Tomoo Ogi

List of Publications by Year in descending order

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89 papers	5,959 citations	117625 34 h-index	76900 74 g-index
93 all docs	93 docs citations	93 times ranked	7098 citing authors

#	Article	IF	Citations
1	Clinical practice guidelines for pseudoxanthoma elasticum (2017). Journal of Dermatology, 2022, 49, .	1.2	8
2	Case of ichthyosis with confetti caused by <scp>KRT10</scp> mutation, complicated with multiple malignant melanomas. Journal of Dermatology, 2022, 49, .	1.2	1
3	Six yearsâ€™ accomplishment of the Initiative on Rare and Undiagnosed Diseases: nationwide project in Japan to discover causes, mechanisms, and cures. Journal of Human Genetics, 2022, 67, 505-513.	2.3	17
4	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	2.4	5
5	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
6	A novel ZC4H2 variant in a female with severe respiratory complications. Brain and Development, 2022, 44, 571-577.	1.1	0
7	Exome sequencing of Japanese schizophrenia multiplex families supports the involvement of calcium ion channels. PLoS ONE, 2022, 17, e0268321.	2.5	0
8	Exome sequencing analysis of Japanese autism spectrum disorder case-control sample supports an increased burden of synaptic function-related genes. Translational Psychiatry, 2022, 12, .	4.8	4
9	Expanding the phenotype of biallelic lossâ€ofâ€function variants in the <scp>NSUN2</scp> gene: Description of four individuals with juvenile cataract, chronic nephritis, or brain anomaly as novel complications. American Journal of Medical Genetics, Part A, 2021, 185, 282-285.	1.2	5
10	The wide-ranging clinical and genetic features in Japanese families with valosin-containing protein proteinopathy. Neurobiology of Aging, 2021, 100, 120.e1-120.e6.	3.1	8
11	Hereditary Mucoepithelial Dysplasia and Autosomal-Dominant IFAP Syndrome Is a Clinical Spectrum Due to SREBF1 Variants. Journal of Investigative Dermatology, 2021, 141, 1596-1598.	0.7	6
12	Microglial gene signature reveals loss of homeostatic microglia associated with neurodegeneration of Alzheimerâ€™s disease. Acta Neuropathologica Communications, 2021, 9, 1.	5.2	172
13	Identification of a novel causative mutation in KRT1 in diffuse palmoplantar keratoderma, facilitated by whole-exome sequencing. European Journal of Dermatology, 2021, 31, 264-265.	0.6	0
14	Temporal dynamics of the plasma microbiome in recipients at early post-liver transplantation: a retrospective study. BMC Microbiology, 2021, 21, 104.	3.3	5
15	Protein instability associated with AARS1 and MARS1 mutations causes trichothiodystrophy. Human Molecular Genetics, 2021, 30, 1711-1720.	2.9	20
16	Next-Generation Sequencing to Detect Pathogens in Pediatric Febrile Neutropenia: A Single-Center Retrospective Study of 112 Cases. Open Forum Infectious Diseases, 2021, 8, ofab223.	0.9	11
17	Transcription-Coupled DNA Repair: From Mechanism to Human Disorder. Trends in Cell Biology, 2021, 31, 359-371.	7.9	49
18	Successful dupilumab treatment for ichthyotic and atopic features of Netherton syndrome. Journal of Dermatological Science, 2021, 102, 126-129.	1.9	29

