Alessandro Stella

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6719209/publications.pdf

Version: 2024-02-01

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	Histopathological characteristics of synovitis in Familial Mediterranean Fever (FMF). Joint Bone Spine, 2022, 89, 105259.	0.8	0
2	Beyond BRCA1/2: Homologous Recombination Repair Genetic Profile in a Large Cohort of Apulian Ovarian Cancers. Cancers, 2022, 14, 365.	1.7	5
3	Novel Insights into Autophagy and Prostate Cancer: A Comprehensive Review. International Journal of Molecular Sciences, 2022, 23, 3826.	1.8	31
4	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
5	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
6	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
7	The unsolved mystery of MEFV variants variable expressivity in Familial Mediterranean Fever. Internal and Emergency Medicine, 2022, 17, 1255-1259.	1.0	1
8	Distinguishing between recent balancing selection and incomplete sweep using deep neural networks. Molecular Ecology Resources, 2021, 21, 2706-2718.	2.2	30
9	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
10	Country-level factors dynamics and ABO/Rh blood groups contribution to COVID-19 mortality. Scientific Reports, 2021, 11, 24527.	1.6	4
11	The grandfatherâ€™s fever. Clinical Rheumatology, 2020, 39, 585-594.	1.0	7
12	Improvement of MEFV gene variants classification to aid treatment decision making in familial Mediterranean fever. Rheumatology, 2020, 59, 754-761.	0.9	26
13	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
14	Familial Mediterranean Fever and COVID-19: Friends or Foes?. Frontiers in Immunology, 2020, 11, 574593.	2.2	20
15	Gut Microbiota between Environment and Genetic Background in Familial Mediterranean Fever (FMF). Genes, 2020, 11, 1041.	1.0	16
16	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
17	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
18	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

#	Article	IF	Citations
19	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
20	Blood-based test for diagnosis and functional subtyping of familial Mediterranean fever. Annals of the Rheumatic Diseases, 2020, 79, 960-968.	0.5	29
21	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
22	Familial Mediterranean fever: breaking all the (genetic) rules. Rheumatology, 2019, 58, 463-467.	0.9	21
23	Accurate Classification of NF1 Gene Variants in 84 Italian Patients with Neurofibromatosis Type 1. Genes, 2018, 9, 216.	1.0	22
24	Establishment and characterization of a highly immunogenic human renal carcinoma cell line. International Journal of Oncology, 2016, 49, 457-470.	1.4	3
25	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
26	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
27	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
28	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
29	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
30	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
31	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
32	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
33	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
34	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
35	Papillary Thyroid Carcinoma in Peutz-Jeghers Syndrome. Thyroid, 2011, 21, 1273-1277.	2.4	21
36	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

#	Article	IF	Citations
37	Analysis of telomere dynamics in peripheral blood cells from patients with Lynch syndrome. Cancer, 2011, 117, 4325-4335.	2.0	12
38	Anticipation in Lynch Syndrome: Where We Are Where We Go. Current Genomics, 2011, 12, 451-465.	0.7	21
39	Breakpoint determination of 15 large deletions in Peutzâ€“Jeghers subjects. Human Genetics, 2010, 128, 373-382.	1.8	26
40	APC Is Essential for Targeting Phosphorylated Î²-Catenin to the SCFÎ²-TrCP Ubiquitin Ligase. Molecular Cell, 2008, 32, 652-661.	4.5	149
41	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
42	Germline novel MSH2 deletions and a founder MSH2 deletion associated with anticipation effects in HNPCC. Clinical Genetics, 2007, 71, 130-139.	1.0	44
43	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
44	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
45	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
46	Mutations in theNDUFS4gene of mitochondrial complex I alter stability of the splice variants. FEBS Letters, 2005, 579, 3770-3776.	1.3	19
47	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
48	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
49	Two novel mutations and a new STK11/LKB1 gene isoform in Peutz-Jeghers patients. Human Mutation, 2002, 20, 78-79.	1.1	37
50	Nine novel APC mutations in Italian FAP patients. Human Mutation, 2001, 17, 434-435.	1.1	20
51	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
52	Colorectal Cancer and The Muir-Torre Syndrome in A Gypsy Family: A Review. American Journal of Gastroenterology, 1999, 94, 575-580.	0.2	30
53	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
54	The familial adenomatous polyposis region exhibits many different haplotypes. Human Genetics, 1998, 102, 624-628.	1.8	3

#	Article	IF	Citations
55	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
56	Laminin Î±2 muscular dystrophy. Neurology, 1998, 51, 101-110.	1.5	118
57	Relief of transcriptional polarity by a mutation that creates a promoter in the. Molecular Genetics and Genomics, 1998, 257, 529.	2.4	0
58	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
59	Four novel mutations of the APC (adenomatous polyposis coli) gene in FAP patients. Human Molecular Genetics, 1994, 3, 1918-1918.	1.4	6
60	Four novel mutations of the APC (adenomatous polyposis coli) gene in FAP patients. Human Molecular Genetics, 1994, 3, 1687-1688.	1.4	25
61	Linkage studies in Italian families with familial adenomatous polyposis. Human Genetics, 1993, 90, 545-550.	1.8	4
62	Infertility in carriers of two bisatellited marker chromosomes. Clinical Genetics, 1993, 44, 71-75.	1.0	28
63	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
64	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
65	Depressed level of natural killer cells in cancer family syndrome. Cancer Immunology, Immunotherapy, 1989, 30, 307-311.	2.0	4