Xavier De la Cruz

List of Publications by Year in descending order

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XAVIED DE LA COUZ

#	Article	IF	CITATIONS
1	PirePred. Journal of Molecular Diagnostics, 2022, 24, 406-425.	1.2	1
2	A Collaborative Effort to Define Classification Criteria for <i>ATM</i> Variants in Hereditary Cancer Patients. Clinical Chemistry, 2021, 67, 518-533.	1.5	14
3	The computational approach to variant interpretation. , 2021, , 89-119.		6
4	The histone demethylase PHF8 regulates astrocyte differentiation and function. Development (Cambridge), 2021, 148, .	1.2	5
5	Towards a New, Endophenotype-Based Strategy for Pathogenicity Prediction in BRCA1 and BRCA2: In Silico Modeling of the Outcome of HDR/SGE Assays for Missense Variants. International Journal of Molecular Sciences, 2021, 22, 6226.	1.8	0
6	New genes involved in Angelman syndrome-like: Expanding the genetic spectrum. PLoS ONE, 2021, 16, e0258766.	1.1	13
7	Loss of microRNA-135b Enhances Bone Metastasis in Prostate Cancer and Predicts Aggressiveness in Human Prostate Samples. Cancers, 2021, 13, 6202.	1.7	8
8	Molecular analysis of the novel L243R mutation in STXBP2 reveals impairment of degranulation activity. International Journal of Hematology, 2020, 111, 440-450.	0.7	2
9	FHLdb: A Comprehensive Database on the Molecular Basis of Familial Hemophagocytic Lymphohistiocytosis. Frontiers in Immunology, 2020, 11, 107.	2.2	4
10	Assessment of blind predictions of the clinical significance of <i>BRCA1</i> and <i>BRCA2</i> variants. Human Mutation, 2019, 40, 1546-1556.	1.1	19
11	Compensated pathogenic variants in coagulation factors VIII and IX present complex mapping between molecular impact and hemophilia severity. Scientific Reports, 2019, 9, 9538.	1.6	3
12	Front Cover, Volume 40, Issue 9. Human Mutation, 2019, 40, i.	1.1	0
13	PHF2 histone demethylase prevents DNA damage and genome instability by controlling cell cycle progression of neural progenitors. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 19464-19473.	3.3	35
14	BRCA1 ―and BRCA2 â€specific in silico tools for variant interpretation in the CAGI 5 ENIGMA challenge. Human Mutation, 2019, 40, 1593-1611.	1.1	11
15	Increased dNTP pools rescue mtDNA depletion in human POLGâ€deficient fibroblasts. FASEB Journal, 2019, 33, 7168-7179.	0.2	14
16	Structural and Computational Characterization of Disease-Related Mutations Involved in Protein-Protein Interfaces. International Journal of Molecular Sciences, 2019, 20, 1583.	1.8	17
17	F5EPIGENETIC SIGNATURE FOR ATTENTION DEFICIT HYPERACTIVITY DISORDER: IDENTIFICATION OF MIR-23A-5P, MIR-26B-5P, MIR-185-5P AND MIR-191-5P AS A POTENTIAL BIOMARKER IN PERIPHERAL BLOOD MONONUCLEAR CELLS. European Neuropsychopharmacology, 2019, 29, S1112.	0.3	0
18	Epigenetic signature for attention-deficit/hyperactivity disorder: identification of miR-26b-5p, miR-185-5p, and miR-191-5p as potential biomarkers in peripheral blood mononuclear cells. Neuropsychopharmacology, 2019, 44, 890-897.	2.8	31

