Alessandro Bruselles

List of Publications by Year
in descending order

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Version: 2024-02-01

62	2,306	236925 25	233421 45
papers	citations	h-index	g-index

64	64	64	5684
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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.	2.8	208
2	A Specific Mutational Signature Associated with DNA 8-Oxoguanine Persistence in MUTYH-defective Colorectal Cancer. EBioMedicine, 2017, 20, 39-49.	6.1	170
3	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, 1718-1726.	3.4	143
4	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. European Journal of Human Genetics, 2015, 23, 1068-1071.	2.8	113
5	Organoids as a new model for improving regenerative medicine and cancer personalized therapy in renal diseases. Cell Death and Disease, 2019, 10, 201.	6.3	105
6	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, 816-825.	6.2	102
7	Massively parallel pyrosequencing highlights minority variants in the HIV-1 env quasispecies deriving from lymphomonocyte sub-populations. Retrovirology, 2009, 6, 15.	2.0	89
8	Single Mutation in the Linker Domain Confers Protein Flexibility and Camptothecin Resistance to Human Topoisomerase I. Journal of Biological Chemistry, 2003, 278, 43268-43275.	3.4	81
9	Mutations in KCNK4 that Affect Gating Cause a Recognizable Neurodevelopmental Syndrome. American Journal of Human Genetics, 2018, 103, 621-630.	6.2	73
10	Combinatorial analysis and algorithms for quasispecies reconstruction using next-generation sequencing. BMC Bioinformatics, 2011, 12, 5.	2.6	72
11	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	2.5	67
12	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.	1.4	52
13	Cancer Stem Cell-Based Models of Colorectal Cancer Reveal Molecular Determinants of Therapy Resistance. Stem Cells Translational Medicine, 2016, 5, 511-523.	3.3	48
14	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.	6.2	48
15	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
16	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.	6.2	48
17	Effect on DNA relaxation of the single Thr718Ala mutation in human topoisomerase I: a functional and molecular dynamics study. Nucleic Acids Research, 2005, 33, 3339-3350.	14.5	47
18	Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846.	3.2	44

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19	Detection of quasispecies variants predicted to use CXCR4 by ultra-deep pyrosequencing during early HIV infection. Aids, 2011, 25, 611-617.	2.2	40
20	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. American Journal of Human Genetics, 2020, 107, 1129-1148.	6.2	38
21	A new bioavailable fenretinide formulation with antiproliferative, antimetabolic, and cytotoxic effects on solid tumors. Cell Death and Disease, 2019, 10, 529.	6.3	37
22	Analysis of coâ€receptor usage of circulating viral and proviral HIV genome quasispecies by ultraâ€deep pyrosequencing in patients who are candidates for CCR5 antagonist treatment. Clinical Microbiology and Infection, 2011, 17, 725-731.	6.0	32
23	An international multicenter study on HIV-1 drug resistance testing by 454 ultra-deep pyrosequencing. Journal of Virological Methods, 2014, 204, 31-37.	2.1	31
24	Mutations in the <scp>IRBIT</scp> domain of <scp>ITPR1</scp> are a frequent cause of autosomal dominant nonprogressive congenital ataxia. Clinical Genetics, 2017, 91, 86-91.	2.0	30
25	Congenital immunodeficiency in an individual with Wiedemannâ€“Steiner syndrome due to a novel missense mutation in KMT2A. American Journal of Medical Genetics, Part A, 2016, 170, 2389-2393.	1.2	29
26	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Human Molecular Genetics, 2018, 27, 1892-1904.	2.9	29
27	Identification of novel and hotspot mutations in the channel domain of ITPR1 in two patients with Gillespie syndrome. Gene, 2017, 628, 141-145.	2.2	27
28	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proofâ€ofâ€concept examples. American Journal of Medical Genetics, Part A, 2016, 170, 1772-1779.	1.2	26
29	NBAS pathogenic variants: Defining the associated clinical and facial phenotype and genotypeâ€“phenotype correlations. Human Mutation, 2019, 40, 721-728.	2.5	26
30	Role of the Linker Domain and the 203â€“214 N-Terminal Residues in the Human Topoisomerase I DNA Complex Dynamics. Biophysical Journal, 2004, 87, 4087-4097.	0.5	25
31	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, 937-942.	1.1	24
32	Expanding the molecular diversity and phenotypic spectrum of glycerol 3â€phosphate dehydrogenase 1 deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 689-695.	3.6	24
33	Biallelic mutations in <scp>DYNC2LI1</scp> are a rare cause of Ellisâ€van Creveld syndrome. Clinical Genetics, 2018, 93, 632-639.	2.0	23
34	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.	2.6	23
35	The open state of human topoisomerase I as probed by molecular dynamics simulation. Nucleic Acids Research, 2007, 35, 3032-3038.	14.5	21
36	Novel SEC61Gâ€“EGFR Fusion Gene in Pediatric Ependymomas Discovered by Clonal Expansion of Stem Cells in Absence of Exogenous Mitogens. Cancer Research, 2017, 77, 5860-5872.	0.9	21

