Pierre Lindenbaum

List of Publications by Year
in descending order

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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	APOB CRISPR-Cas9 Engineering in Hypobetalipoproteinemia: A Promising Tool for Functional Studies of Novel Variants. International Journal of Molecular Sciences, 2022, 23, 4281.	4.1	6
2	Phenotypic Differences Between Polygenic and Monogenic Hypobetalipoproteinemia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2021, 41, e63-e71.	2.4	12
3	Fryns type mesomelic dysplasia of the upper limbs caused by inverted duplications of the HOXD gene cluster. European Journal of Human Genetics, 2020, 28, 324-332.	2.8	12
4	RPL13 Variants Cause Spondyloepimetaphyseal Dysplasia with Severe Short Stature. American Journal of Human Genetics, 2019, 105, 1040-1047.	6.2	17
5	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. Human Mutation, 2019, 40, 1993-2000.	2.5	4
6	bioalcidae, samjs and vcffilterjs: object-oriented formatters and filters for bioinformatics files. Bioinformatics, 2018, 34, 1224-1225.	4.1	27
7	Rare Coding Variants in ANGPTL6 Are Associated with Familial Forms of Intracranial Aneurysm. American Journal of Human Genetics, 2018, 102, 133-141.	6.2	37
8	Accurate Identification and Quantification of DNA Species by Next-Generation Sequencing in Adeno-Associated Viral Vectors Produced in Insect Cells. Human Gene Therapy Methods, 2017, 28, 148-162.	2.1	31
9	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
10	Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 695-703.	6.2	87
11	Dysfunction of the Voltageâ€Gated K ⁺ Channel Î²2 Subunit in a Familial Case of Brugada Syndrome. Journal of the American Heart Association, 2016, 5, .	3.7	20
12	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 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. Circulation: Cardiovascular Genetics, 2016, 9, 86-94.	5.1	38
14	Targeted resequencing identifies TRPM4 as a major gene predisposing to progressive familial heart block type I. International Journal of Cardiology, 2016, 207, 349-358.	1.7	62
15	Short-lived recombinant adeno-associated virus transgene expression in dystrophic muscle is associated with oxidative damage to transgene mRNA. Molecular Therapy - Methods and Clinical Development, 2015, 2, 15010.	4.1	18
16	Testing the burden of rare variation in arrhythmia-susceptibility genes provides new insights into molecular diagnosis for Brugada syndrome. Human Molecular Genetics, 2015, 24, 2757-2763.	2.9	130
17	Advanced Characterization of DNA Molecules in rAAV Vector Preparations by Single-stranded Virus Next-generation Sequencing. Molecular Therapy - Nucleic Acids, 2015, 4, e260.	5.1	57
18	mod_bio: Apache modules for Next-Generation sequencing data. Bioinformatics, 2015, 31, 112-113.	4.1	1

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19	NGS library preparation may generate artifactual integration sites of AAV vectors. Nature Medicine, 2014, 20, 577-578.	30.7	13
20	The 3rd DBCLS BioHackathon: improving life science data integration with Semantic Web technologies. Journal of Biomedical Semantics, 2013, 4, 6.	1.6	26
21	Common variants at SCN5A-SCN10A and HEY2 are associated with Brugada syndrome, a rare disease with high risk of sudden cardiac death. Nature Genetics, 2013, 45, 1044-1049.	21.4	467
22	Mutations in FAM111B Cause Hereditary Fibrosing Poikiloderma with Tendon Contracture, Myopathy, and Pulmonary Fibrosis. American Journal of Human Genetics, 2013, 93, 1100-1107.	6.2	76
23	Mass Spectrometry-Based Identification of Native Cardiac Nav1.5 Channel Î± Subunit Phosphorylation Sites. Journal of Proteome Research, 2012, 11, 5994-6007.	3.7	47
24	Acknowledging contributions to online expert assistance. Nature Precedings, 2011, , .	0.1	0
25	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. Nature Genetics, 2011, 43, 306-308.	21.4	181
26	Knime4Bio: a set of custom nodes for the interpretation of next-generation sequencing data with KNIME. Bioinformatics, 2011, 27, 3200-3201.	4.1	26
27	BioStar: An Online Question & Answer Resource for the Bioinformatics Community. PLoS Computational Biology, 2011, 7, e1002216.	3.2	82
28	The Gene Wiki: community intelligence applied to human gene annotation. Nucleic Acids Research, 2010, 38, D633-D639.	14.5	67
29	Robust physical methods that enrich genomic regions identical by descent for linkage studies: confirmation of a locus for osteogenesis imperfecta. BMC Genetics, 2009, 10, 16.	2.7	7
30	Association of autism with polymorphisms in the paired-like homeodomain transcription factor 1 (PITX1) on chromosome 5q31: a candidate gene analysis. BMC Medical Genetics, 2007, 8, 74.	2.1	35
31	RoXaN, a Novel Cellular Protein Containing TPR, LD, and Zinc Finger Motifs, Forms a Ternary Complex with Eukaryotic Initiation Factor 4G and Rotavirus NSP3. Journal of Virology, 2004, 78, 3851-3862.	3.4	36