Emanuele Bellacchio

List of Publications by Year
in descending order

88	3,878	196777 29	145109 60
papers	citations	h-index	g-index

90	90	90	7557
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Intramolecular Interaction with the E6 Region Stabilizes the Closed Conformation of the N-SH2 Domain and Concurs with the Self-Inhibitory Docking in Downregulating the Activity of the SHP2 Tyrosine Phosphatase: A Molecular Dynamics Study. International Journal of Molecular Sciences, 2022, 23, 4794.	1.8	1
2	Clinical and molecular characterization of patients with adenylosuccinate lyase deficiency. Orphanet Journal of Rare Diseases, 2021, 16, 112.	1.2	12
3	Ectodermal Dysplasia-Syndactyly Syndrome with Toe-Only Minimal Syndactyly Due to a Novel Mutation in NECTIN4: A Case Report and Literature Review. Genes, 2021, 12, 748.	1.0	3
4	Novel KCND3 Variant Underlying Nonprogressive Congenital Ataxia or SCA19/22 Disrupt KV4.3 Protein Expression and K+ Currents with Variable Effects on Channel Properties. International Journal of Molecular Sciences, 2021, 22, 4986.	1.8	12
5	The Recruitment-Secretory Block (â€œR-SBâ€) Phenomenon and Endoplasmic Reticulum Storage Diseases. International Journal of Molecular Sciences, 2021, 22, 6807.	1.8	1
6	Homozygous HESX1 and COL1A1 Gene Variants in a Boy with Growth Hormone Deficiency and Early Onset Osteoporosis. International Journal of Molecular Sciences, 2021, 22, 750.	1.8	1
7	Hereditary spastic paraplegia is a novel phenotype for germline de novo ATP1A1 mutation. Clinical Genetics, 2020, 97, 521-526.	1.0	14
8	Genetic identification and molecular modeling characterization of a novel POU3F4 variant in two Italian deaf brothers. International Journal of Pediatric Otorhinolaryngology, 2020, 129, 109790.	0.4	8
9	Infantile-Onset Syndromic Cerebellar Ataxia and CACNA1G Mutations. Pediatric Neurology, 2020, 104, 40-45.	1.0	17
10	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
11	Structural Characteristics in the Î³ Chain Variants Associated with Fibrinogen Storage Disease Suggest the Underlying Pathogenic Mechanism. International Journal of Molecular Sciences, 2020, 21, 5139.	1.8	11
12	Mutations Causing Mild or No Structural Damage in Interfaces of Multimerization of the Fibrinogen Î³-Module More Likely Confer Negative Dominant Behaviors. International Journal of Molecular Sciences, 2020, 21, 9016.	1.8	3
13	Very mild isolated intellectual disability caused by adenylosuccinate lyase deficiency: a new phenotype. Molecular Genetics and Metabolism Reports, 2020, 23, 100592.	0.4	4
14	TUBB Variants Underlying Different Phenotypes Result in Altered Vesicle Trafficking and Microtubule Dynamics. International Journal of Molecular Sciences, 2020, 21, 1385.	1.8	20
15	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. American Journal of Human Genetics, 2019, 105, 640-657.	2.6	31
16	Heterozygous missense variants of SPTBN2 are a frequent cause of congenital cerebellar ataxia. Clinical Genetics, 2019, 96, 169-175.	1.0	27
17	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. Cerebellum, 2019, 18, 422-432.	1.4	10
18	Clinical-genetic features and peculiar muscle histopathology in infantileDNM1L-related mitochondrial epileptic encephalopathy. Human Mutation, 2019, 40, 601-618.	1.1	31

