Alessandro Stella

List of Publications by Year
in descending order

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65 papers	1,574 citations	304602 22 h-index	330025 37 g-index
68 all docs	68 docs citations	68 times ranked	2715 citing authors

#	Article	IF	Citations
1	APC Is Essential for Targeting Phosphorylated Î²-Catenin to the SCFÎ²-TrCP Ubiquitin Ligase. Molecular Cell, 2008, 32, 652-661.	4.5	149
2	Laminin Î±2 muscular dystrophy. Neurology, 1998, 51, 101-110.	1.5	118
3	Cancer risk associated with STK11/LKB1 germline mutations in Peutzâ€“Jeghers syndrome patients: Results of an Italian multicenter study. Digestive and Liver Disease, 2013, 45, 606-611.	0.4	113
4	A long diagnostic delay in patients with Hereditary Haemorrhagic Telangiectasia: a questionnaire-based retrospective study. Orphanet Journal of Rare Diseases, 2012, 7, 33.	1.2	78
5	Molecular and Functional Characterization of Three Different Postzygotic Mutations in PIK3CA-Related Overgrowth Spectrum (PROS) Patients: Effects on PI3K/AKT/mTOR Signaling and Sensitivity to PIK3 Inhibitors. PLoS ONE, 2015, 10, e0123092.	1.1	72
6	An LKB1 AT-AC intron mutation causes Peutz-Jeghers syndrome via splicing at noncanonical cryptic splice sites. Nature Structural and Molecular Biology, 2005, 12, 54-59.	3.6	62
7	A silent mutation in exon 14 of the APC gene is associated with exon skipping in a FAP family. Journal of Medical Genetics, 2001, 38, 863-867.	1.5	54
8	In silico and in vivo splicing analysis of MLH1 and MSH2 missense mutations shows exon- and tissue-specific effects. BMC Genomics, 2006, 7, 243.	1.2	54
9	A homozygous frameshift mutation in theESCO2 gene: Evidence of intertissue and interindividual variation in Nmd efficiency. Journal of Cellular Physiology, 2006, 209, 67-73.	2.0	48
10	Germline novel MSH2 deletions and a founder MSH2 deletion associated with anticipation effects in HNPCC. Clinical Genetics, 2007, 71, 130-139.	1.0	44
11	Metabolomic Approaches for Detection and Identification of Biomarkers and Altered Pathways in Bladder Cancer. International Journal of Molecular Sciences, 2022, 23, 4173.	1.8	40
12	Two novel mutations and a new STK11/LKB1 gene isoform in Peutz-Jeghers patients. Human Mutation, 2002, 20, 78-79.	1.1	37
13	Functional analysis of LKB1/STK11 mutants and two aberrant isoforms found in Peutz-Jeghers Syndrome patients. Human Mutation, 2003, 21, 172-172.	1.1	35
14	Novel Insights into Autophagy and Prostate Cancer: A Comprehensive Review. International Journal of Molecular Sciences, 2022, 23, 3826.	1.8	31
15	Colorectal Cancer and The Muir-Torre Syndrome in A Gypsy Family: A Review. American Journal of Gastroenterology, 1999, 94, 575-580.	0.2	30
16	Distinguishing between recent balancing selection and incomplete sweep using deep neural networks. Molecular Ecology Resources, 2021, 21, 2706-2718.	2.2	30
17	Blood-based test for diagnosis and functional subtyping of familial Mediterranean fever. Annals of the Rheumatic Diseases, 2020, 79, 960-968.	0.5	29
18	Infertility in carriers of two bisatellited marker chromosomes. Clinical Genetics, 1993, 44, 71-75.	1.0	28

