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	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
2	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	3.7	15
3	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
4	Gain of Function of Malate Dehydrogenase 2 and Familial Hyperglycemia. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 668-684.	1.8	4
5	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
6	<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
7	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
8	C. elegans-based chemosensation strategy for the early detection of cancer metabolites in urine samples. Scientific Reports, 2021, 11, 17133.	1.6	22
9	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
10	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
11	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
12	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	1.8	40
13	Biallelic mutations in the TOGARAM1 gene cause a novel primary ciliopathy. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-106833.	1.5	12
14	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	2.6	48
15	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
16	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
17	A novel disorder involving dyshematopoiesis, inflammation, and HLH due to aberrant CDC42 function. Journal of Experimental Medicine, 2019, 216, 2778-2799.	4.2	132
18	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

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19	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
20	Clinical Presentation and Natural History of Hypertrophic Cardiomyopathy in RASopathies. Heart Failure Clinics, 2018, 14, 225-235.	1.0	44
21	Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology, 2018, 91, e319-e330.	1.5	35
22	Malate Dehydrogenase 2 (MDH2) as a New Diabetogene Causing Hyperglycemia in Families with Multigenerational Diabetes. Diabetes, 2018, 67, 262-OR.	0.3	2
23	Structural, Functional, and Clinical Characterization of a NovelPTPN11Mutation Cluster Underlying Noonan Syndrome. Human Mutation, 2017, 38, 451-459.	1.1	39
24	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
25	Activating Mutations Affecting the Dbl Homology Domain of SOS2 Cause Noonan Syndrome. Human Mutation, 2015, 36, 1080-1087.	1.1	67
26	Molecular Diversity and Associated Phenotypic Spectrum of GermlineCBLMutations. Human Mutation, 2015, 36, 787-796.	1.1	36
27	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
28	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
29	Therapeutic targeting of Chk1 in NSCLC stem cells during chemotherapy. Cell Death and Differentiation, 2012, 19, 768-778.	5.0	157
30	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
31	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
32	Efficient one-step chromatographic purification and functional characterization of recombinant human Saposin C. Protein Expression and Purification, 2011, 78, 209-215.	0.6	2
33	Germline PTPN11 mutation affecting exon 8 in a case of syndromic juvenile myelomonocytic leukemia. Leukemia Research, 2011, 35, e13-e14.	0.4	0
34	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
35	RAS signaling dysregulation in human embryonal Rhabdomyosarcoma. Genes Chromosomes and Cancer, 2009, 48, 975-982.	1.5	88
36	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

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37	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
38	Somatically acquired JAK1 mutations in adult acute lymphoblastic leukemia. Journal of Experimental Medicine, 2008, 205, 751-758.	4.2	318
39	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
40	Gain-of-function SOS1 mutations cause a distinctive form of Noonan syndrome. Nature Genetics, 2007, 39, 75-79.	9.4	523
41	Gain-of-function RAF1 mutations cause Noonan and LEOPARD syndromes with hypertrophic cardiomyopathy. Nature Genetics, 2007, 39, 1007-1012.	9.4	624
42	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
43	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
44	Activating PTPN11 mutations play a minor role in pediatric and adult solid tumors. Cancer Genetics and Cytogenetics, 2006, 166, 124-129.	1.0	48
45	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
46	Acquired PTPN11 mutations occur rarely in adult patients with myelodysplastic syndromes and chronic myelomonocytic leukemia. Leukemia Research, 2005, 29, 459-462.	0.4	64
47	Somatic PTPN11 mutations in childhood acute myeloid leukaemia. British Journal of Haematology, 2005, 129, 333-339.	1.2	78
48	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
49	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
50	Sensitivity to DNA cross-linking chemotherapeutic agents in mismatch repair-defective cellsin vitro and in xenografts. , 2000, 85, 590-596.		48
51	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
52	Reversal of methylation tolerance by transfer of human chromosome 2. Mutation Research DNA Repair, 1997, 385, 115-126.	3.8	7