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19	Heme and sensory neuropathy: insights from novel mutations in the heme exporter feline leukemia virus subgroup C receptor 1. Pain, 2019, 160, 2766-2775.	2.0	16
20	Dominant Noonan syndrome-causing LZTR1 mutations specifically affect the Kelch domain substrate-recognition surface and enhance RAS-MAPK signaling. Human Molecular Genetics, 2019, 28, 1007-1022.	1.4	58
21	Novel exostosinâ€2 missense variants in a family with autosomal recessive exostosinâ€2â€related syndrome: further evidences on the phenotype. Clinical Genetics, 2019, 95, 165-171.	1.0	3
22	Novel Homozygous KCNJ10 Mutation in a Patient with Non-syndromic Early-Onset Cerebellar Ataxia. Cerebellum, 2018, 17, 499-503.	1.4	10
23	Proliferative vasculopathy and hydranencephalyâ€“hydrocephaly syndrome or Fowler syndrome: Report of a family and insight into the disease's mechanism. Molecular Genetics & Genomic Medicine, 2018, 6, 446-451.	0.6	11
24	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	1.1	22
25	Expanding the histopathological spectrum of CFL2â€related myopathies. Clinical Genetics, 2018, 93, 1234-1239.	1.0	11
26	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Human Molecular Genetics, 2018, 27, 1892-1904.	1.4	29
27	Expanding the clinical phenotype of IARS2-related mitochondrial disease. BMC Medical Genetics, 2018, 19, 196.	2.1	16
28	Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846.	1.5	44
29	Next-Generation Sequencing Identifies Different Genetic Defects in 2 Patients with Primary Adrenal Insufficiency and Gonadotropin-Independent Precocious Puberty. Hormone Research in Paediatrics, 2018, 90, 203-211.	0.8	11
30	A Novel Homozygous Mutation of the AIRE Gene in an APECED Patient From Pakistan: Case Report and Review of the Literature. Frontiers in Immunology, 2018, 9, 1835.	2.2	7
31	Mineralization of alpha-1-antitrypsin inclusion bodies in Mmalton alpha-1-antitrypsin deficiency. Orphanet Journal of Rare Diseases, 2018, 13, 79.	1.2	13
32	Blood malignancies presenting with mutations at equivalent residues in RUNX1â€“2 suggest a common leukemogenic pathway. Leukemia and Lymphoma, 2017, 58, 2002-2004.	0.6	4
33	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. Brain, 2017, 140, e34-e34.	3.7	17
34	Novel mutations in <scp>KARS</scp> cause hypertrophic cardiomyopathy and combined mitochondrial respiratory chain defect. Clinical Genetics, 2017, 91, 918-923.	1.0	27
35	Expanding the clinical phenotype of CAPN1- associated mutations: A new case with congenital-onset pure spastic paraplegia. Journal of the Neurological Sciences, 2017, 378, 210-212.	0.3	21
36	Missense mutations of CACNA1A are a frequent cause of autosomal dominant nonprogressive congenital ataxia. European Journal of Paediatric Neurology, 2017, 21, 450-456.	0.7	37

