Nobuhiko Okamoto

List of Publications by Year
in descending order

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91 papers	3,142 citations	236833 25 h-index	175177 52 g-index
91 all docs	91 docs citations	91 times ranked	6061 citing authors

#	Article	IF	Citations
1	Four pedigrees with aminoacyl-tRNA synthetase abnormalities. Neurological Sciences, 2022, 43, 2765-2774.	0.9	7
2	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
3	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	1.1	16
4	Genome Analysis in Sick Neonates and Infants: High-yield Phenotypes and Contribution of Small Copy Number Variations. Journal of Pediatrics, 2022, 244, 38-48.e1.	0.9	8
5	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	1.1	8
6	Maternal Uniparental Isodisomy of Chromosome 4 and 8 in Patients with Retinal Dystrophy: SRD5A3-Congenital Disorders of Glycosylation and RP1-Related Retinitis Pigmentosa. Genes, 2022, 13, 359.	1.0	4
7	Back Cover, Volume 43, Issue 7. Human Mutation, 2022, 43, .	1.1	0
8	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. Journal of Human Genetics, 2021, 66, 401-407.	1.1	15
9	Severe course with lethal hepatocellular injury and skeletal muscular dysgenesis in a neonate with infantile liver failure syndrome type 1 caused by novel <scp>LARS1</scp> mutations. American Journal of Medical Genetics, Part A, 2021, 185, 866-870.	0.7	4
10	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	1.1	18
11	Primary ovarian insufficiency in a female with phosphomannomutase-2 gene (PMM2) mutations for congenital disorder of glycosylation. Endocrine Journal, 2021, 68, 605-611.	0.7	4
12	Homozygous ADCY5 mutation causes early-onset movement disorder with severe intellectual disability. Neurological Sciences, 2021, 42, 2975-2978.	0.9	3
13	Blended phenotype of combination of <scp>HERC2</scp> and <scp>AP3B2</scp> deficiency and Angelman syndrome caused by paternal isodisomy of chromosome 15. American Journal of Medical Genetics, Part A, 2021, 185, 3092-3098.	0.7	2
14	A Japanese adult and two girls with NEDMIAL caused by de novo missense variants in DHX30. Human Genome Variation, 2021, 8, 24.	0.4	1
15	Novel CLTC variants cause new brain and kidney phenotypes. Journal of Human Genetics, 2021, , .	1.1	4
16	Siblings with MAN1B1-CDG Showing Novel Biochemical Profiles. Cells, 2021, 10, 3117.	1.8	5
17	Intellectual disability and dysmorphic features in male siblings arising from a novel TAF1 mutation. Congenital Anomalies (discontinued), 2020, 60, 40-41.	0.3	3
18	Breakpoint junction analysis for complex genomic rearrangements with the caldera volcanoâ€like pattern. Human Mutation, 2020, 41, 2119-2127.	1.1	2