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19	ELOF1 is a transcription-coupled DNA repair factor that directs RNA polymerase II ubiquitylation. Nature Cell Biology, 2021, 23, 595-607.	10.3	38
20	Pediatric sepsis cases diagnosed with group B streptococcal meningitis using next-generation sequencing: a report of two cases. BMC Infectious Diseases, 2021, 21, 531.	2.9	5
21	Predominant cellular mitochondrial dysfunction in the TOP3A gene-caused Bloom syndrome-like disorder. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166106.	3.8	7
22	The sodium-glucose cotransporter-2 inhibitor Tofogliflozin prevents the progression of nonalcoholic steatohepatitisâ€“associated liver tumors in a novel murine model. Biomedicine and Pharmacotherapy, 2021, 140, 111738.	5.6	11
23	Updated allele frequencies of SERPINB7 founder mutations in Asian patients with Nagashima-type palmoplantar keratosis/keratoderma. Journal of Dermatological Science, 2021, 103, 116-119.	1.9	5
24	Extensive Multiple Organ Involvement in VEXAS Syndrome. Arthritis and Rheumatology, 2021, 73, 1896-1897.	5.6	25
25	Dealing with transcription-blocking DNA damage: Repair mechanisms, RNA polymerase II processing and human disorders. DNA Repair, 2021, 106, 103192.	2.8	25
26	Odontogenic keratocysts are an important clue for diagnosing basal cell nevus syndrome. Nagoya Journal of Medical Science, 2021, 83, 393-396.	0.3	1
27	Reduced stratum corneum acylceramides in autosomal recessive congenital ichthyosis with a NIPAL4 mutation. Journal of Dermatological Science, 2020, 97, 50-56.	1.9	16
28	InMeRF: prediction of pathogenicity of missense variants by individual modeling for each amino acid substitution. NAR Genomics and Bioinformatics, 2020, 2, lqaa038.	3.2	16
29	Gene Expression Profile at the Motor Endplate of the Neuromuscular Junction of Fast-Twitch Muscle. Frontiers in Molecular Neuroscience, 2020, 13, 154.	2.9	12
30	Astrocytic phagocytosis is a compensatory mechanism for microglial dysfunction. EMBO Journal, 2020, 39, e104464.	7.8	105
31	NUS1 mutation in a family with epilepsy, cerebellar ataxia, and tremor. Epilepsy Research, 2020, 164, 106371.	1.6	18
32	Topoisomerase I-driven repair of UV-induced damage in NER-deficient cells. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 14412-14420.	7.1	16
33	Ubiquitination of DNA Damage-Stalled RNAPII Promotes Transcription-Coupled Repair. Cell, 2020, 180, 1228-1244.e24.	28.9	132
34	Severe achondroplasia due to two de novo variants in the transmembrane domain of FGFR3 on the same allele: A case report. Molecular Genetics & Genomic Medicine, 2020, 8, e1148.	1.2	2
35	Digenic mutations in ALDH2 and ADH5 impair formaldehyde clearance and cause a multisystem disorder, AMeD syndrome. Science Advances, 2020, 6, .	10.3	39
36	A heterozygous SERPINB7 mutation is a possible modifying factor for epidermolytic palmoplantar keratoderma. Journal of Dermatological Science, 2020, 100, 148-151.	1.9	3

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37	1212. Temporal Dynamics of the Plasma Microbiome in Recipients at Early Post-liver Transplantation. Open Forum Infectious Diseases, 2020, 7, S627-S628.	0.9	0
38	Diagnostic Whole Exome Sequencing for 166 Patients with Inherited Bone Marrow Failure Syndrome. Blood, 2020, 136, 9-9.	1.4	1
39	Bi-allelic TARS Mutations Are Associated with Brittle Hair Phenotype. American Journal of Human Genetics, 2019, 105, 434-440.	6.2	42
40	Functional Comparison of XPF Missense Mutations Associated to Multiple DNA Repair Disorders. Genes, 2019, 10, 60.	2.4	8
41	JAK/STAT3 and NF-Î²B Signaling Pathways Regulate Cancer Stem-Cell Properties in Anaplastic Thyroid Cancer Cells. Thyroid, 2019, 29, 674-682.	4.5	39
42	242. Comprehensive Pathogen Detection for Pediatric Febrile Neutropenia by Metagenomic Next-Generation Sequencing. Open Forum Infectious Diseases, 2019, 6, S137-S138.	0.9	0
43	Disorders with Deficiency in TC-NER: Molecular Pathogenesis of Cockayne Syndrome and UV-Sensitive Syndrome. , 2019, , 25-40.		0
44	A Japanese Case of Galli-Galli Disease due to a Previously Unreported POGLUT1 Mutation. Acta Dermato-Venereologica, 2019, 99, 458-459.	1.3	7
45	Hailey-Hailey disease with oesophageal involvement due to a previously unreported ATP2C1 mutation. European Journal of Dermatology, 2019, , .	0.6	1
46	Cerebellar ataxia-dominant phenotype in patients with ERCC4 mutations. Journal of Human Genetics, 2018, 63, 417-423.	2.3	15
47	Functional and clinical relevance of novel mutations in a large cohort of patients with Cockayne syndrome. Journal of Medical Genetics, 2018, 55, 329-343.	3.2	55
48	An adolescent case of xeroderma pigmentosum variant confirmed by the onset of sun exposureâ€related skin cancer during Crohn's disease treatment. Journal of Cutaneous Immunology and Allergy, 2018, 1, 23-26.	0.3	1
49	Novel function of HATs and HDACs in homologous recombination through acetylation of human RAD52 at double-strand break sites. PLoS Genetics, 2018, 14, e1007277.	3.5	25
50	Analysis of clinical symptoms and <scp>ABCC</scp>6 mutations in 76 Japanese patients with pseudoxanthoma elasticum. Journal of Dermatology, 2017, 44, 644-650.	1.2	20
51	Common TFIIH recruitment mechanism in global genome and transcription-coupled repair subpathways. Nucleic Acids Research, 2017, 45, 13043-13055.	14.5	83
52	Transplantation of bioengineered rat lungs recellularized with endothelial and adipose-derived stromal cells. Scientific Reports, 2017, 7, 8447.	3.3	58
53	Phosphorylated HBO1 at UV irradiated sites is essential for nucleotide excision repair. Nature Communications, 2017, 8, 16102.	12.8	29
54	PCNA ubiquitylation ensures timely completion of unperturbed DNA replication in fission yeast. PLoS Genetics, 2017, 13, e1006789.	3.5	20