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37	A novel fibrinogen gamma chain mutation (c.1096C>G; p.His340Asp), fibrinogen Ankara, causing hypofibrinogenaemia and hepatic storage. Pathology, 2017, 49, 534-537.	0.3	12
38	Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome. Gene, 2017, 628, 141-145.	1.0	27
39	A mild form of adenylosuccinate lyase deficiency in absence of typical brain MRI features diagnosed by whole exome sequencing. Italian Journal of Pediatrics, 2017, 43, 65.	1.0	9
40	Fibrinogen Gamma Chain Mutations Provoke Fibrinogen and Apolipoprotein B Plasma Deficiency and Liver Storage. International Journal of Molecular Sciences, 2017, 18, 2717.	1.8	17
41	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	2.6	49
42	Acute myeloid leukemia in a 3 years old child with cleidocranial dysplasia. Leukemia and Lymphoma, 2016, 57, 2189-2191.	0.6	9
43	A novel AIFM1 mutation expands the phenotype to an infantile motor neuron disease. European Journal of Human Genetics, 2016, 24, 463-466.	1.4	51
44	A Novel Mutation inRPL10(Ribosomal Protein L10) Causes X-Linked Intellectual Disability, Cerebellar Hypoplasia, and Spondylo-Epiphyseal Dysplasia. Human Mutation, 2015, 36, 1155-1158.	1.1	28
45	Genetics and Molecular Modeling of New Mutations of Familial Intrahepatic Cholestasis in a Single Italian Center. PLoS ONE, 2015, 10, e0145021.	1.1	18
46	Kabuki syndrome: clinical and molecular diagnosis in the first year of life. Archives of Disease in Childhood, 2015, 100, 158-164.	1.0	69
47	The possible implication of the S250C variant of the autoimmune regulator protein in a patient with autoimmunity and immunodeficiency: in silico analysis suggests a molecular pathogenic mechanism for the variant. Gene, 2014, 549, 286-294.	1.0	13
48	Muscle magnetic resonance imaging and histopathology in ACTA1â€related congenital nemaline myopathy. Muscle and Nerve, 2014, 50, 1011-1016.	1.0	15
49	The human papillomavirus-16 E7 oncoprotein exerts antiapoptotic effects via its physical interaction with the actin-binding protein gelsolin. Carcinogenesis, 2013, 34, 2424-2433.	1.3	9
50	Understanding the targeting of the RB family proteins by viral oncoproteins to defeat their oncogenic machinery. Journal of Cellular Physiology, 2013, 228, 285-291.	2.0	26
51	A new de novo missense mutation in MYH2 expands clinical and genetic findings in hereditary myosin myopathies. Neuromuscular Disorders, 2013, 23, 437-440.	0.3	17
52	Description of a novel TUBA1A mutation in Arg-390 associated with asymmetrical polymicrogyria and mid-hindbrain dysgenesis. European Journal of Paediatric Neurology, 2013, 17, 361-365.	0.7	23
53	Bile salt export pump deficiency: A de novo mutation in a child compound heterozygous for ABCB11. Laboratory investigation to study pathogenic role and transmission of two novel ABCB11 mutations. Hepatology Research, 2013, 43, 315-319.	1.8	6
54	Targeting Proliferating Cell Nuclear Antigen and Its Protein Interactions Induces Apoptosis in Multiple Myeloma Cells. PLoS ONE, 2013, 8, e70430.	1.1	77

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55	Mechanism of Neurotoxicity of Prion and Alzheimer's Disease-Related Proteins: Molecular Insights from Bioinformatically Identified Ï‰-Conotoxin-Like Pharmacophores. Critical Reviews in Eukaryotic Gene Expression, 2013, 23, 355-374.	0.4	3
56	ATM gene alterations in chronic lymphocytic leukemia patients induce a distinct gene expression profile and predict disease progression. Haematologica, 2012, 97, 47-55.	1.7	92
57	A novel heterozygous mutation of the AIRE gene in a patient with autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome (APECED). Gene, 2012, 511, 113-117.	1.0	23
58	In silico analysis of the two tandem somatomedin B domains of ENPP1 reveals hints on the homodimerization of the protein. Journal of Cellular Physiology, 2012, 227, 3566-3574.	2.0	7
59	Two novel mutations in African and Asian children with progressive familial intrahepatic cholestasis type 3. Digestive and Liver Disease, 2011, 43, 567-570.	0.4	12
60	The SH2B1 obesity locus is associated with myocardial infarction in diabetic patients and with NO synthase activity in endothelial cells. Atherosclerosis, 2011, 219, 667-672.	0.4	17
61	Mutations in PVRL4, Encoding Cell Adhesion Molecule Nectin-4, Cause Ectodermal Dysplasia-Syndactyly Syndrome. American Journal of Human Genetics, 2010, 87, 265-273.	2.6	98
62	Familial transposition of the great arteries caused by multiple mutations in laterality genes. Heart, 2010, 96, 673-677.	1.2	126
63	Human Papillomavirus-16 E7 Interacts with Glutathione S-Transferase P1 and Enhances Its Role in Cell Survival. PLoS ONE, 2009, 4, e7254.	1.1	30
64	Molecular analysis of PRKAG2, LAMP2, and NKX2â€5 genes in a cohort of 125 patients with accessory atrioventricular connection. American Journal of Medical Genetics, Part A, 2009, 149A, 1574-1577.	0.7	18
65	Mutation screening of the DYT6/THAP1 gene in Italy. Movement Disorders, 2009, 24, 2424-2427.	2.2	43
66	Sgk1 activates MDM2-dependent p53 degradation and affects cell proliferation, survival, and differentiation. Journal of Molecular Medicine, 2009, 87, 1221-1239.	1.7	88
67	Are MYO1C and MYO1F associated with hearing loss?. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 27-32.	1.8	28
68	Identification of a novel mutation in the SLC26A4 gene in an Italian with fluctuating sensorineural hearing loss. International Journal of Pediatric Otorhinolaryngology, 2009, 73, 1458-1463.	0.4	10
69	Proteaseâ€mediated arsenic prodrug strategy in cancer and infectious diseases: A hypothesis for targeted activation. Journal of Cellular Physiology, 2008, 214, 681-686.	2.0	3
70	TRIB3 R84 Variant Is Associated With Impaired Insulin-Mediated Nitric Oxide Production in Human Endothelial Cells. Arteriosclerosis, Thrombosis, and Vascular Biology, 2008, 28, 1355-1360.	1.1	53
71	Interaction Between the Cdk2/Cyclin A Complex and a Small Molecule Derived from the pRb2/p130 Spacer Domain: A Theoretical Model. Cell Cycle, 2007, 6, 2591-2593.	1.3	13
72	Redox regulation of cyclophilin A by glutathionylation. Proteomics, 2006, 6, 817-825.	1.3	43

