Alessandro Bruselles

List of Publications by Citations

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	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. *Journal of Virology*, **2009**, 83, 1718-26	6.6	137
58	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. *European Journal of Human Genetics*, **2020**, 28, 1602-1614	5.3	132
57	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. *EBioMedicine*, **2017**, 20, 39-49	8.8	112
56	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. *European Journal of Human Genetics*, **2015**, 23, 1068-71	5.3	89
55	Massively parallel pyrosequencing highlights minority variants in the HIV-1 env quasispecies deriving from lymphomonocyte sub-populations. *Retrovirology*, **2009**, 6, 15	3.6	79
54	Co-receptor usage prediction at quasispecies level using ultra-deep pyrosequencing on both circulating and proviral hiv in patients candidates to CCR5 antagonist treatment. *Retrovirology*, **2010**, 7,	3.6	78
53	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. *American Journal of Human Genetics*, **2015**, 96, 816-25	11	75
52	Single mutation in the linker domain confers protein flexibility and camptothecin resistance to human topoisomerase I. *Journal of Biological Chemistry*, **2003**, 278, 43268-75	5.4	75
51	Organoids as a new model for improving regenerative medicine and cancer personalized therapy in renal diseases. *Cell Death and Disease*, **2019**, 10, 201	9.8	61
50	Combinatorial analysis and algorithms for quasispecies reconstruction using next-generation sequencing. *BMC Bioinformatics*, **2011**, 12, 5	3.6	58
49	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. *Human Mutation*, **2015**, 36, 1080-7	4.7	51
48	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. *American Journal of Human Genetics*, **2018**, 103, 621-630	11	45
47	Effect on DNA relaxation of the single Thr718Ala mutation in human topoisomerase I: a functional and molecular dynamics study. *Nucleic Acids Research*, **2005**, 33, 3339-50	20.1	43
46	Detection of quasispecies variants predicted to use CXCR4 by ultra-deep pyrosequencing during early HIV infection. *Aids*, **2011**, 25, 611-7	3.5	37
45	Cancer Stem Cell-Based Models of Colorectal Cancer Reveal Molecular Determinants of Therapy Resistance. *Stem Cells Translational Medicine*, **2016**, 5, 511-23	6.9	35
44	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. *Clinical Microbiology and Infection*, **2011**, 17, 725-31	9.5	31
43	Specific combinations of biallelic variants cause Wiedemann-Rautenstrauch syndrome. *Journal of Medical Genetics*, **2018**, 55, 837-846	5.8	31

42	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. *American Journal of Human Genetics*, **2019**, 105, 493-508	11	30
41	The impact of next-generation sequencing on the diagnosis of pediatric-onset hereditary spastic paraplegias: new genotype-phenotype correlations for rare HSP-related genes. *Neurogenetics*, **2018**, 19, 111-121	3	28
40	An international multicenter study on HIV-1 drug resistance testing by 454 ultra-deep pyrosequencing. *Journal of Virological Methods*, **2014**, 204, 31-7	2.6	27
39	Mutations in the IRBIT domain of ITPR1 are a frequent cause of autosomal dominant nonprogressive congenital ataxia. *Clinical Genetics*, **2017**, 91, 86-91	4	26
38	ACE2gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population		25
37	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. *American Journal of Human Genetics*, **2020**, 107, 499-513	11	25
36	Role of the linker domain and the 203-214 N-terminal residues in the human topoisomerase I DNA complex dynamics. *Biophysical Journal*, **2004**, 87, 4087-97	2.9	24
35	Congenital immunodeficiency in an individual with Wiedemann-Steiner syndrome due to a novel missense mutation in KMT2A. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2389-93	2.5	24
34	A new bioavailable fenretinide formulation with antiproliferative, antimetabolic, and cytotoxic effects on solid tumors. *Cell Death and Disease*, **2019**, 10, 529	9.8	23
33	Use of massive parallel pyrosequencing for near full-length characterization of a unique HIV Type 1 BF recombinant associated with a fatal primary infection. *AIDS Research and Human Retroviruses*, **2009**, 25, 937-42	1.6	22
32	Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome. *Gene*, **2017**, 628, 141-145	3.8	20
31	The open state of human topoisomerase I as probed by molecular dynamics simulation. *Nucleic Acids Research*, **2007**, 35, 3032-8	20.1	20
30	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. *Human Molecular Genetics*, **2018**, 27, 1892-1904	5.6	18
29	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proof-of-concept examples. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1772-9	2.5	18
28	Expanding the molecular diversity and phenotypic spectrum of glycerol 3-phosphate dehydrogenase 1 deficiency. *Journal of Inherited Metabolic Disease*, **2016**, 39, 689-695	5.4	18
27	Biallelic mutations in DYNC2LI1 are a rare cause of Ellis-van Creveld syndrome. *Clinical Genetics*, **2018**, 93, 632-639	4	16
26	Archived HIV-1 minority variants detected by ultra-deep pyrosequencing in provirus may be fully replication competent. *Aids*, **2009**, 23, 2541-3	3.5	16
25	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. *Brain*, **2017**, 140, 2550-2556	11.2	15

