Simone Martinelli

List of Publications by Year
in descending order

52 papers	4,688 citations	201385 27 h-index	189595 50 g-index
54 all docs	54 docs citations	54 times ranked	7344 citing authors

#	Article	IF	Citations
1	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	9.4	624
2	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	9.4	523
3	Mutation of SHOC2 promotes aberrant protein N-myristoylation and causes Noonan-like syndrome with loose anagen hair. Nature Genetics, 2009, 41, 1022-1026.	9.4	358
4	Diversity and Functional Consequences of Germline and Somatic PTPN11 Mutations in Human Disease. American Journal of Human Genetics, 2006, 78, 279-290.	2.6	352
5	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
6	Somatically acquired JAK1 mutations in adult acute lymphoblastic leukemia. Journal of Experimental Medicine, 2008, 205, 751-758.	4.2	318
7	Genetic evidence for lineage-related and differentiation stage-related contribution of somatic PTPN11 mutations to leukemogenesis in childhood acute leukemia. Blood, 2004, 104, 307-313.	0.6	265
8	Heterozygous Germline Mutations in the CBL Tumor-Suppressor Gene Cause a Noonan Syndrome-like Phenotype. American Journal of Human Genetics, 2010, 87, 250-257.	2.6	221
9	Therapeutic targeting of Chk1 in NSCLC stem cells during chemotherapy. Cell Death and Differentiation, 2012, 19, 768-778.	5.0	157
10	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
11	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	4.2	132
12	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. Human Molecular Genetics, 2014, 23, 4315-4327.	1.4	114
13	RAS signaling dysregulation in human embryonal Rhabdomyosarcoma. Genes Chromosomes and Cancer, 2009, 48, 975-982.	1.5	88
14	Diverse driving forces underlie the invariant occurrence of the T42A, E139D, I282V and T468M SHP2 amino acid substitutions causing Noonan and LEOPARD syndromes. Human Molecular Genetics, 2008, 17, 2018-2029.	1.4	79
15	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. British Journal of Haematology, 2005, 129, 333-339.	1.2	78
16	Spectrum of MEK1 and MEK2 gene mutations in cardio-facio-cutaneous syndrome and genotypeâ€“phenotype correlations. European Journal of Human Genetics, 2009, 17, 733-740.	1.4	74
17	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	1.1	67
18	Acquired PTPN11 mutations occur rarely in adult patients with myelodysplastic syndromes and chronic myelomonocytic leukemia. Leukemia Research, 2005, 29, 459-462.	0.4	64

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19	Sensitivity to DNA cross-linking chemotherapeutic agents in mismatch repair-defective cellsin vitro and in xenografts. , 2000, 85, 590-596.		48
20	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. Cancer Genetics and Cytogenetics, 2006, 166, 124-129.	1.0	48
21	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.	2.6	48
22	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
23	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235.	1.0	44
24	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	1.8	40
25	Structural, Functional, and Clinical Characterization of a NovelPTPN11Mutation Cluster Underlying Noonan Syndrome. Human Mutation, 2017, 38, 451-459.	1.1	39
26	Molecular Diversity and Associated Phenotypic Spectrum of GermlineCBLMutations. Human Mutation, 2015, 36, 787-796.	1.1	36
27	Counteracting Effects Operating on Src Homology 2 Domain-containing Protein-tyrosine Phosphatase 2 (SHP2) Function Drive Selection of the Recurrent Y62D and Y63C Substitutions in Noonan Syndrome*. Journal of Biological Chemistry, 2012, 287, 27066-27077.	1.6	35
28	Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology, 2018, 91, e319-e330.	1.5	35
29	Caenorhabditis elegans provides an efficient drug screening platform for GNAO1-related disorders and highlights the potential role of caffeine in controlling dyskinesia. Human Molecular Genetics, 2022, 31, 929-941.	1.4	32
30	Structural and functional effects of disease-causing amino acid substitutions affecting residues Ala72 and Glu76 of the protein tyrosine phosphatase SHP-2. Proteins: Structure, Function and Bioinformatics, 2006, 66, 963-974.	1.5	31
31	Differences in the prevalence of PTPN11 mutations in FAB M5 paediatric acute myeloid leukaemia. British Journal of Haematology, 2005, 130, 801-803.	1.2	23
32	C. elegans-based chemosensation strategy for the early detection of cancer metabolites in urine samples. Scientific Reports, 2021, 11, 17133.	1.6	22
33	Combined mismatch and nucleotide excision repair defects in a human cell line: mismatch repair processes methylation but not UV- or ionizing radiation-induced DNA damage. Carcinogenesis, 1999, 20, 799-804.	1.3	20
34	Targeting Oncogenic Src Homology 2 Domain-Containing Phosphatase 2 (SHP2) by Inhibiting Its Proteinâ€“Protein Interactions. Journal of Medicinal Chemistry, 2021, 64, 15973-15990.	2.9	17
35	Mutations at the C-terminus of CDC42 cause distinct hematopoietic and autoinflammatory disorders. Journal of Allergy and Clinical Immunology, 2022, 150, 223-228.	1.5	17
36	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. Parkinsonism and Related Disorders, 2020, 72, 75-79.	1.1	16