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19	Loss of STK11 expression is an early event in prostate carcinogenesis and predicts therapeutic response to targeted therapy against MAPK/p38. Autophagy, 2015, 11, 2102-2113.	4.3	27
20	Breakpoint determination of 15 large deletions in Peutzâ€“Jeghers subjects. Human Genetics, 2010, 128, 373-382.	1.8	26
21	Improvement of MEFV gene variants classification to aid treatment decision making in familial Mediterranean fever. Rheumatology, 2020, 59, 754-761.	0.9	26
22	Four novel mutations of the APC (adenomatous polyposis coli) gene in FAP patients. Human Molecular Genetics, 1994, 3, 1687-1688.	1.4	25
23	Familial adenomatous polyposis: Identification of a new frameshift mutation of the APC gene in an Italian family. Biochemical and Biophysical Research Communications, 1992, 184, 1357-1363.	1.0	24
24	Survey of KRAS, BRAF and PIK3CA mutational status in 209 consecutive Italian colorectal cancer patients. International Journal of Biological Markers, 2012, 27, 366-374.	0.7	22
25	Accurate Classification of NF1 Gene Variants in 84 Italian Patients with Neurofibromatosis Type 1. Genes, 2018, 9, 216.	1.0	22
26	Papillary Thyroid Carcinoma in Peutz-Jeghers Syndrome. Thyroid, 2011, 21, 1273-1277.	2.4	21
27	Familial Mediterranean fever: breaking all the (genetic) rules. Rheumatology, 2019, 58, 463-467.	0.9	21
28	Anticipation in Lynch Syndrome: Where We Are Where We Go. Current Genomics, 2011, 12, 451-465.	0.7	21
29	Nine novel APC mutations in Italian FAP patients. Human Mutation, 2001, 17, 434-435.	1.1	20
30	Familial Mediterranean Fever and COVID-19: Friends or Foes?. Frontiers in Immunology, 2020, 11, 574593.	2.2	20
31	Mutations in theNDUFS4gene of mitochondrial complex I alter stability of the splice variants. FEBS Letters, 2005, 579, 3770-3776.	1.3	19
32	Site directed mutagenesis of hMLH1 exonic splicing enhancers does not correlate with splicing disruption. Journal of Medical Genetics, 2004, 41, e72-e72.	1.5	17
33	Novel splice isoforms of STRADÎ± differentially affect LKB1 activity, complex assembly and subcellular localization.. Cancer Biology and Therapy, 2007, 6, 1627-1631.	1.5	16
34	Gut Microbiota between Environment and Genetic Background in Familial Mediterranean Fever (FMF). Genes, 2020, 11, 1041.	1.0	16
35	Large expert-curated database for benchmarking document similarity detection in biomedical literature search. Database: the Journal of Biological Databases and Curation, 2019, 2019, .	1.4	15
36	Comparison of clinical and demographic features between affected pairs of Italian Multiple Sclerosis multiplex families; relation to tumour necrosis factor genomic polymorphisms. Journal of the Neurological Sciences, 1999, 162, 194-200.	0.3	13