24	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotype-phenotype correlations. *Human Mutation*, **2019**, 40, 721-728	4.7	15
23	Novel - Fusion Gene in Pediatric Ependymomas Discovered by Clonal Expansion of Stem Cells in Absence of Exogenous Mitogens. *Cancer Research*, **2017**, 77, 5860-5872	10.1	14
22	The genotypic false positive rate determined by V3 population sequencing can predict the burden of HIV-1 CXCR4-using species detected by pyrosequencing. *PLoS ONE*, **2013**, 8, e53603	3.7	14
21	Comparison of real-time PCR methods for measurement of HIV-1 proviral DNA. *Journal of Virological Methods*, **2010**, 164, 135-8	2.6	14
20	\Sentinel\mutations in standard population sequencing can predict the presence of HIV-1 reverse transcriptase major mutations detectable only by ultra-deep pyrosequencing. *Journal of Antimicrobial Chemotherapy*, **2011**, 66, 2615-23	5.1	13
19	Colorectal cancer spheroid biobanks: multi-level approaches to drug sensitivity studies. *Cell Biology and Toxicology*, **2018**, 34, 459-469	7.4	12
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. *Journal of Medical Virology*, **2012**, 84, 839-44	19.7	12
17	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. *American Journal of Human Genetics*, **2021**, 108, 502-516	11	12
16	A Follow-Up of the Multicenter Collaborative Study on HIV-1 Drug Resistance and Tropism Testing Using 454 Ultra Deep Pyrosequencing. *PLoS ONE*, **2016**, 11, e0146687	3.7	12
15	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. *American Journal of Human Genetics*, **2020**, 107, 1129-1148	11	11
14	VarGenius executes cohort-level DNA-seq variant calling and annotation and allows to manage the resulting data through a PostgreSQL database. *BMC Bioinformatics*, **2018**, 19, 477	3.6	9
13	Low-abundance drug resistance mutations: extending the HIV paradigm to hepatitis B virus. *Journal of Infectious Diseases*, **2009**, 200, 1798-9; author reply 1799-1800	7	8
12	Exome sequencing in children of women with skewed X-inactivation identifies atypical cases and complex phenotypes. *European Journal of Paediatric Neurology*, **2017**, 21, 475-484	3.8	6
11	A syndromic extreme insulin resistance caused by biallelic mutations in exon 10. *European Journal of Endocrinology*, **2017**, 177, K21-K27	6.5	6
10	HIPK2-T566 autophosphorylation diversely contributes to UV- and doxorubicin-induced HIPK2 activation. *Oncotarget*, **2017**, 8, 16744-16754	3.3	5
9	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. *European Journal of Paediatric Neurology*, **2017**, 21, 873-883	3.8	4
8	The activating p.Ser466Arg change in STAT1 causes a peculiar phenotype with features of interferonopathies. *Clinical Genetics*, **2019**, 96, 585-589	4	3
7	Refinement of the clinical and mutational spectrum of UBE2A deficiency syndrome. *Clinical Genetics*, **2020**, 98, 172-178	4	2

6	Functional analysis of variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. *Journal of Medical Genetics*, **2020**,	5.8	2
5	Role of flexibility and long range communication on the function of human topoisomerase I. *Italian Journal of Biochemistry*, **2007**, 56, 110-4		2
4	Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
3	Expanding the clinical phenotype of the ultra-rare Skraban-Deardorff syndrome: Two novel individuals with WDR26 loss-of-function variants and a literature review. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1712-1720	2.5	1
2	Broadening the phenotypic spectrum of Beta3GalT6-associated phenotypes. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3153-3160	2.5	0
1	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in. *Genes*, **2021**, 12,	4.2	0