Tetsuya Niihori

List of Publications by Year
in descending order

78	5,314	147726	88593
papers	citations	31 h-index	70 g-index

79	79	79	7995
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Duplications in the G3 domain or switch II region in HRAS identified in patients with Costello syndrome. Human Mutation, 2022, 43, 3-15.	1.1	7
2	NovelPOLEmutations identified in patients with IMAGE-I syndrome cause aberrant subcellular localisation and protein degradation in the nucleus. Journal of Medical Genetics, 2022, 59, 1116-1122.	1.5	2
3	A novel deletion in the C-terminal region of HSPB8 in a family with rimmed vacuolar myopathy. Journal of Human Genetics, 2021, 66, 965-972.	1.1	6
4	A de novo CHD3 variant in a child with intellectual disability, autism, joint laxity, and dysmorphisms. Brain and Development, 2021, 43, 563-565.	0.6	7
5	Detection of intracellular histological abnormalities using cardiac magnetic resonance T1 mapping in patients with Danon disease: a case series. European Heart Journal - Case Reports, 2021, 5, ytab145.	0.3	3
6	LZTR1 facilitates polyubiquitination and degradation of RAS-GTPases. Cell Death and Differentiation, 2020, 27, 1023-1035.	5.0	70
7	Variants That Affect Function of Calcium Channel TRPV6 Are Associated With Early-Onset Chronic Pancreatitis. Gastroenterology, 2020, 158, 1626-1641.e8.	0.6	77
8	Utility of a bridged nucleic acid clamp for liquid biopsy: Detecting BRAF V600E in the cerebrospinal fluid of a patient with brain tumor. Pediatric Blood and Cancer, 2020, 67, e28651.	0.8	3
9	Costello syndrome model mice with a Hras G12S mutation are susceptible to develop house dust mite-induced atopic dermatitis. Cell Death and Disease, 2020, 11, 617.	2.7	2
10	The genetic profile of dysferlinopathy in a cohort of 209 cases: Genotypeâ€“phenotype relationship and a hotspot on the inner DysF domain. Human Mutation, 2020, 41, 1540-1554.	1.1	27
11	A somatic activating KRAS variant identified in an affected lesion of a patient with Gorhamâ€“Stout disease. Journal of Human Genetics, 2020, 65, 995-1001.	1.1	30
12	Metabolic and pathologic profiles of human LSS deficiency recapitulated in mice. PLoS Genetics, 2020, 16, e1008628.	1.5	21
13	Biallelic variants/mutations of IL1RAP in patients with steroid-sensitive nephrotic syndrome. International Immunology, 2020, 32, 283-292.	1.8	3
14	Genotype-phenotype correlation analysis in Japanese patients with Noonan syndrome. Endocrine Journal, 2019, 66, 983-994.	0.7	12
15	Detection of NRAS mutation in cell-free DNA biological fluids from patients with kaposiform lymphangiomatosis. Orphanet Journal of Rare Diseases, 2019, 14, 215.	1.2	43
16	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	2.6	35
17	New Noonan syndrome model mice with RIT1 mutation exhibit cardiac hypertrophy and susceptibility to Î²-adrenergic stimulation-induced cardiac fibrosis. EBioMedicine, 2019, 42, 43-53.	2.7	23
18	Leucine-485 deletion variant of BRAF may exhibit the severe end of the clinical spectrum of CFC syndrome. Journal of Human Genetics, 2019, 64, 499-504.	1.1	3

