

Alessia David

List of Publications by Year in descending order

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Version: 2024-02-01

30
papers

1,461
citations

471509

17
h-index

501196

28
g-index

34
all docs

34
docs citations

34
times ranked

2327
citing authors

#	ARTICLE	IF	CITATIONS
1	PDBe-KB: collaboratively defining the biological context of structural data. <i>Nucleic Acids Research</i> , 2022, 50, D534-D542.	14.5	46
2	The AlphaFold Database of Protein Structures: A Biologist's Guide. <i>Journal of Molecular Biology</i> , 2022, 434, 167336.	4.2	126
3	A common Tmprss2 variant has a protective effect against severe COVID-19. <i>Current Research in Translational Medicine</i> , 2022, 70, 103333.	1.8	30
4	GWYRE: A Resource for Mapping Variants onto Experimental and Modeled Structures of Human Protein Complexes. <i>Journal of Molecular Biology</i> , 2022, 434, 167608.	4.2	4
5	Missense3D-DB web catalogue: an atom-based analysis and repository of 4M human protein-coding genetic variants. <i>Human Genetics</i> , 2021, 140, 805-812.	3.8	39
6	Whole exome sequencing identifies deleterious rare variants in CCDC141 in familial self-limited delayed puberty. <i>Npj Genomic Medicine</i> , 2021, 6, 107.	3.8	4
7	Ligand-Specific Factors Influencing GLP-1 Receptor Post-Endocytic Trafficking and Degradation in Pancreatic Beta Cells. <i>International Journal of Molecular Sciences</i> , 2020, 21, 8404.	4.1	28
8	A polygenic biomarker to identify patients with severe hypercholesterolemia of polygenic origin. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1248.	1.2	5
9	LGR4 deficiency results in delayed puberty through impaired Wnt/ β -catenin signaling. <i>JCI Insight</i> , 2020, 5, .	5.0	25
10	PhyreRisk: A Dynamic Web Application to Bridge Genomics, Proteomics and 3D Structural Data to Guide Interpretation of Human Genetic Variants. <i>Journal of Molecular Biology</i> , 2019, 431, 2460-2466.	4.2	21
11	EAP1 regulation of GnRH promoter activity is important for human pubertal timing. <i>Human Molecular Genetics</i> , 2019, 28, 1357-1368.	2.9	29
12	Identification of disease-associated loci using machine learning for genotype and network data integration. <i>Bioinformatics</i> , 2019, 35, 5182-5190.	4.1	7
13	Can Predicted Protein 3D Structures Provide Reliable Insights into whether Missense Variants Are Disease Associated?. <i>Journal of Molecular Biology</i> , 2019, 431, 2197-2212.	4.2	344
14	PhenoRank: reducing study bias in gene prioritization through simulation. <i>Bioinformatics</i> , 2018, 34, 2087-2095.	4.1	30
15	Contributions of Function-Altering Variants in Genes Implicated in Pubertal Timing and Body Mass for Self-Limited Delayed Puberty. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 649-659.	3.6	31
16	Properties of human genes guided by their enrichment in rare and common variants. <i>Human Mutation</i> , 2018, 39, 365-370.	2.5	13
17	Structural Biology Helps Interpret Variants of Uncertain Significance in Genes Causing Endocrine and Metabolic Disorders. <i>Journal of the Endocrine Society</i> , 2018, 2, 842-854.	0.2	7
18	Landscape of Pleiotropic Proteins Causing Human Disease: Structural and System Biology Insights. <i>Human Mutation</i> , 2017, 38, 289-296.	2.5	26

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19	In-frame seven amino-acid duplication in AIP arose over the last 3000 years, disrupts protein interaction and stability and is associated with gigantism. <i>European Journal of Endocrinology</i> , 2017, 177, 257-266.	3.7	12
20	Novel Dominant-Negative GH Receptor Mutations Expands the Spectrum of GHI and IGF-I Deficiency. <i>Journal of the Endocrine Society</i> , 2017, 1, 345-358.	0.2	26
21	<i><sc>IGSF</sc> 10</i> mutations dysregulate gonadotropinâ€releasing hormone neuronal migration resulting in delayed puberty. <i>EMBO Molecular Medicine</i> , 2016, 8, 626-642.	6.9	109
22	Threeâ€Dimensional Model of Human Nicotinamide Nucleotide Transhydrogenase (NNT) and Sequenceâ€Structure Analysis of its Diseaseâ€Causing Variations. <i>Human Mutation</i> , 2016, 37, 1074-1084.	2.5	16
23	Exploring the cellular basis of human disease through a large-scale mapping of deleterious genes to cell types. <i>Genome Medicine</i> , 2015, 7, 95.	8.2	13
24	The Contribution of Missense Mutations in Core and Rim Residues of Proteinâ€Protein Interfaces to Human Disease. <i>Journal of Molecular Biology</i> , 2015, 427, 2886-2898.	4.2	105
25	A new structural model of the acid-labile subunit: pathogenetic mechanisms of short stature-causing mutations. <i>Journal of Molecular Endocrinology</i> , 2012, 49, 213-220.	2.5	16
26	Protein-protein interaction sites are hot spots for disease-associated nonsynonymous SNPs. <i>Human Mutation</i> , 2012, 33, 359-363.	2.5	149
27	Evidence for a Continuum of Genetic, Phenotypic, and Biochemical Abnormalities in Children with Growth Hormone Insensitivity. <i>Endocrine Reviews</i> , 2011, 32, 472-497.	20.1	171
28	Repair of Aberrant Splicing in Growth Hormone Receptor by Antisense Oligonucleotides Targeting the Splice Sites of a Pseudoexon. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, 3542-3546.	3.6	10
29	A Common <i>TMPRSS2</i> Variant Protects Against Severe COVID-19. <i>SSRN Electronic Journal</i> , 0, , .	0.4	2
30	Annotation and curation of human genomic variations: an ELIXIR Implementation Study. <i>F1000Research</i> , 0, 9, 1207.	1.6	0