

Alessandro Bruselles

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

59
papers

1,742
citations

24
h-index

41
g-index

64
ext. papers

2,148
ext. citations

6.1
avg, IF

3.81
L-index

#	Paper	IF	Citations
59	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epistatue of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021 , 108, 502-516	11	12
58	Broadening the phenotypic spectrum of Beta3GalT6-associated phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 3153-3160	2.5	0
57	Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	1
56	Expanding the clinical phenotype of the ultra-rare Skraban-Deardorff syndrome: Two novel individuals with WDR26 loss-of-function variants and a literature review. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 1712-1720	2.5	1
55	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in. <i>Genes</i> , 2021 , 12,	4.2	0
54	Refinement of the clinical and mutational spectrum of UBE2A deficiency syndrome. <i>Clinical Genetics</i> , 2020 , 98, 172-178	4	2
53	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. <i>American Journal of Human Genetics</i> , 2020 , 107, 1129-1148	11	11
52	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. <i>European Journal of Human Genetics</i> , 2020 , 28, 1602-1614	5.3	132
51	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. <i>American Journal of Human Genetics</i> , 2020 , 107, 499-513	11	25
50	Functional analysis of variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. <i>Journal of Medical Genetics</i> , 2020 ,	5.8	2
49	The activating p.Ser466Arg change in STAT1 causes a peculiar phenotype with features of interferonopathies. <i>Clinical Genetics</i> , 2019 , 96, 585-589	4	3
48	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. <i>American Journal of Human Genetics</i> , 2019 , 105, 493-508	11	30
47	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotype-phenotype correlations. <i>Human Mutation</i> , 2019 , 40, 721-728	4.7	15
46	A new bioavailable fenretinide formulation with antiproliferative, antimetabolic, and cytotoxic effects on solid tumors. <i>Cell Death and Disease</i> , 2019 , 10, 529	9.8	23
45	Organoids as a new model for improving regenerative medicine and cancer personalized therapy in renal diseases. <i>Cell Death and Disease</i> , 2019 , 10, 201	9.8	61
44	Colorectal cancer spheroid biobanks: multi-level approaches to drug sensitivity studies. <i>Cell Biology and Toxicology</i> , 2018 , 34, 459-469	7.4	12
43	The impact of next-generation sequencing on the diagnosis of pediatric-onset hereditary spastic paraplegias: new genotype-phenotype correlations for rare HSP-related genes. <i>Neurogenetics</i> , 2018 , 19, 111-121	3	28

42	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling saccinopathy. <i>Human Molecular Genetics</i> , 2018 , 27, 1892-1904	5.6	18
41	Biallelic mutations in DYNC2LI1 are a rare cause of Ellis-van Creveld syndrome. <i>Clinical Genetics</i> , 2018 , 93, 632-639	4	16
40	VarGenius executes cohort-level DNA-seq variant calling and annotation and allows to manage the resulting data through a PostgreSQL database. <i>BMC Bioinformatics</i> , 2018 , 19, 477	3.6	9
39	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. <i>American Journal of Human Genetics</i> , 2018 , 103, 621-630	11	45
38	Specific combinations of biallelic variants cause Wiedemann-Rautenstrauch syndrome. <i>Journal of Medical Genetics</i> , 2018 , 55, 837-846	5.8	31
37	Mutations in the IRBIT domain of ITPR1 are a frequent cause of autosomal dominant nonprogressive congenital ataxia. <i>Clinical Genetics</i> , 2017 , 91, 86-91	4	26
36	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. <i>EBioMedicine</i> , 2017 , 20, 39-49	8.8	112
35	Exome sequencing in children of women with skewed X-inactivation identifies atypical cases and complex phenotypes. <i>European Journal of Paediatric Neurology</i> , 2017 , 21, 475-484	3.8	6
34	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. <i>Brain</i> , 2017 , 140, 2550-2556	11.2	15
33	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. <i>European Journal of Paediatric Neurology</i> , 2017 , 21, 873-883	3.8	4
32	A syndromic extreme insulin resistance caused by biallelic mutations in exon 10. <i>European Journal of Endocrinology</i> , 2017 , 177, K21-K27	6.5	6
31	Novel - Fusion Gene in Pediatric Ependymomas Discovered by Clonal Expansion of Stem Cells in Absence of Exogenous Mitogens. <i>Cancer Research</i> , 2017 , 77, 5860-5872	10.1	14
30	Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome. <i>Gene</i> , 2017 , 628, 141-145	3.8	20
29	HIPK2-T566 autophosphorylation diversely contributes to UV- and doxorubicin-induced HIPK2 activation. <i>Oncotarget</i> , 2017 , 8, 16744-16754	3.3	5
28	Cancer Stem Cell-Based Models of Colorectal Cancer Reveal Molecular Determinants of Therapy Resistance. <i>Stem Cells Translational Medicine</i> , 2016 , 5, 511-23	6.9	35
27	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proof-of-concept examples. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 1772-9	2.5	18
26	A Follow-Up of the Multicenter Collaborative Study on HIV-1 Drug Resistance and Tropism Testing Using 454 Ultra Deep Pyrosequencing. <i>PLoS ONE</i> , 2016 , 11, e0146687	3.7	12
25	Congenital immunodeficiency in an individual with Wiedemann-Steiner syndrome due to a novel missense mutation in KMT2A. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2389-93	2.5	24