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37	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	3.7	15
38	De novo pure 12q22q24.33 duplication: First report of a case with mental retardation, ADHD, and Dandy-Walker malformation. American Journal of Medical Genetics, Part A, 2006, 140A, 1203-1207.	0.7	12
39	A PTPN11 allele encoding a catalytically impaired SHP2 protein in a patient with a Noonan syndrome phenotype. American Journal of Medical Genetics, Part A, 2014, 164, 2351-2355.	0.7	12
40	Biallelic mutations in the TOGARAM1 gene cause a novel primary ciliopathy. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-106833.	1.5	12
41	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.	1.5	9
42	Reversal of methylation tolerance by transfer of human chromosome 2. Mutation Research DNA Repair, 1997, 385, 115-126.	3.8	7
43	Loss of <scp>CBL</scp> E3â€ligase activity in Bâ€lineage childhood acute lymphoblastic leukaemia. British Journal of Haematology, 2012, 159, 115-119.	1.2	6
44	Clinical variability of neurofibromatosis 1: A modifying role of cooccurring <scp>PTPN11</scp> variants and atypical brain <scp>MRI</scp> findings. Clinical Genetics, 2021, 100, 563-572.	1.0	6
45	Gain of Function of Malate Dehydrogenase 2 and Familial Hyperglycemia. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 668-684.	1.8	4
46	â€œAtypicalâ€•Krabbe disease in two siblings harboring biallelic GALC mutations including a deep intronic variant. European Journal of Human Genetics, 2022, , .	1.4	4
47	Pathogenic PTPN11 variants involving the polyâ€glutamine Gln ²⁵⁵ â€Gln ²⁵⁶ â€Gln ²⁵⁷ stretch highlight the relevance of helix B in SHP2's functional regulation. Human Mutation, 2020, 41, 1171-1182.	1.1	3
48	Efficient one-step chromatographic purification and functional characterization of recombinant human Saposin C. Protein Expression and Purification, 2011, 78, 209-215.	0.6	2
49	Compound heterozygosity for <scp>PTPN11</scp> variants in a subject with Noonan syndrome provides insights into the mechanism of <scp>SHP2</scp>â€related disorders. Clinical Genetics, 2021, 99, 457-461.	1.0	2
50	Malate Dehydrogenase 2 (MDH2) as a New Diabetogene Causing Hyperglycemia in Families with Multigenerational Diabetes. Diabetes, 2018, 67, 262-OR.	0.3	2
51	<scp>SHP2</scp>'s gainâ€ofâ€function in <scp>Werner</scp> syndrome causes childhood disease onset likely resulting from negative genetic interaction. Clinical Genetics, 2022, 102, 12-21.	1.0	2
52	Germline PTPN11 mutation affecting exon 8 in a case of syndromic juvenile myelomonocytic leukemia. Leukemia Research, 2011, 35, e13-e14.	0.4	0