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73	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	9.4	368
74	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	2.8	125
75	Mitochondrial import and enzymatic activity of PINK1 mutants associated to recessive parkinsonism. Human Molecular Genetics, 2005, 14, 3477-3492.	1.4	413
76	NF1 Gene Mutations Represent the Major Molecular Event Underlying Neurofibromatosis-Noonan Syndrome. American Journal of Human Genetics, 2005, 77, 1092-1101.	2.6	139
77	Mutations of the Nogo-66 receptor (RTN4R) gene in schizophrenia. Human Mutation, 2004, 24, 534-535.	1.1	77
78	Calcium EXAFS Establishes the Mn-Ca Cluster in the Oxygen-Evolving Complex of Photosystem II. Biochemistry, 2002, 41, 12928-12933.	1.2	131
79	Diffusion of gases in PEEKs membranes: molecular dynamics simulations. Journal of Membrane Science, 2002, 206, 389-398.	4.1	31
80	Absence of Mn-Centered Oxidation in the S2â† S3Transition:Â Implications for the Mechanism of Photosynthetic Water Oxidation. Journal of the American Chemical Society, 2001, 123, 7804-7820.	6.6	295
81	High-resolution X-ray spectroscopy of rare events: a different look at local structure and chemistry. Journal of Synchrotron Radiation, 2001, 8, 199-203.	1.0	45
82	Counting the number of disulfides and thiol groups in proteins and a novel approach for determining the local pKafor cysteine groups in proteinsin vivo. Journal of Synchrotron Radiation, 2001, 8, 1056-1058.	1.0	9
83	J-Type Aggregates of the Anionic Meso-Tetrakis(4-Sulfonatophenyl)Porphine Induced by â€šHinderedâ€™ Cationic Porphyrins. Supramolecular Chemistry, 2000, 12, 193-202.	1.5	42
84	Spectroscopic characterization of porphyrin supramolecular aggregates on poly-lysine and their application to quantitative DNA determination. Inorganica Chimica Acta, 1999, 286, 121-126.	1.2	27
85	Nanomolar determination of copper(II) and zinc(II) using supramolecular complexes of meso-tetrakis(4-N-methylpyridyl)porphine on polyglutamate. Chemical Communications, 1998, , 1333-1334.	2.2	21
86	Chiral H- and J-Type Aggregates of meso-Tetrakis(4-sulfonatophenyl)porphine on Î±-Helical Polyglutamic Acid Induced by Cationic Porphyrins. Inorganic Chemistry, 1998, 37, 3647-3648.	1.9	118
87	Template-Imprinted Chiral Porphyrin Aggregates. Journal of the American Chemical Society, 1998, 120, 12353-12354.	6.6	154
88	pH Modulation of Porphyrins Self-Assembly onto Polylysine. Journal of Physical Chemistry B, 1998, 102, 8852-8857.	1.2	88