

Pierre Lindenbaum

List of Publications by Year in descending order

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Version: 2024-02-01

31
papers

1,816
citations

361413

20
h-index

434195

31
g-index

33
all docs

33
docs citations

33
times ranked

4410
citing authors

#	ARTICLE	IF	CITATIONS
1	Common variants at SCN5A-SCN10A and HEY2 are associated with Brugada syndrome, a rare disease with high risk of sudden cardiac death. <i>Nature Genetics</i> , 2013, 45, 1044-1049.	21.4	467
2	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. <i>Nature Genetics</i> , 2011, 43, 306-308.	21.4	181
3	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	6.2	136
4	Testing the burden of rare variation in arrhythmia-susceptibility genes provides new insights into molecular diagnosis for Brugada syndrome. <i>Human Molecular Genetics</i> , 2015, 24, 2757-2763.	2.9	130
5	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 99, 695-703.	6.2	87
6	BioStar: An Online Question & Answer Resource for the Bioinformatics Community. <i>PLoS Computational Biology</i> , 2011, 7, e1002216.	3.2	82
7	Mutations in FAM111B Cause Hereditary Fibrosing Poikiloderma with Tendon Contracture, Myopathy, and Pulmonary Fibrosis. <i>American Journal of Human Genetics</i> , 2013, 93, 1100-1107.	6.2	76
8	The Gene Wiki: community intelligence applied to human gene annotation. <i>Nucleic Acids Research</i> , 2010, 38, D633-D639.	14.5	67
9	Targeted resequencing identifies TRPM4 as a major gene predisposing to progressive familial heart block type I. <i>International Journal of Cardiology</i> , 2016, 207, 349-358.	1.7	62
10	Advanced Characterization of DNA Molecules in rAAV Vector Preparations by Single-stranded Virus Next-generation Sequencing. <i>Molecular Therapy - Nucleic Acids</i> , 2015, 4, e260.	5.1	57
11	Mass Spectrometry-Based Identification of Native Cardiac Nav1.5 Channel $\hat{\pm}$ Subunit Phosphorylation Sites. <i>Journal of Proteome Research</i> , 2012, 11, 5994-6007.	3.7	47
12	De Novo Truncating Mutations in the Kinetochores-Microtubules Attachment Gene <i>CHAMP1</i> Cause Syndromic Intellectual Disability. <i>Human Mutation</i> , 2016, 37, 354-358.	2.5	40
13	Search for Rare Copy-Number Variants in Congenital Heart Defects Identifies Novel Candidate Genes and a Potential Role for FOXC1 in Patients With Coarctation of the Aorta. <i>Circulation: Cardiovascular Genetics</i> , 2016, 9, 86-94.	5.1	38
14	Rare Coding Variants in ANGPTL6 Are Associated with Familial Forms of Intracranial Aneurysm. <i>American Journal of Human Genetics</i> , 2018, 102, 133-141.	6.2	37
15	RoXaN, a Novel Cellular Protein Containing TPR, LD, and Zinc Finger Motifs, Forms a Ternary Complex with Eukaryotic Initiation Factor 4G and Rotavirus NSP3. <i>Journal of Virology</i> , 2004, 78, 3851-3862.	3.4	36
16	Association of autism with polymorphisms in the paired-like homeodomain transcription factor 1 (PITX1) on chromosome 5q31: a candidate gene analysis. <i>BMC Medical Genetics</i> , 2007, 8, 74.	2.1	35
17	Accurate Identification and Quantification of DNA Species by Next-Generation Sequencing in Adeno-Associated Viral Vectors Produced in Insect Cells. <i>Human Gene Therapy Methods</i> , 2017, 28, 148-162.	2.1	31
18	bioalcidae, samjs and vcfilterjs: object-oriented formatters and filters for bioinformatics files. <i>Bioinformatics</i> , 2018, 34, 1224-1225.	4.1	27

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19	Knime4Bio: a set of custom nodes for the interpretation of next-generation sequencing data with KNIME. <i>Bioinformatics</i> , 2011, 27, 3200-3201.	4.1	26
20	The 3rd DBCLS BioHackathon: improving life science data integration with Semantic Web technologies. <i>Journal of Biomedical Semantics</i> , 2013, 4, 6.	1.6	26
21	Dysfunction of the Voltage-Gated K ⁺ Channel β 2 Subunit in a Familial Case of Brugada Syndrome. <i>Journal of the American Heart Association</i> , 2016, 5, .	3.7	20
22	Short-lived recombinant adeno-associated virus transgene expression in dystrophic muscle is associated with oxidative damage to transgene mRNA. <i>Molecular Therapy - Methods and Clinical Development</i> , 2015, 2, 15010.	4.1	18
23	RPL13 Variants Cause Spondyloepimetaphyseal Dysplasia with Severe Short Stature. <i>American Journal of Human Genetics</i> , 2019, 105, 1040-1047.	6.2	17
24	NGS library preparation may generate artifactual integration sites of AAV vectors. <i>Nature Medicine</i> , 2014, 20, 577-578.	30.7	13
25	Fryns type mesomelic dysplasia of the upper limbs caused by inverted duplications of the HOXD gene cluster. <i>European Journal of Human Genetics</i> , 2020, 28, 324-332.	2.8	12
26	Phenotypic Differences Between Polygenic and Monogenic Hypobetalipoproteinemia. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2021, 41, e63-e71.	2.4	12
27	Robust physical methods that enrich genomic regions identical by descent for linkage studies: confirmation of a locus for osteogenesis imperfecta. <i>BMC Genetics</i> , 2009, 10, 16.	2.7	7
28	APOB CRISPR-Cas9 Engineering in Hypobetalipoproteinemia: A Promising Tool for Functional Studies of Novel Variants. <i>International Journal of Molecular Sciences</i> , 2022, 23, 4281.	4.1	6
29	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. <i>Human Mutation</i> , 2019, 40, 1993-2000.	2.5	4
30	mod_bio: Apache modules for Next-Generation sequencing data. <i>Bioinformatics</i> , 2015, 31, 112-113.	4.1	1
31	Acknowledging contributions to online expert assistance. <i>Nature Precedings</i> , 2011, , .	0.1	0