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55	Calcification in dermal fibroblasts from a patient with GGCX syndrome accompanied by upregulation of osteogenic molecules. PLoS ONE, 2017, 12, e0177375.	2.5	8
56	ALC1/CHD1L, a chromatin-remodeling enzyme, is required for efficient base excision repair. PLoS ONE, 2017, 12, e0188320.	2.5	34
57	A 10â€year followâ€up of a child with mild case of xeroderma pigmentosum complementation group D diagnosed by wholeâ€genome sequencing. Photodermatology Photoimmunology and Photomedicine, 2016, 32, 174-180.	1.5	9
58	SETDB1, HP1 and SUV39 promote repositioning of 53BP1 to extend resection during homologous recombination in G2 cells. Nucleic Acids Research, 2015, 43, 7931-7944.	14.5	69
59	XRCC4 deficiency in human subjects causes a marked neurological phenotype but no overt immunodeficiency. Journal of Allergy and Clinical Immunology, 2015, 136, 1007-1017.	2.9	44
60	Sensitivity and dose dependency of radiation-induced injury in hematopoietic stem/progenitor cells in mice. Scientific Reports, 2015, 5, 8055.	3.3	29
61	Novel compound heterozygous DNA ligase IV mutations in an adolescent with a slowly-progressing radiosensitive-severe combined immunodeficiency. Clinical Immunology, 2015, 160, 255-260.	3.2	29
62	A rapid, comprehensive system for assaying DNA repair activity and cytotoxic effects of DNA-damaging reagents. Nature Protocols, 2015, 10, 12-24.	12.0	39
63	Hypomorphic PCNA mutation underlies a human DNA repair disorder. Journal of Clinical Investigation, 2014, 124, 3137-3146.	8.2	77
64	Functional characterization of the novel BRAF complex mutation, BRAF^{V600delinsYM}, identified in papillary thyroid carcinoma. International Journal of Cancer, 2013, 132, 738-743.	5.1	16
65	Malfunction of Nuclease ERCC1-XPF Results in Diverse Clinical Manifestations and Causes Cockayne Syndrome, Xeroderma Pigmentosum, and Fanconi Anemia. American Journal of Human Genetics, 2013, 92, 807-819.	6.2	178
66	PRKDC mutations in a SCID patient with profound neurological abnormalities. Journal of Clinical Investigation, 2013, 123, 2969-2980.	8.2	121
67	Identification of the First ATRIPâ€“Deficient Patient and Novel Mutations in ATR Define a Clinical Spectrum for ATRâ€“ATRIP Seckel Syndrome. PLoS Genetics, 2012, 8, e1002945.	3.5	104
68	miR-196a Downregulation Increases the Expression of Type I and III Collagens in Keloid Fibroblasts. Journal of Investigative Dermatology, 2012, 132, 1597-1604.	0.7	123
69	Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase IIo processing in transcription-coupled nucleotide-excision repair. Nature Genetics, 2012, 44, 586-592.	21.4	162
70	Two unrelated patients with MRE11A mutations and Nijmegen breakage syndrome-like severe microcephaly. DNA Repair, 2011, 10, 314-321.	2.8	49
71	A semi-automated non-radioactive system for measuring recovery of RNA synthesis and unscheduled DNA synthesis using ethynyluracil derivatives. DNA Repair, 2010, 9, 506-516.	2.8	69
72	Collaborative Action of Brca1 and CtIP in Elimination of Covalent Modifications from Double-Strand Breaks to Facilitate Subsequent Break Repair. PLoS Genetics, 2010, 6, e1000828.	3.5	133

