Robert Huether

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/994395/publications.pdf

Version: 2024-02-01

53	9,794	236925	254184
papers	citations	25 h-index	43 g-index

54	54	54	16691
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Specifications of the ACMG/AMP variant interpretation guidelines for germline TP53 variants. Human Mutation, 2021, 42, 223-236.	2.5	81
2	Real-world Evidence of Diagnostic Testing and Treatment Patterns in US Patients With Breast Cancer With Implications for Treatment Biomarkers From RNA Sequencing Data. Clinical Breast Cancer, 2021, 21, e340-e361.	2.4	10
3	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
4	Tumor Mutational Burden From Tumor-Only Sequencing Compared With Germline Subtraction From Paired Tumor and Normal Specimens. JAMA Network Open, 2020, 3, e200202.	5.9	40
5	Association of Breast and Ovarian Cancers With Predisposition Genes Identified by Large-Scale Sequencing. JAMA Oncology, 2019, 5, 51.	7.1	145
6	Integrated genomic profiling expands clinical options for patients with cancer. Nature Biotechnology, 2019, 37, 1351-1360.	17.5	103
7	Mutations in MAP3K1 that cause 46,XY disorders of sex development disrupt distinct structural domains in the protein. Human Molecular Genetics, 2019, 28, 1620-1628.	2.9	21
8	Clinical validation of the tempus xT next-generation targeted oncology sequencing assay. Oncotarget, 2019, 10, 2384-2396.	1.8	119
9	Pan-cancer genome and transcriptome analyses of 1,699 paediatric leukaemias and solid tumours. Nature, 2018, 555, 371-376.	27.8	649
10	De novo variants in KLF7 are a potential novel cause of developmental delay/intellectual disability, neuromuscular and psychiatric symptoms. Clinical Genetics, 2018, 93, 1030-1038.	2.0	9
11	Expansion and further delineation of the SETD5 phenotype leading to global developmental delay, variable dysmorphic features, and reduced penetrance. Clinical Genetics, 2018, 93, 752-761.	2.0	23
12	Clinical validation of the Tempus xO assay. Oncotarget, 2018, 9, 25826-25832.	1.8	43
13	Geneâ€specific criteria for PTEN variant curation: Recommendations from the ClinGen PTEN Expert Panel. Human Mutation, 2018, 39, 1581-1592.	2.5	123
14	Associations Between Cancer Predisposition Testing Panel Genes and Breast Cancer. JAMA Oncology, 2017, 3, 1190.	7.1	472
15	Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases. Genetics in Medicine, 2017, 19, 224-235.	2.4	47
16	Further evidence that de novo missense and truncating variants in <scp>ZBTB18</scp> cause intellectual disability with variable features. Clinical Genetics, 2017, 91, 697-707.	2.0	29
17	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. Annals of Neurology, 2016, 80, .	5.3	49
18	The genomic landscape of core-binding factor acute myeloid leukemias. Nature Genetics, 2016, 48, 1551-1556.	21.4	215

#	Article	IF	Citations
19	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. European Journal of Human Genetics, 2016, 24, 1761-1770.	2.8	36
20	Abstract 2597: Breast and ovarian cancer risks associated with cancer predisposition gene mutations identified by multigene panel testing. , 2016, , .		1
21	Mutations in RASA1 and GDF2 identified in patients with clinical features of hereditary hemorrhagic telangiectasia. Human Genome Variation, 2015, 2, 15040.	0.7	53
22	Germline activating MTOR mutation arising through gonadal mosaicism in two brothers with megalencephaly and neurodevelopmental abnormalities. BMC Medical Genetics, 2015, 16, 102.	2.1	23
23	The landscape of somatic mutations in infant MLL-rearranged acute lymphoblastic leukemias. Nature Genetics, 2015, 47, 330-337.	21.4	405
24	C11ORF95-RELA FUSIONS DRIVE ONCOGENIC NF-KB SIGNALING IN EPENDYMOMA. Neuro-Oncology, 2014, 16, iii16-iii16.	1.2	1
25	The landscape of somatic mutations in epigenetic regulators across 1,000 paediatric cancer genomes. Nature Communications, 2014, 5, 3630.	12.8	342
26	C11orf95â€“RELA fusions drive oncogenic NF-Î²B signalling in ependymoma. Nature, 2014, 506, 451-455.	27.8	559
27	The genomic landscape of diffuse intrinsic pontine glioma and pediatric non-brainstem high-grade glioma. Nature Genetics, 2014, 46, 444-450.	21.4	871
28	Survival analysis of infected mice reveals pathogenic variations in the genome of avian H1N1 viruses. Scientific Reports, 2014, 4, 7455.	3.3	13
29	Incidence of Germline Mutations in Cancer-Predisposition Genes in Children with Hematologic Malignancies: a Report from the Pediatric Cancer Genome Project. Blood, 2014, 124, 127-127.	1.4	9
30	RB1 gene inactivation by chromothripsis in human retinoblastoma. Oncotarget, 2014, 5, 438-450.	1.8	104
31	Abstract 2930: Global chromatin profiling reveals NSD2 mutation in pediatric ALL. , 2014, , .		0
32	Abstract PR03: The genomic landscape of diffuse intrinsic pontine glioma and pediatric non-brainstem high-grade glioma. , 2014, , .		2
33	Global chromatin profiling reveals NSD2 mutations in pediatric acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 1386-1391.	21.4	238
34	Whole-genome sequencing identifies genetic alterations in pediatric low-grade gliomas. Nature Genetics, 2013, 45, 602-612.	21.4	704
35	The genomic landscape of hypodiploid acute lymphoblastic leukemia. Nature Genetics, 2013, 45, 242-252.	21.4	588
36	Evolution of bacterial ribosomal protein L1. International Journal of Bioinformatics Research and Applications, 2012, 8, 99.	0.2	0

