Alessia David

List of Publications by Year
in descending order

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	Can Predicted Protein 3D Structures Provide Reliable Insights into whether Missense Variants Are Disease Associated?. Journal of Molecular Biology, 2019, 431, 2197-2212.	4.2	344
2	Evidence for a Continuum of Genetic, Phenotypic, and Biochemical Abnormalities in Children with Growth Hormone Insensitivity. Endocrine Reviews, 2011, 32, 472-497.	20.1	171
3	Protein-protein interaction sites are hot spots for disease-associated nonsynonymous SNPs. Human Mutation, 2012, 33, 359-363.	2.5	149
4	The AlphaFold Database of Protein Structures: A Biologistâ€™s Guide. Journal of Molecular Biology, 2022, 434, 167336.	4.2	126
5	<scp>IGSF</scp> 10 mutations dysregulate gonadotropinâ€releasing hormone neuronal migration resulting in delayed puberty. EMBO Molecular Medicine, 2016, 8, 626-642.	6.9	109
6	The Contribution of Missense Mutations in Core and Rim Residues of Proteinâ€“Protein Interfaces to Human Disease. Journal of Molecular Biology, 2015, 427, 2886-2898.	4.2	105
7	PDBe-KB: collaboratively defining the biological context of structural data. Nucleic Acids Research, 2022, 50, D534-D542.	14.5	46
8	Missense3D-DB web catalogue: an atom-based analysis and repository of 4M human protein-coding genetic variants. Human Genetics, 2021, 140, 805-812.	3.8	39
9	Contributions of Function-Altering Variants in Genes Implicated in Pubertal Timing and Body Mass for Self-Limited Delayed Puberty. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 649-659.	3.6	31
10	PhenoRank: reducing study bias in gene prioritization through simulation. Bioinformatics, 2018, 34, 2087-2095.	4.1	30
11	A common TMPRSS2 variant has a protective effect against severe COVID-19. Current Research in Translational Medicine, 2022, 70, 103333.	1.8	30
12	EAP1 regulation of GnRH promoter activity is important for human pubertal timing. Human Molecular Genetics, 2019, 28, 1357-1368.	2.9	29
13	Ligand-Specific Factors Influencing GLP-1 Receptor Post-Endocytic Trafficking and Degradation in Pancreatic Beta Cells. International Journal of Molecular Sciences, 2020, 21, 8404.	4.1	28
14	Landscape of Pleiotropic Proteins Causing Human Disease: Structural and System Biology Insights. Human Mutation, 2017, 38, 289-296.	2.5	26
15	Novel Dominant-Negative GH Receptor Mutations Expands the Spectrum of GHI and IGF-I Deficiency. Journal of the Endocrine Society, 2017, 1, 345-358.	0.2	26
16	LGR4 deficiency results in delayed puberty through impaired Wnt/Î²-catenin signaling. JCI Insight, 2020, 5, .	5.0	25
17	PhyreRisk: A Dynamic Web Application to Bridge Genomics, Proteomics and 3D Structural Data to Guide Interpretation of Human Genetic Variants. Journal of Molecular Biology, 2019, 431, 2460-2466.	4.2	21
18	A new structural model of the acid-labile subunit: pathogenetic mechanisms of short stature-causing mutations. Journal of Molecular Endocrinology, 2012, 49, 213-220.	2.5	16

#	Article	IF	Citations
19	ThreeâEDimensional Model of Human Nicotinamide Nucleotide Transhydrogenase (NNT) and SequenceâEStructure Analysis of its DiseaseâECausing Variations. Human Mutation, 2016, 37, 1074-1084.	2.5	16
20	Exploring the cellular basis of human disease through a large-scale mapping of deleterious genes to cell types. Genome Medicine, 2015, 7, 95.	8.2	13
21	Properties of human genes guided by their enrichment in rare and common variants. Human Mutation, 2018, 39, 365-370.	2.5	13
22	In-frame seven amino-acid duplication in AIP arose over the last 3000 years, disrupts protein interaction and stability and is associated with gigantism. European Journal of Endocrinology, 2017, 177, 257-266.	3.7	12
23	Repair of Aberrant Splicing in Growth Hormone Receptor by Antisense Oligonucleotides Targeting the Splice Sites of a Pseudoexon. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 3542-3546.	3.6	10
24	Structural Biology Helps Interpret Variants of Uncertain Significance in Genes Causing Endocrine and Metabolic Disorders. Journal of the Endocrine Society, 2018, 2, 842-854.	0.2	7
25	Identification of disease-associated loci using machine learning for genotype and network data integration. Bioinformatics, 2019, 35, 5182-5190.	4.1	7
26	A polygenic biomarker to identify patients with severe hypercholesterolemia of polygenic origin. Molecular Genetics & Genomic Medicine, 2020, 8, e1248.	1.2	5
27	Whole exome sequencing identifies deleterious rare variants in CCDC141 in familial self-limited delayed puberty. Npj Genomic Medicine, 2021, 6, 107.	3.8	4
28	GWYRE: A Resource for Mapping Variants onto Experimental and Modeled Structures of Human Protein Complexes. Journal of Molecular Biology, 2022, 434, 167608.	4.2	4
29	A Common TMPRSS2 Variant Protects Against Severe COVID-19. SSRN Electronic Journal, 0, , .	0.4	2
30	Annotation and curation of human genomic variations: an ELIXIR Implementation Study. F1000Research, 0, 9, 1207.	1.6	0