency vectors reveal evolving metabolic network organisation. <i>Molecular BioSystems</i> , 2015, 11, 77-85.	2.9	7
57	Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. <i>Clinical Genetics</i> , 2013, 84, 552-559.	1.0	6
58	Predicting novel genomic regions linked to genetic disorders using GWAS and chromosome conformation data – a case study of schizophrenia. <i>Scientific Reports</i> , 2019, 9, 17940.	1.6	6
59	Identification of novel genes associated with longevity in <i>Drosophila melanogaster</i> - a computational approach. <i>Aging</i> , 2019, 11, 11244-11267.	1.4	6
60	A rare complex DNA rearrangement in the murine Steel gene results in exon duplication and a lethal phenotype. <i>Blood</i> , 2003, 102, 3548-3555.	0.6	5
61	Two sisters with Rett syndrome and non-identical paternally-derived microdeletions in the MECP2 gene. <i>Genomic Medicine</i> , 2008, 2, 77-81.	0.6	4
62	Screening in silico predicted remotely acting NF1 gene regulatory elements for mutations in patients with neurofibromatosis type 1. <i>Human Genomics</i> , 2013, 7, 18.	1.4	4
63	The Somatic Mutational Spectrum of the NF1 Gene. , 2012, , 211-233.		3
64	Indel in the FIC1/ATP8B1 gene? a novel rare type of mutation associated with benign recurrent intrahepatic cholestasis. <i>Hepatology Research</i> , 2004, 30, 1-3.	1.8	1
65	Remotely acting SMCHD1 gene regulatory elements: in silico prediction and identification of potential regulatory variants in patients with FSHD. <i>Human Genomics</i> , 2015, 9, 25.	1.4	0