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37	Analysis of telomere dynamics in peripheral blood cells from patients with Lynch syndrome. Cancer, 2011, 117, 4325-4335.	2.0	12
38	Gastric polyposis and desmoid tumours as a new familial adenomatous polyposis clinical variant associated with APC mutation at the extreme 3â€²-end. Journal of Medical Genetics, 2020, 57, 356-360.	1.5	12
39	Identification and surveillance of 19 Lynch syndrome families in southern Italy: report of six novel germline mutations and a common founder mutation. Familial Cancer, 2011, 10, 285-295.	0.9	11
40	The mechanism of alternative splicing of the X-linked NDUFB11 gene of the respiratory chain complex I, impact of rotenone treatment in neuroblastoma cells. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2013, 1829, 211-218.	0.9	9
41	In-silico Analysis of NF1 Missense Variants in ClinVar: Translating Variant Predictions into Variant Interpretation and Classification. International Journal of Molecular Sciences, 2020, 21, 721.	1.8	9
42	Population data for 17 Y-chromosome STRs in a sample from Apulia (Southern Italy). Forensic Science International: Genetics, 2013, 7, e3-e4.	1.6	8
43	The grandfatherâ€™s fever. Clinical Rheumatology, 2020, 39, 585-594.	1.0	7
44	Four novel mutations of the APC (adenomatous polyposis coli) gene in FAP patients. Human Molecular Genetics, 1994, 3, 1918-1918.	1.4	6
45	A rare MSH2 mutation causes defective binding to hMSH6, normal hMSH2 staining, and loss of hMSH6 at advanced cancer stage. Human Pathology, 2014, 45, 2162-2167.	1.1	6
46	The recurrent SETBP1 c.2608Gâ€‰>â€‰A, p.(Gly870Ser) variant in a patient with Schinzel-Giedion syndrome: an illustrative case of the utility of whole exome sequencing in a critically ill neonate. Italian Journal of Pediatrics, 2020, 46, 74.	1.0	6
47	Cancer family syndrome: cytogenetic investigations, in vitro tetraploidy, and biomarker studies in a large family.. Journal of Medical Genetics, 1990, 27, 441-445.	1.5	5
48	Beyond BRCA1/2: Homologous Recombination Repair Genetic Profile in a Large Cohort of Apulian Ovarian Cancers. Cancers, 2022, 14, 365.	1.7	5
49	Depressed level of natural killer cells in cancer family syndrome. Cancer Immunology, Immunotherapy, 1989, 30, 307-311.	2.0	4
50	Linkage studies in Italian families with familial adenomatous polyposis. Human Genetics, 1993, 90, 545-550.	1.8	4
51	Clinical Findings in a Family with Familial Adenomatous Polyposis and a Missense Mutation of the Adenomatous Polyposis Coli Gene. Scandinavian Journal of Gastroenterology, 1996, 31, 917-920.	0.6	4
52	Country-level factors dynamics and ABO/Rh blood groups contribution to COVID-19 mortality. Scientific Reports, 2021, 11, 24527.	1.6	4
53	Lateralized overgrowth with vascular malformation caused by a somatic PTPN11 pathogenic variant: Another piece added to the puzzle of mosaic <scp>RASopathies</scp>. Genes Chromosomes and Cancer, 2022, 61, 689-695.	1.5	4
54	The familial adenomatous polyposis region exhibits many different haplotypes. Human Genetics, 1998, 102, 624-628.	1.8	3

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55	401 Loss of LKB1/STK11 expression is an early event in prostate cancer development and predicts therapeutic response to p38Î± inhibitor. European Urology Supplements, 2015, 14, e401-e401a.	0.1	3
56	Establishment and characterization of a highly immunogenic human renal carcinoma cell line. International Journal of Oncology, 2016, 49, 457-470.	1.4	3
57	Neurofibromatosis type 1 and melanoma of the iris arising from a dysplastic nevus: A rare yet casual association?. European Journal of Ophthalmology, 2020, 31, 112067212090699.	0.7	3
58	Oro-Dental Manifestations in a Pediatric Patient Affected by Helsmoortel-Van der Aa Syndrome. International Journal of Environmental Research and Public Health, 2021, 18, 8957.	1.2	3
59	Clinical presentation and genetic analyses of neurofibromatosis type 1 in independent patients with monoallelic double de novo closely spaced mutations in the NF1 gene. Human Mutation, 2022, 43, 1354-1360.	1.1	3
60	The hUPF1-NMD factor controls the cellular transcript levels of different genes of complex I of the respiratory chain. Biochimie, 2012, 94, 2600-2607.	1.3	2
61	Comment on: Improvement of MEFV gene variants classification to aid treatment decision making in familial Mediterranean fever: reply. Rheumatology, 2020, 59, 911-912.	0.9	1
62	The unsolved mystery of MEFV variants variable expressivity in Familial Mediterranean Fever. Internal and Emergency Medicine, 2022, 17, 1255-1259.	1.0	1
63	Relief of transcriptional polarity by a mutation that creates a promoter in the hisG gene of Salmonella typhimurium LT2. Molecular Genetics and Genomics, 1998, 257, 529-533.	2.4	0
64	Histopathological characteristics of synovitis in Familial Mediterranean Fever (FMF). Joint Bone Spine, 2022, 89, 105259.	0.8	0
65	Relief of transcriptional polarity by a mutation that creates a promoter in the. Molecular Genetics and Genomics, 1998, 257, 529.	2.4	0