24	Expanding the molecular diversity and phenotypic spectrum of glycerol 3-phosphate dehydrogenase 1 deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2016 , 39, 689-695	5.4	18
23	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. <i>American Journal of Human Genetics</i> , 2015 , 96, 816-25	11	75
22	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. <i>European Journal of Human Genetics</i> , 2015 , 23, 1068-71	5.3	89
21	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. <i>Human Mutation</i> , 2015 , 36, 1080-7	4.7	51
20	An international multicenter study on HIV-1 drug resistance testing by 454 ultra-deep pyrosequencing. <i>Journal of Virological Methods</i> , 2014 , 204, 31-7	2.6	27
19	The genotypic false positive rate determined by V3 population sequencing can predict the burden of HIV-1 CXCR4-using species detected by pyrosequencing. <i>PLoS ONE</i> , 2013 , 8, e53603	3.7	14
18	Ultra-deep sequencing reveals hidden HIV-1 minority lineages and shifts of viral population between the main cellular reservoirs of the infection after therapy interruption. <i>Journal of Medical Virology</i> , 2012 , 84, 839-44	19.7	12
17	Detection of quasispecies variants predicted to use CXCR4 by ultra-deep pyrosequencing during early HIV infection. <i>Aids</i> , 2011 , 25, 611-7	3.5	37
16	Analysis of co-receptor usage of circulating viral and proviral HIV genome quasispecies by ultra-deep pyrosequencing in patients who are candidates for CCR5 antagonist treatment. <i>Clinical Microbiology and Infection</i> , 2011 , 17, 725-31	9.5	31
15	Sentinel mutations in standard population sequencing can predict the presence of HIV-1 reverse transcriptase major mutations detectable only by ultra-deep pyrosequencing. <i>Journal of Antimicrobial Chemotherapy</i> , 2011 , 66, 2615-23	5.1	13
14	Combinatorial analysis and algorithms for quasispecies reconstruction using next-generation sequencing. <i>BMC Bioinformatics</i> , 2011 , 12, 5	3.6	58
13	Comparison of real-time PCR methods for measurement of HIV-1 proviral DNA. <i>Journal of Virological Methods</i> , 2010 , 164, 135-8	2.6	14
12	Co-receptor usage prediction at quasispecies level using ultra-deep pyrosequencing on both circulating and proviral hiv in patients candidates to CCR5 antagonist treatment. <i>Retrovirology</i> , 2010 , 7,	3.6	78
11	Low-abundance drug resistance mutations: extending the HIV paradigm to hepatitis B virus. <i>Journal of Infectious Diseases</i> , 2009 , 200, 1798-9; author reply 1799-1800	7	8
10	Archived HIV-1 minority variants detected by ultra-deep pyrosequencing in provirus may be fully replication competent. <i>Aids</i> , 2009 , 23, 2541-3	3.5	16
9	Use of massive parallel pyrosequencing for near full-length characterization of a unique HIV Type 1 BF recombinant associated with a fatal primary infection. <i>AIDS Research and Human Retroviruses</i> , 2009 , 25, 937-42	1.6	22
8	Use of massively parallel ultradeep pyrosequencing to characterize the genetic diversity of hepatitis B virus in drug-resistant and drug-naive patients and to detect minor variants in reverse transcriptase and hepatitis B S antigen. <i>Journal of Virology</i> , 2009 , 83, 1718-26	6.6	137
7	Massively parallel pyrosequencing highlights minority variants in the HIV-1 env quasispecies deriving from lymphomonocyte sub-populations. <i>Retrovirology</i> , 2009 , 6, 15	3.6	79

6	The open state of human topoisomerase I as probed by molecular dynamics simulation. <i>Nucleic Acids Research</i> , 2007 , 35, 3032-8	20.1	20
5	Role of flexibility and long range communication on the function of human topoisomerase I. <i>Italian Journal of Biochemistry</i> , 2007 , 56, 110-4		2
4	Effect on DNA relaxation of the single Thr718Ala mutation in human topoisomerase I: a functional and molecular dynamics study. <i>Nucleic Acids Research</i> , 2005 , 33, 3339-50	20.1	43
3	Role of the linker domain and the 203-214 N-terminal residues in the human topoisomerase I DNA complex dynamics. <i>Biophysical Journal</i> , 2004 , 87, 4087-97	2.9	24
2	Single mutation in the linker domain confers protein flexibility and camptothecin resistance to human topoisomerase I. <i>Journal of Biological Chemistry</i> , 2003 , 278, 43268-75	5.4	75
1	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population		25