#	Article	IF	Citations
37	Somatic histone H3 alterations in pediatric diffuse intrinsic pontine gliomas and non-brainstem glioblastomas. Nature Genetics, 2012, 44, 251-253.	21.4	1,402
38	The genetic basis of early T-cell precursor acute lymphoblastic leukaemia. Nature, 2012, 481, 157-163.	27.8	1,430
39	Novel mutations target distinct subgroups of medulloblastoma. Nature, 2012, 488, 43-48.	27.8	742
40	Use of whole genome sequencing to identify novel mutations in distinct subgroups of medulloblastoma.. Journal of Clinical Oncology, 2012, 30, 9518-9518.	1.6	0
41	Discovery of Novel Recurrent Mutations in Childhood Early T-Cell Precursor Acute Lymphoblastic Leukemia by Whole Genome Sequencing - a Report From the St Jude Children's Research Hospital - Washington University Pediatric Cancer Genome Project. Blood, 2011, 118, 68-68.	1.4	0
42	The shortâ€chain oxidoreductase Q9HYA2 from Pseudomonas aeruginosa PAO1 contains an atypical catalytic center. Protein Science, 2010, 19, 1097-1103.	7.6	1
43	Sequence fingerprint and structural analysis of the SCOR enzyme A3DFK9 from Clostridium thermocellum. Proteins: Structure, Function and Bioinformatics, 2010, 78, 603-613.	2.6	1
44	Correlation in species, cofactor, oligomerization and substrate specificity in shortâ€chain oxidoreductase enzymes. FASEB Journal, 2010, 24, lb163.	0.5	0
45	Ribosomal protein structures and sequences define the prokaryotic tree of life. Acta Crystallographica Section A: Foundations and Advances, 2010, 66, s21-s21.	0.3	0
46	Divergent evolution of a Rossmann fold and identification of its oldest surviving ancestor. International Journal of Bioinformatics Research and Applications, 2009, 5, 280.	0.2	9
47	Multiple open reading frames GARP content and a 32-letter genetic code. Acta Crystallographica Section A: Foundations and Advances, 2009, 65, s163-s163.	0.3	0
48	Divergent evolution of a Rossmann fold and identification of its oldest surviving ancestor. International Journal of Bioinformatics Research and Applications, 2009, 5, 280-94.	0.2	5
49	Structure/function of human type 1 3Î²-hydroxysteroid dehydrogenase: An intrasubunit disulfide bond in the Rossmann-fold domain and a Cys residue in the active site are critical for substrate and coenzyme utilization. Journal of Steroid Biochemistry and Molecular Biology, 2007, 107, 80-87.	2.5	14
50	Rational proteomics of PKD1. I. Modeling the three dimensional structure and ligand specificity of the C_lectin binding domain of Polycystin-1.. Journal of Molecular Modeling, 2007, 13, 891-896.	1.8	7
51	Multiple open reading frames, codon bias and the evolution of the genetic code. FASEB Journal, 2006, 20, A927.	0.5	0
52	Determining Structure and Function of Steroid Dehydrogenase Enzymes by Sequence Analysis, Homology Modeling, and Rational Mutational Analysis. Annals of the New York Academy of Sciences, 2005, 1061, 135-148.	3.8	19
53	Rational genomics I: Antisense open reading frames and codon bias in short-chain oxido reductase enzymes and the evolution of the genetic code. Proteins: Structure, Function and Bioinformatics, 2005, 61, 900-906.	2.6	13