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19	Lineage specific transcription factors and epigenetic regulators mediate TGFÎ ² -dependent enhancer activation. Nucleic Acids Research, 2018, 46, 3351-3365.	6.5	24
20	Improving the diagnosis of cobalamin and related defects by genomic analysis, plus functional and structural assessment of novel variants. Orphanet Journal of Rare Diseases, 2018, 13, 125.	1.2	3
21	The histone demethylase PHF8 is a molecular safeguard of the IFNÎ ³ response. Nucleic Acids Research, 2017, 45, gkw1346.	6.5	12
22	Elucidating the clinical significance of two PMS2 missense variants coexisting in a family fulfilling hereditary cancer criteria. Familial Cancer, 2017, 16, 501-507.	0.9	3
23	PMut: a web-based tool for the annotation of pathological variants on proteins, 2017 update. Nucleic Acids Research, 2017, 45, W222-W228.	6.5	184
24	Elucidating the molecular basis of MSH2â€deficient tumors by combined germline and somatic analysis. International Journal of Cancer, 2017, 141, 1365-1380.	2.3	26
25	Early Versus Late Diagnosis of Complement Factor I Deficiency: Clinical Consequences Illustrated in Two Families with Novel Homozygous CFI Mutations. Journal of Clinical Immunology, 2017, 37, 781-789.	2.0	13
26	Identification and characterization of the novel point mutation m.3634A>G in the mitochondrial MT - ND1 gene associated with LHON syndrome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 182-187.	1.8	8
27	Development of pathogenicity predictors specific for variants that do not comply with clinical guidelines for the use of computational evidence. BMC Genomics, 2017, 18, 569.	1.2	14
28	EZH2 regulates neuroepithelium structure and neuroblast proliferation by repressing p21. Open Biology, 2016, 6, 150227.	1.5	30
29	Novel Mutations Causing C5 Deficiency in Three North-African Families. Journal of Clinical Immunology, 2016, 36, 388-396.	2.0	13
30	The Complementarity Between Protein-Specific and General Pathogenicity Predictors for Amino Acid Substitutions. Human Mutation, 2016, 37, 1013-1024.	1.1	42
31	Clinical and structural impact of mutations affecting the residue Phe367 of FOXP3 in patients with IPEX syndrome. Clinical Immunology, 2016, 163, 60-65.	1.4	14
32	Functional consequences of transferrin receptorâ€⊋ mutations causing hereditary hemochromatosis type 3. Molecular Genetics & Genomic Medicine, 2015, 3, 221-232.	0.6	19
33	Cerebrospinal fluid-derived circulating tumour DNA better represents the genomic alterations of brain tumours than plasma. Nature Communications, 2015, 6, 8839.	5.8	605
34	Molecular damage in <scp>F</scp> abry disease: Characterization and prediction of alphaâ€galactosidase <scp>A</scp> pathological mutations. Proteins: Structure, Function and Bioinformatics, 2015, 83, 91-104.	1.5	23
35	Molecular Dynamics Study of Naturally Existing Cavity Couplings in Proteins. PLoS ONE, 2015, 10, e0119978.	1.1	10
36	Prediction of pathological mutations in proteins: the challenge of integrating sequence conservation and structure stability principles. Wiley Interdisciplinary Reviews: Computational Molecular Science, 2014, 4, 249-268.	6.2	19

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37	Identification and characterization of a novel splice site mutation in the SERPING1 gene in a family with hereditary angioedema. Clinical Immunology, 2014, 150, 143-148.	1.4	10
38	The Relationship between Gene Isoform Multiplicity, Number of Exons and Protein Divergence. PLoS ONE, 2013, 8, e72742.	1.1	5
39	Genome-wide analysis reveals that Smad3 and JMJD3 HDM co-activate the neural developmental program. Development (Cambridge), 2012, 139, 2681-2691.	1.2	100
40	Alternative Splicing as a Source of Phenotypic Differences Between Species: Protein-Level Mechanisms. , 2012, , 343-356.		1
41	Characterization of the impact of alternative splicing on protein dynamics: The cases of glutathione Sâ€ŧransferase and ectodysplasinâ€A isoforms. Proteins: Structure, Function and Bioinformatics, 2012, 80, 2235-2249.	1.5	5
42	Scoring by Intermolecular Pairwise Propensities of Exposed Residues (SIPPER): A New Efficient Potential for Proteinâ^'Protein Docking. Journal of Chemical Information and Modeling, 2011, 51, 370-377.	2.5	70
43	Characterization of structural variability sheds light on the specificity determinants of the interaction between effector domains and histone tails. Epigenetics, 2010, 5, 137-148.	1.3	7
44	Autoacetylation Regulates P/CAF Nuclear Localization. Journal of Biological Chemistry, 2009, 284, 1343-1352.	1.6	62
45	Alternative Splicing of Transcription Factors' Genes: Beyond the Increase of Proteome Diversity. Comparative and Functional Genomics, 2009, 2009, 1-6.	2.0	14
46	An Atomistic View to the Gas Phase Proteome. Structure, 2009, 17, 88-95.	1.6	44
47	Preservation of protein clefts in comparative models. BMC Structural Biology, 2008, 8, 2.	2.3	11
48	The (In)dependence of Alternative Splicing and Gene Duplication. PLoS Computational Biology, 2007, 3, e33.	1.5	66
49	Involvement of chromatin and histone deacetylation in SV40 T antigen transcription regulation. Nucleic Acids Research, 2007, 35, 1958-1968.	6.5	20
50	Characterization of Compensated Mutations in Terms of Structural and Physico-Chemical Properties. Journal of Molecular Biology, 2007, 365, 249-256.	2.0	43
51	A procedure for identifying homologous alternative splicing events. BMC Bioinformatics, 2007, 8, 260.	1.2	5
52	The functional modulation of epigenetic regulators by alternative splicing. BMC Genomics, 2007, 8, 252.	1.2	14
53	Data Mining of Molecular Dynamics Trajectories of Nucleic Acids. Journal of Biomolecular Structure and Dynamics, 2006, 23, 447-455.	2.0	12
54	A fast method for the determination of fractional contributions to solvation in proteins. Protein Science, 2006, 15, 2525-2533.	3.1	3

