Emanuela Leonardi

List of Publications by Year
in descending order

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	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. Journal of Medical Genetics, 2005, 42, 588-594.	1.5	282
2	DisProt 7.0: a major update of the database of disordered proteins. Nucleic Acids Research, 2017, 45, D219-D227.	6.5	242
3	DisProt: intrinsic protein disorder annotation in 2020. Nucleic Acids Research, 2020, 48, D269-D276.	6.5	141
4	DisProt in 2022: improved quality and accessibility of protein intrinsic disorder annotation. Nucleic Acids Research, 2022, 50, D480-D487.	6.5	117
5	Fly cryptochrome and the visual system. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 6163-6168.	3.3	103
6	INGA: protein function prediction combining interaction networks, domain assignments and sequence similarity. Nucleic Acids Research, 2015, 43, W134-W140.	6.5	73
7	Characterization of intellectual disability and autism comorbidity through gene panel sequencing. Human Mutation, 2019, 40, 1346-1363.	1.1	54
8	Familial temporal lobe epilepsy with psychic auras associated with a novel LGI1 mutation. Neurology, 2011, 76, 1173-1176.	1.5	49
9	ConnexinÂ26 35delG does not represent a mutational hotspot. Human Genetics, 2003, 113, 18-23.	1.8	46
10	Working toward precision medicine: Predicting phenotypes from exomes in the Critical Assessment of Genome Interpretation (CAGI) challenges. Human Mutation, 2017, 38, 1182-1192.	1.1	39
11	VHLdb: A database of von Hippel-Lindau protein interactors and mutations. Scientific Reports, 2016, 6, 31128.	1.6	36
12	Spectrum and Frequency of SLC26A4 Mutations Among Czech Patients with Early Hearing Loss with and without Enlarged Vestibular Aqueduct (EVA). Annals of Human Genetics, 2010, 74, 299-307.	0.3	35
13	A Computational Model of the LGI1 Protein Suggests a Common Binding Site for ADAM Proteins. PLoS ONE, 2011, 6, e18142.	1.1	33
14	Molecular Genetics Applied to Clinical Practice: The Cx26 Hearing Impairment. International Journal of Audiology, 1999, 33, 291-295.	0.7	32
15	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. European Journal of Human Genetics, 2012, 20, 1234-1239.	1.4	31
16	BOOGIE: Predicting Blood Groups from High Throughput Sequencing Data. PLoS ONE, 2015, 10, e0124579.	1.1	31
17	A Novel WT1 Gene Mutation in a Three-Generation Family with Progressive Isolated Focal Segmental Glomerulosclerosis. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 698-702.	2.2	30
18	A novel <scp>SACS</scp> mutation results in nonâ€ataxic spastic paraplegia and peripheral neuropathy. European Journal of Neurology, 2013, 20, 1486-1491.	1.7	30

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19	Adding structural information to the von Hippelâ€“Lindau (VHL) tumor suppressor interaction network. FEBS Letters, 2009, 583, 3704-3710.	1.3	25
20	Molecular Characterization of Large Deletions in the von Hippel-Lindau (VHL) Gene by Quantitative Real-Time PCR. Molecular Diagnosis and Therapy, 2006, 10, 243-249.	1.6	24
21	Secretion-Positive LGI1 Mutations Linked to Lateral Temporal Epilepsy Impair Binding to ADAM22 and ADAM23 Receptors. PLoS Genetics, 2016, 12, e1006376.	1.5	23
22	Deletions and Mutations in the Acidic Lipid-binding Region of the Plasma Membrane Ca2+ Pump. Journal of Biological Chemistry, 2010, 285, 30779-30791.	1.6	22
23	Dynamic scaffolds for neuronal signaling: in silico analysis of the TANC protein family. Scientific Reports, 2017, 7, 6829.	1.6	21
24	Identification and In Silico Analysis of Novel von Hippel-Lindau (VHL) Gene Variants from a Large Population. Annals of Human Genetics, 2011, 75, 483-496.	0.3	19
25	Mapping pathogenic mutations suggests an innovative structural model for the pendrin (SLC26A4) transmembrane domain. Biochimie, 2017, 132, 109-120.	1.3	19
26	Identification of Four NovelPCDH19Mutations and Prediction of Their Functional Impact. Annals of Human Genetics, 2014, 78, 389-398.	0.3	17
27	Matching phenotypes to whole genomes: Lessons learned from four iterations of the personal genome project community challenges. Human Mutation, 2017, 38, 1266-1276.	1.1	14
28	A Novel WAC Loss of Function Mutation in an Individual Presenting with Encephalopathy Related to Status Epilepticus during Sleep (ESES). Genes, 2020, 11, 344.	1.0	14
29	Feasibility of Screening for Chromosome 15 Imprinting Disorders in 16â€¯579 Newborns by Using a Novel Genomic Workflow. JAMA Network Open, 2022, 5, e2141911.	2.8	14
30	Earlyâ€onset epileptic encephalopathy in a girl carrying a truncating mutation of the <scp>ARX</scp> gene: rethinking the <scp>ARX</scp> phenotype in females. Clinical Genetics, 2013, 84, 82-85.	1.0	13
31	Performance of in silico tools for the evaluation of p16INK4a (CDKN2A) variants in CAGI. Human Mutation, 2017, 38, 1042-1050.	1.1	13
32	Crohn disease risk predictionâ€"Best practices and pitfalls with exome data. Human Mutation, 2017, 38, 1193-1200.	1.1	12
33	Assessment of patient clinical descriptions and pathogenic variants from gene panel sequences in the CAGIâ€5 intellectual disability challenge. Human Mutation, 2019, 40, 1330-1345.	1.1	11
34	Identification of SETBP1 Mutations by Gene Panel Sequencing in Individuals With Intellectual Disability or With â€œDevelopmental and Epileptic Encephalopathyâ€• Frontiers in Neurology, 2020, 11, 593446.	1.1	10
35	2mit, an Intronic Gene of Drosophila melanogaster timeless2, Is Involved in Behavioral Plasticity. PLoS ONE, 2013, 8, e76351.	1.1	9
36	Performance of computational methods for the evaluation of pericentriolar material 1 missense variants in CAGIâ€5. Human Mutation, 2019, 40, 1474-1485.	1.1	8

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37	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	0.7	7
38	Lessons from the CAGIâ€4 Hopkins clinical panel challenge. Human Mutation, 2017, 38, 1155-1168.	1.1	6
39	Frequency of Usher gene mutations in non-syndromic hearing loss: higher variability of the Usher phenotype. Journal of Human Genetics, 2020, 65, 855-864.	1.1	6
40	Assessing computational predictions of the phenotypic effect of cystathionineâ€betaâ€synthase variants. Human Mutation, 2019, 40, 1530-1545.	1.1	5
41	A Missense De Novo Variant in the CASK-interactor KIRREL3 Gene Leading to Neurodevelopmental Disorder with Mild Cerebellar Hypoplasia. Neuropediatrics, 2021, 52, 484-488.	0.3	3
42	CNTNAP2 mutations and autosomal dominant epilepsy with auditory features. Epilepsy Research, 2018, 139, 51-53.	0.8	3
43	Expanding the genetic landscape of Rett syndrome to include lysine acetyltransferase 6A (KAT6A). Journal of Genetics and Genomics, 2020, 47, 650-654.	1.7	2
44	Molecular Effects of Mutations in Human Genetic Diseases. International Journal of Molecular Sciences, 2022, 23, 6408.	1.8	0