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37	Biallelic mutations in the homeodomain of NKX6-2 underlie a severe hypomyelinating leukodystrophy. Brain, 2017, 140, 2550-2556.	7.6	18
38	Archived HIV-1 minority variants detected by ultra-deep pyrosequencing in provirus may be fully replication competent. Aids, 2009, 23, 2541-2543.	2.2	17
39	Comparison of real-time PCR methods for measurement of HIV-1 proviral DNA. Journal of Virological Methods, 2010, 164, 135-138.	2.1	17
40	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.	2.5	15
41	'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, 2615-2623.	3.0	14
42	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.	2.5	14
43	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.	1.6	14
44	Colorectal cancer spheroid biobanks: multi-level approaches to drug sensitivity studies. Cell Biology and Toxicology, 2018, 34, 459-469.	5.3	14
45	Ultraâ€deep sequencing reveals hidden HIVâ€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, 839-844.	5.0	12
46	The activating p.Ser466Arg change in STAT1 causes a peculiar phenotype with features of interferonopathies. Clinical Genetics, 2019, 96, 585-589.	2.0	10
47	An Orthotopic Patient-Derived Xenograft (PDX) Model Allows the Analysis of Metastasis-Associated Features in Colorectal Cancer. Frontiers in Oncology, 0, 12, .	2.8	10
48	Functional analysis of TLK2 variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. Journal of Medical Genetics, 2022, 59, 170-179.	3.2	9
49	Lowâ€Abundance Drug Resistance Mutations: Extending the HIV Paradigm to Hepatitis B Virus. Journal of Infectious Diseases, 2009, 200, 1798-1799.	4.0	8
50	A syndromic extreme insulin resistance caused by biallelic POC1A mutations in exon 10. European Journal of Endocrinology, 2017, 177, K21-K27.	3.7	8
51	Expanding the clinical phenotype of the ultraâ€rare <scp>Skrabanâ€Deardorff</scp> syndrome: Two novel individuals with <scp>WDR26</scp> lossâ€ofâ€function variants and a literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1712-1720.	1.2	6
52	HIPK2-T566 autophosphorylation diversely contributes to UV- and doxorubicin-induced HIPK2 activation. Oncotarget, 2017, 8, 16744-16754.	1.8	6
53	Refinement of the clinical and mutational spectrum of <scp>UBE2A</scp> deficiency syndrome. Clinical Genetics, 2020, 98, 172-178.	2.0	5
54	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. European Journal of Paediatric Neurology, 2017, 21, 873-883.	1.6	4

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55	Broadening the phenotypic spectrum of Beta3GalT6 â€œassociated phenotypes. American Journal of Medical Genetics, Part A, 2021, 185, 3153-3160.	1.2	3
56	KCNK18 Biallelic Variants Associated with Intellectual Disability and Neurodevelopmental Disorders Alter TRESK Channel Activity. International Journal of Molecular Sciences, 2021, 22, 6064.	4.1	3
57	A Rare Case of Brachyolmia with Amelogenesis Imperfecta Caused by a New Pathogenic Splicing Variant in LTBP3. Genes, 2021, 12, 1406.	2.4	2
58	Role of flexibility and long range communication on the function of human topoisomerase I. Italian Journal of Biochemistry, 2007, 56, 110-4.	0.3	2
59	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, .	2.0	1
60	MiRLog and dbmiR: Prioritization and functional annotation tools to study human microRNA sequence variants. Human Mutation, 2022, , .	2.5	1
61	Cover Image, Volume 170A, Number 7, July 2016. , 2016, 170, i-i.		0
62	Front Cover, Volume 40, Issue 6. Human Mutation, 2019, 40, i.	2.5	0