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73	Three DNA Polymerases, Recruited by Different Mechanisms, Carry Out NER Repair Synthesis in Human Cells. Molecular Cell, 2010, 37, 714-727.	9.7	313
74	A rapid non-radioactive technique for measurement of repair synthesis in primary human fibroblasts by incorporation of ethynyl deoxyuridine (EdU). Nucleic Acids Research, 2008, 37, e31-e31.	14.5	102
75	Differential Bvg Phase-Dependent Regulation and Combinatorial Role in Pathogenesis of Two Bordetella Paralogs, BipA and BcfA. Journal of Bacteriology, 2007, 189, 3695-3704.	2.2	30
76	Translesion synthesis: Y-family polymerases and the polymerase switch. DNA Repair, 2007, 6, 891-899.	2.8	335
77	The Y-family DNA polymerase Î¹ (pol Î¹) functions in mammalian nucleotide-excision repair. Nature Cell Biology, 2006, 8, 640-642.	10.3	193
78	Involvement of Vertebrate PolÎ¹ in Translesion DNA Synthesis across DNA Monoalkylation Damage. Journal of Biological Chemistry, 2006, 281, 2000-2004.	3.4	33
79	Localisation of human Y-family DNA polymerase Î¹: relationship to PCNA foci. Journal of Cell Science, 2005, 118, 129-136.	2.0	79
80	Binding and transcriptional activation of non-flagellar genes by the Escherichia coli flagellar master regulator FlhD2C2. Microbiology (United Kingdom), 2005, 151, 1779-1788.	1.8	60
81	Mammalian Pol Î¹: Regulation of its Expression and Lesion Substrates. Advances in Protein Chemistry, 2004, 69, 265-278.	4.4	15
82	Elevated expression of DNA polymerase Î¹ in human lung cancer is associated with p53 inactivation: Negative regulation of POLK promoter activity by p53. International Journal of Oncology, 2004, 25, 161.	3.3	10
83	The absence of DNA polymerase Î¹ does not affect somatic hypermutation of the mouse immunoglobulin heavy chain gene. Immunology Letters, 2003, 86, 265-270.	2.5	41
84	Identification, Timing, and Signal Specificity ofPseudomonas aeruginosaQuorum-Controlled Genes: a Transcriptome Analysis. Journal of Bacteriology, 2003, 185, 2066-2079.	2.2	1,037
85	Nonlinear partial differential equations and applications: PolÂ protects mammalian cells against the lethal and mutagenic effects of benzo[a]pyrene. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 15548-15553.	7.1	222
86	Expression of human and mouse genes encoding polÎ¹: testis-specific developmental regulation and AhR-dependent inducible transcription. Genes To Cells, 2001, 6, 943-953.	1.2	79
87	Identification of additional genes belonging to the LexA regulon in Escherichia coli. Molecular Microbiology, 2000, 35, 1560-1572.	2.5	492
88	Error-prone bypass of certain DNA lesions by the human DNA polymerase Î¹. Genes and Development, 2000, 14, 1589-1594.	5.9	250
89	Mutation enhancement by DINB1, a mammalian homologue of the Escherichia coli mutagenesis protein DinB. Genes To Cells, 1999, 4, 607-618.	1.2	135