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19	Recurrent de novo MAPK8IP3 variants cause neurological phenotypes. Annals of Neurology, 2019, 85, 927-933.	2.8	34
20	Biallelic GALM pathogenic variants cause a novel type of galactosemia. Genetics in Medicine, 2019, 21, 1286-1294.	1.1	40
21	Delineation of LZTR1 mutation-positive patients with Noonan syndrome and identification of LZTR1 binding to RAF1â€“PPP1CB complexes. Human Genetics, 2019, 138, 21-35.	1.8	50
22	Genomic analysis identifies masqueraders of fullâ€term cerebral palsy. Annals of Clinical and Translational Neurology, 2018, 5, 538-551.	1.7	73
23	Rett-like features and cortical visual impairment in a Japanese patient with HECW2 mutation. Brain and Development, 2018, 40, 410-414.	0.6	10
24	Mice with an Oncogenic HRAS Mutation are Resistant to High-Fat Diet-Induced Obesity and Exhibit Impaired Hepatic Energy Homeostasis. EBioMedicine, 2018, 27, 138-150.	2.7	33
25	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. Nature Communications, 2018, 9, 1960.	5.8	90
26	Novel IARS2 mutations in Japanese siblings with CAGSSS, Leigh, and West syndrome. Brain and Development, 2018, 40, 934-938.	0.6	15
27	Coâ€occurrence of hypertrophic cardiomyopathy and juvenile myelomonocytic leukemia in a neonate with Noonan syndrome, leading to premature death. Clinical Case Reports (discontinued), 2018, 6, 1202-1207.	0.2	1
28	Comprehensive targeted next-generation sequencing in Japanese familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2017, 53, 194.e1-194.e8.	1.5	47
29	Patient with a novel purineâ€rich element binding protein A mutation. Congenital Anomalies (discontinued), 2017, 57, 201-204.	0.3	15
30	Exome sequencing deciphers a germline <scp>MET</scp> mutation in familial epidermal growth factor receptorâ€mutant lung cancer. Cancer Science, 2017, 108, 1263-1270.	1.7	11
31	Phenotype-genotype correlations of PIGO deficiency with variable phenotypes from infantile lethality to mild learning difficulties. Human Mutation, 2017, 38, 805-815.	1.1	29
32	Dramatic response after functional hemispherectomy in a patient with epileptic encephalopathy carrying a de novo COL4A1 mutation. Brain and Development, 2017, 39, 337-340.	0.6	6
33	Activated Braf induces esophageal dilation and gastric epithelial hyperplasia in mice. Human Molecular Genetics, 2017, 26, 4715-4727.	1.4	13
34	Long-term outcome of a 26-year-old woman with West syndrome and an nuclear receptor subfamily 2 group F member 1 gene (NR2F1) mutation. Seizure: the Journal of the British Epilepsy Association, 2017, 50, 144-146.	0.9	11
35	Craniosynostosis in patients with RASopathies: Accumulating clinical evidence for expanding the phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 2346-2352.	0.7	32
36	Novel causative gene for radioulnar synostosis with amegakaryocytic thrombocytopenia. Japanese Journal of Thrombosis and Hemostasis, 2017, 28, 16-23.	0.1	0

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37	Metachondromatosis without Enchondromas. JBJS Case Connector, 2016, 6, e30.	0.1	3
38	Prominent sensory involvement in a case of familial amyotrophic lateral sclerosis carrying the L8V SOD1 mutation. Clinical Neurology and Neurosurgery, 2016, 150, 194-196.	0.6	6
39	Variants in the UBR1 gene are not associated with chronic pancreatitis in Japan. Pancreatology, 2016, 16, 814-818.	0.5	3
40	Somatic BRAF c.1799T>A p.V600E Mosaicism syndrome characterized by a linear syringocystadenoma papilliferum, anaplastic astrocytoma, and ocular abnormalities. American Journal of Medical Genetics, Part A, 2016, 170, 189-194.	0.7	22
41	Human genetic variation database, a reference database of genetic variations in the Japanese population. Journal of Human Genetics, 2016, 61, 547-553.	1.1	270
42	Spectrum of mutations and genotypeâ€“phenotype analysis in Noonan syndrome patients with RIT1 mutations. Human Genetics, 2016, 135, 209-222.	1.8	75
43	Recent advances in RASopathies. Journal of Human Genetics, 2016, 61, 33-39.	1.1	290
44	Adult mice expressing aBrafQ241R mutation on an ICR/CD-1 background exhibit a cardio-facio-cutaneous syndrome phenotype. Human Molecular Genetics, 2015, 24, 7349-7360.	1.4	17
45	Isolated inclusion body myopathy caused by a multisystem proteinopathyâ€“linked hnRNPA1 mutation. Neurology: Genetics, 2015, 1, e23.	0.9	34
46	Variants in pancreatic carboxypeptidase genesCPA2andCPB1are not associated with chronic pancreatitis. American Journal of Physiology - Renal Physiology, 2015, 309, G688-G694.	1.6	19
47	A postzygotic NRAS mutation in a patient with Schimmelpenning syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2223-2225.	0.7	24
48	Targeted Next-Generation Sequencing Effectively Analyzed the Cystic Fibrosis Transmembrane Conductance Regulator Gene in Pancreatitis. Digestive Diseases and Sciences, 2015, 60, 1297-1307.	1.1	19
49	Mutations in MECOM, Encoding Oncoprotein EVI1, Cause Radioulnar Synostosis with Amegakaryocytic Thrombocytopenia. American Journal of Human Genetics, 2015, 97, 848-854.	2.6	97
50	Genetic profile for suspected dysferlinopathy identified by targeted next-generation sequencing. Neurology: Genetics, 2015, 1, e36.	0.9	22
51	A novel heterozygous MAP2K1 mutation in a patient with Noonan syndrome with multiple lentigines. American Journal of Medical Genetics, Part A, 2015, 167, 407-411.	0.7	25
52	Mutations in PIGL in a patient with Mabry syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 777-785.	0.7	30
53	Genomic analysis identifies candidate pathogenic variants in 9 of 18 patients with unexplained West syndrome. Human Genetics, 2015, 134, 649-658.	1.8	51
54	TBX1 Mutation Identified by Exome Sequencing in a Japanese Family with 22q11.2 Deletion Syndrome-Like Craniofacial Features and Hypocalcemia. PLoS ONE, 2014, 9, e91598.	1.1	49

