

# Emanuela Leonardi

## List of Publications by Year in descending order

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Version: 2024-02-01

44  
papers

1,773  
citations

331259

21  
h-index

288905

40  
g-index

47  
all docs

47  
docs citations

47  
times ranked

2838  
citing authors

#	ARTICLE	IF	CITATIONS
1	DisProt in 2022: improved quality and accessibility of protein intrinsic disorder annotation. <i>Nucleic Acids Research</i> , 2022, 50, D480-D487.	6.5	117
2	Feasibility of Screening for Chromosome 15 Imprinting Disorders in 16 579 Newborns by Using a Novel Genomic Workflow. <i>JAMA Network Open</i> , 2022, 5, e2141911.	2.8	14
3	Molecular Effects of Mutations in Human Genetic Diseases. <i>International Journal of Molecular Sciences</i> , 2022, 23, 6408.	1.8	0
4	A Missense De Novo Variant in the CASK-interactor KIRREL3 Gene Leading to Neurodevelopmental Disorder with Mild Cerebellar Hypoplasia. <i>Neuropediatrics</i> , 2021, 52, 484-488.	0.3	3
5	DisProt: intrinsic protein disorder annotation in 2020. <i>Nucleic Acids Research</i> , 2020, 48, D269-D276.	6.5	141
6	Expanding the genetic landscape of Rett syndrome to include lysine acetyltransferase 6A (KAT6A). <i>Journal of Genetics and Genomics</i> , 2020, 47, 650-654.	1.7	2
7	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). <i>European Journal of Medical Genetics</i> , 2020, 63, 104004.	0.7	7
8	Frequency of Usher gene mutations in non-syndromic hearing loss: higher variability of the Usher phenotype. <i>Journal of Human Genetics</i> , 2020, 65, 855-864.	1.1	6
9	A Novel WAC Loss of Function Mutation in an Individual Presenting with Encephalopathy Related to Status Epilepticus during Sleep (ESES). <i>Genes</i> , 2020, 11, 344.	1.0	14
10	Identification of SETBP1 Mutations by Gene Panel Sequencing in Individuals With Intellectual Disability or With Developmental and Epileptic Encephalopathy. <i>Frontiers in Neurology</i> , 2020, 11, 593446.	1.1	10
11	Assessing computational predictions of the phenotypic effect of cystathionine beta synthase variants. <i>Human Mutation</i> , 2019, 40, 1530-1545.	1.1	5
12	Performance of computational methods for the evaluation of pericentriolar material 1 missense variants in CAGI5. <i>Human Mutation</i> , 2019, 40, 1474-1485.	1.1	8
13	Characterization of intellectual disability and autism comorbidity through gene panel sequencing. <i>Human Mutation</i> , 2019, 40, 1346-1363.	1.1	54
14	Assessment of patient clinical descriptions and pathogenic variants from gene panel sequences in the CAGI5 intellectual disability challenge. <i>Human Mutation</i> , 2019, 40, 1330-1345.	1.1	11
15	CNTNAP2 mutations and autosomal dominant epilepsy with auditory features. <i>Epilepsy Research</i> , 2018, 139, 51-53.	0.8	3
16	Crohn disease risk prediction Best practices and pitfalls with exome data. <i>Human Mutation</i> , 2017, 38, 1193-1200.	1.1	12
17	Performance of in silico tools for the evaluation of p16INK4a (CDKN2A) variants in CAGI. <i>Human Mutation</i> , 2017, 38, 1042-1050.	1.1	13
18	DisProt 7.0: a major update of the database of disordered proteins. <i>Nucleic Acids Research</i> , 2017, 45, D219-D227.	6.5	242