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55	Do protein motifs read the histone code?. BioEssays, 2005, 27, 164-175.	1.2	220
56	Use of bioinformatics tools for the annotation of disease-associated mutations in animal models. Proteins: Structure, Function and Bioinformatics, 2005, 61, 878-887.	1.5	19
57	PMUT: a web-based tool for the annotation of pathological mutations on proteins. Bioinformatics, 2005, 21, 3176-3178.	1.8	441
58	PupasView: a visual tool for selecting suitable SNPs, with putative pathological effect in genes, for genotyping purposes. Nucleic Acids Research, 2005, 33, W501-W505.	6.5	253
59	Exploring the Essential Dynamics of B-DNA. Journal of Chemical Theory and Computation, 2005, 1, 790-800.	2.3	61
60	Triplex-forming oligonucleotide target sequences in the human genome. Nucleic Acids Research, 2004, 32, 354-360.	6.5	149
61	Linear response theory: An alternative to PB and GB methods for the analysis of molecular dynamics trajectories?. Proteins: Structure, Function and Bioinformatics, 2004, 57, 458-467.	1.5	8
62	Sequence-based prediction of pathological mutations. Proteins: Structure, Function and Bioinformatics, 2004, 57, 811-819.	1.5	156
63	Partition of protein solvation into group contributions from molecular dynamics simulations. Proteins: Structure, Function and Bioinformatics, 2004, 58, 101-109.	1.5	8
64	Exploring the Binding Mode of Semicarbazide-Sensitive Amine Oxidase/VAP-1:Â Identification of Novel Substrates with Insulin-like Activity. Journal of Medicinal Chemistry, 2004, 47, 4865-4874.	2.9	27
65	Alternative Splicing Mechanisms for the Modulation of Protein Function: Conservation Between Human and Other Species. Journal of Molecular Biology, 2004, 335, 495-502.	2.0	21
66	The SV40 T antigen modulates CBP histone acetyltransferase activity. Nucleic Acids Research, 2003, 31, 3114-3122.	6.5	34
67	Toward predicting protein topology: An approach to identifying hairpins. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 11157-11162.	3.3	48
68	hSos1 Contains a New Amino-terminal Regulatory Motif with Specific Binding Affinity for Its Pleckstrin Homology Domain. Journal of Biological Chemistry, 2002, 277, 44171-44179.	1.6	25
69	Characterization of disease-associated single amino acid polymorphisms in terms of sequence and structure properties 1 1Edited by J. Thornton. Journal of Molecular Biology, 2002, 315, 771-786.	2.0	194
70	Use of structure comparison methods for the refinement of protein structure predictions. I. Identifying the structural family of a protein from low-resolution models. Proteins: Structure, Function and Bioinformatics, 2002, 46, 72-84.	1.5	4
71	Classical molecular interaction potentials: Improved setup procedure in molecular dynamics simulations of proteins. Proteins: Structure, Function and Bioinformatics, 2001, 45, 428-437.	1.5	87

Use of surface area computations to describe atom-atom interactions. , 2001, 15, 521-532.

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73	Factors limiting the performance of predictionâ€based fold recognition methods. Protein Science, 1999, 8, 750-759.	3.1	18
74	A new procedure for constructing peptides into a given C. Folding & Design, 1998, 3, 1-10.	4.5	22
75	Discrete representations of the protein Cα chain. Folding & Design, 1997, 2, 223-234.	4.5	8
76	The structural homology between uteroglobin and the poreâ€forming domain of colicin A suggests a possible mechanism of action for uteroglobin. Protein Science, 1996, 5, 857-861.	3.1	10
77	Investigation of shape variations in the antibody binding site by molecular dynamics computer simulation. Journal of Molecular Biology, 1994, 236, 1186-1195.	2.0	16
78	Representation of noncovalent interactions in protein structures. Journal of Molecular Graphics, 1992, 10, 96-100.	1.7	8
79	Atomic accessible and contact surfaces as restraints in the Hendrickson & Konnert refinement program. Journal of Applied Crystallography, 1991, 24, 941-946.	1.9	4