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55	New BRAF knockin mice provide a pathogenetic mechanism of developmental defects and a therapeutic approach in cardio-facio-cutaneous syndrome. Human Molecular Genetics, 2014, 23, 6553-6566.	1.4	49
56	GNE myopathy associated with congenital thrombocytopenia: A report of two siblings. Neuromuscular Disorders, 2014, 24, 1068-1072.	0.3	49
57	Sequential analysis of amino acid substitutions with hepatitis <scp>B</scp> virus in association with nucleoside/nucleotide analog treatment detected by deep sequencing. Hepatology Research, 2014, 44, 678-684.	1.8	2
58	A girl with Cardio-facio-cutaneous syndrome complicated with status epilepticus and acute encephalopathy. Brain and Development, 2014, 36, 61-63.	0.6	8
59	Gain-of-Function Mutations in RIT1 Cause Noonan Syndrome, a RAS/MAPK Pathway Syndrome. American Journal of Human Genetics, 2013, 93, 173-180.	2.6	279
60	Exome sequencing identifies a novel TTN mutation in a family with hereditary myopathy with early respiratory failure. Journal of Human Genetics, 2013, 58, 259-266.	1.1	33
61	Mutations in genes encoding the glycine cleavage system predispose to neural tube defects in mice and humans. Human Molecular Genetics, 2012, 21, 1496-1503.	1.4	100
62	Prevalence and clinical features of Costello syndrome and cardioâ€facioâ€cutaneous syndrome in Japan: Findings from a nationwide epidemiological survey. American Journal of Medical Genetics, Part A, 2012, 158A, 1083-1094.	0.7	74
63	Casitas B-cell lymphoma mutation in childhood T-cell acute lymphoblastic leukemia. Leukemia Research, 2012, 36, 1009-1015.	0.4	13
64	A genome-wide association study identifies RNF213 as the first Moyamoya disease gene. Journal of Human Genetics, 2011, 56, 34-40.	1.1	582
65	Non-Hodgkin Lymphoma in a Patient With Cardiofaciocutaneous Syndrome. Journal of Pediatric Hematology/Oncology, 2011, 33, e342-e346.	0.3	13
66	A familial case of LEOPARD syndrome associated with a high-functioning autism spectrum disorder. Brain and Development, 2011, 33, 576-579.	0.6	8
67	HRAS mutants identified in Costello syndrome patients can induce cellular senescence: possible implications for the pathogenesis of Costello syndrome. Journal of Human Genetics, 2011, 56, 707-715.	1.1	29
68	Molecular and clinical analysis ofRAF1in Noonan syndrome and related disorders: dephosphorylation of serine 259 as the essential mechanism for mutant activation. Human Mutation, 2010, 31, 284-294.	1.1	96
69	Mutation analysis of the SHOC2 gene in Noonan-like syndrome and in hematologic malignancies. Journal of Human Genetics, 2010, 55, 801-809.	1.1	38
70	Clinical manifestations in patients with SOS1 mutations range from Noonan syndrome to CFC syndrome. Journal of Human Genetics, 2008, 53, 834-841.	1.1	31
71	The RAS/MAPK syndromes: novel roles of the RAS pathway in human genetic disorders. Human Mutation, 2008, 29, 992-1006.	1.1	322
72	Cardio-facio-cutaneous and Noonan syndromes due to mutations in the RAS/MAPK signalling pathway: genotype phenotype relationships and overlap with Costello syndrome. Journal of Medical Genetics, 2007, 44, 763-771.	1.5	221

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73	Leukemia in Cardio-facio-cutaneous (CFC) Syndrome: A Patient With a Germline Mutation in BRAF Proto-oncogene. Journal of Pediatric Hematology/Oncology, 2007, 29, 287-290.	0.3	53
74	Molecular and clinical characterization of cardio-facio-cutaneous (CFC) syndrome: Overlapping clinical manifestations with Costello syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 799-807.	0.7	96
75	Germline KRAS and BRAF mutations in cardio-facio-cutaneous syndrome. Nature Genetics, 2006, 38, 294-296.	9.4	517
76	A novel KCNQ4 one-base deletion in a large pedigree with hearing loss: implication for the genotypeâ€“phenotype correlation. Journal of Human Genetics, 2006, 51, 455-460.	1.1	50
77	Germline mutations in HRAS proto-oncogene cause Costello syndrome. Nature Genetics, 2005, 37, 1038-1040.	9.4	597
78	Functional analysis of PTPN11/SHP-2 mutants identified in Noonan syndrome and childhood leukemia. Journal of Human Genetics, 2005, 50, 192-202.	1.1	113