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19	Working toward precision medicine: Predicting phenotypes from exomes in the Critical Assessment of Genome Interpretation (CAGI) challenges. <i>Human Mutation</i> , 2017, 38, 1182-1192.	1.1	39
20	Matching phenotypes to whole genomes: Lessons learned from four iterations of the personal genome project community challenges. <i>Human Mutation</i> , 2017, 38, 1266-1276.	1.1	14
21	Dynamic scaffolds for neuronal signaling: in silico analysis of the TANC protein family. <i>Scientific Reports</i> , 2017, 7, 6829.	1.6	21
22	Lessons from the CAGI's Hopkins clinical panel challenge. <i>Human Mutation</i> , 2017, 38, 1155-1168.	1.1	6
23	Mapping pathogenic mutations suggests an innovative structural model for the pendrin (SLC26A4) transmembrane domain. <i>Biochimie</i> , 2017, 132, 109-120.	1.3	19
24	Secretion-Positive LGI1 Mutations Linked to Lateral Temporal Epilepsy Impair Binding to ADAM22 and ADAM23 Receptors. <i>PLoS Genetics</i> , 2016, 12, e1006376.	1.5	23
25	VHLdb: A database of von Hippel-Lindau protein interactors and mutations. <i>Scientific Reports</i> , 2016, 6, 31128.	1.6	36
26	INGA: protein function prediction combining interaction networks, domain assignments and sequence similarity. <i>Nucleic Acids Research</i> , 2015, 43, W134-W140.	6.5	73
27	BOOGIE: Predicting Blood Groups from High Throughput Sequencing Data. <i>PLoS ONE</i> , 2015, 10, e0124579.	1.1	31
28	Identification of Four Novel <i>PCDH19</i> Mutations and Prediction of Their Functional Impact. <i>Annals of Human Genetics</i> , 2014, 78, 389-398.	0.3	17
29	Early-onset epileptic encephalopathy in a girl carrying a truncating mutation of the <i>ARX</i> gene: rethinking the <i>ARX</i> phenotype in females. <i>Clinical Genetics</i> , 2013, 84, 82-85.	1.0	13
30	A novel <i>SACS</i> mutation results in non-ataxic spastic paraplegia and peripheral neuropathy. <i>European Journal of Neurology</i> , 2013, 20, 1486-1491.	1.7	30
31	Fly cryptochrome and the visual system. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 6163-6168.	3.3	103
32	2mit, an Intronic Gene of <i>Drosophila melanogaster timeless2</i> , Is Involved in Behavioral Plasticity. <i>PLoS ONE</i> , 2013, 8, e76351.	1.1	9
33	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. <i>European Journal of Human Genetics</i> , 2012, 20, 1234-1239.	1.4	31
34	Identification and In Silico Analysis of Novel von Hippel-Lindau (VHL) Gene Variants from a Large Population. <i>Annals of Human Genetics</i> , 2011, 75, 483-496.	0.3	19
35	Familial temporal lobe epilepsy with psychic auras associated with a novel <i>LGI1</i> mutation. <i>Neurology</i> , 2011, 76, 1173-1176.	1.5	49
36	A Computational Model of the LGI1 Protein Suggests a Common Binding Site for ADAM Proteins. <i>PLoS ONE</i> , 2011, 6, e18142.	1.1	33

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37	Spectrum and Frequency of <i>SLC26A4</i> Mutations Among Czech Patients with Early Hearing Loss with and without Enlarged Vestibular Aqueduct (EVA). <i>Annals of Human Genetics</i> , 2010, 74, 299-307.	0.3	35
38	Deletions and Mutations in the Acidic Lipid-binding Region of the Plasma Membrane Ca <sup>2+</sup> Pump. <i>Journal of Biological Chemistry</i> , 2010, 285, 30779-30791.	1.6	22
39	A Novel WT1 Gene Mutation in a Three-Generation Family with Progressive Isolated Focal Segmental Glomerulosclerosis. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2010, 5, 698-702.	2.2	30
40	Adding structural information to the von Hippel-Lindau (VHL) tumor suppressor interaction network. <i>FEBS Letters</i> , 2009, 583, 3704-3710.	1.3	25
41	Molecular Characterization of Large Deletions in the von Hippel-Lindau (VHL) Gene by Quantitative Real-Time PCR. <i>Molecular Diagnosis and Therapy</i> , 2006, 10, 243-249.	1.6	24
42	A novel deletion involving the connexin-30 gene, del(GJB6-d13s1854), found in trans with mutations in the GJB2 gene (connexin-26) in subjects with DFNB1 non-syndromic hearing impairment. <i>Journal of Medical Genetics</i> , 2005, 42, 588-594.	1.5	282
43	Connexin <sup>26</sup> 35delG does not represent a mutational hotspot. <i>Human Genetics</i> , 2003, 113, 18-23.	1.8	46
44	Molecular Genetics Applied to Clinical Practice: The Cx26 Hearing Impairment. <i>International Journal of Audiology</i> , 1999, 33, 291-295.	0.7	32