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19	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. Journal of Human Genetics, 2020, 65, 751-757.	1.1	13
20	Complex chromosomal rearrangements of human chromosome 21 in a patient manifesting clinical features partially overlapped with that of Down syndrome. Human Genetics, 2020, 139, 1555-1563.	1.8	5
21	Bardetâ€“Biedl syndrome and related disorders in Japan. Journal of Human Genetics, 2020, 65, 847-853.	1.1	9
22	Genotype-phenotype correlation analysis in Japanese patients with Noonan syndrome. Endocrine Journal, 2019, 66, 983-994.	0.7	12
23	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
24	A case of de novo splice site variant in SLC35A2 showing developmental delays, spastic paraplegia, and delayed myelination. Molecular Genetics & Genomic Medicine, 2019, 7, e814.	0.6	14
25	Genomic backgrounds of Japanese patients with undiagnosed neurodevelopmental disorders. Brain and Development, 2019, 41, 776-782.	0.6	36
26	Clinical and molecular spectrum of CHOPS syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1126-1138.	0.7	20
27	Male CDPX2 patient with EBP mosaicism and asymmetrically lateralized skin lesions with strict midline demarcation. American Journal of Medical Genetics, Part A, 2019, 179, 1315-1318.	0.7	0
28	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	1.5	30
29	Okamoto syndrome has features overlapping with Auâ€“Kline syndrome and is caused by HNRNPK mutation. American Journal of Medical Genetics, Part A, 2019, 179, 822-826.	0.7	9
30	RALA mutation in a patient with autism spectrum disorder and Noonan syndromeâ€like phenotype. Congenital Anomalies (discontinued), 2019, 59, 195-196.	0.3	3
31	Natural histories of patients with Wolfâ€Hirschhorn syndrome derived from variable chromosomal abnormalities. Congenital Anomalies (discontinued), 2019, 59, 169-173.	0.3	6
32	De novo 1p35.2 microdeletion including PUM1 identified in a patient with sporadic West syndrome. Congenital Anomalies (discontinued), 2019, 59, 193-194.	0.3	2
33	Clinically diverse phenotypes and genotypes of patients with branchio-oto-renal syndrome. Journal of Human Genetics, 2018, 63, 647-656.	1.1	31
34	Primary microcephaly caused by novel compound heterozygous mutations in ASPM. Human Genome Variation, 2018, 5, 18015.	0.4	10
35	Infantile spasms related to a 5q31.2-q31.3 microdeletion including PURA. Human Genome Variation, 2018, 5, 18007.	0.4	6
36	Fetal ultrasonographic findings including cerebral hyperechogenicity in a patient with nonâ€lethal form of Raine syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 682-686.	0.7	13

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37	Three patients with Schaafâ€“Yang syndrome exhibiting arthrogryposis and endocrinological abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 707-711.	0.7	15
38	A novel homozygous DPH1 mutation causes intellectual disability and unique craniofacial features. Journal of Human Genetics, 2018, 63, 487-491.	1.1	14
39	Cover Image, Volume 176A, Number 3, March 2018. American Journal of Medical Genetics, Part A, 2018, 176, i.	0.7	0
40	A 10q21.3q22.2 microdeletion identified in a patient with severe developmental delay and multiple congenital anomalies including congenital heart defects. Congenital Anomalies (discontinued), 2018, 58, 36-38.	0.3	3
41	Progressive subglottic stenosis in a child with <scp>P</scp>allisterâ€<scp>K</scp>illian syndrome. Congenital Anomalies (discontinued), 2018, 58, 102-104.	0.3	5
42	Coffinâ€Siris syndrome and cardiac anomaly with a novel SOX11 mutation. Congenital Anomalies (discontinued), 2018, 58, 105-107.	0.3	18
43	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	2.9	132
44	A novel homozygous missense mutation in the SH3-binding motif of STAMBP causing microcephaly-capillary malformation syndrome. Journal of Human Genetics, 2018, 63, 957-963.	1.1	10
45	Neurological manifestations of 2q31 microdeletion syndrome. Congenital Anomalies (discontinued), 2017, 57, 197-200.	0.3	3
46	Patient with a novel purineâ€rich element binding protein A mutation. Congenital Anomalies (discontinued), 2017, 57, 201-204.	0.3	15
47	Novel MCA/ID syndrome with ASH1L mutation. American Journal of Medical Genetics, Part A, 2017, 173, 1644-1648.	0.7	27
48	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
49	MED13L haploinsufficiency syndrome: A de novo frameshift and recurrent intragenic deletions due to parental mosaicism. , 2017, 173, 1264-1269.		23
50	Disturbed chromosome segregation and multipolar spindle formation in a patient with <scp>CHAMP</scp> 1 mutation. Molecular Genetics & Genomic Medicine, 2017, 5, 585-591.	0.6	11
51	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	1.1	28
52	A novel genetic syndrome with STARD9 mutation and abnormal spindle morphology. American Journal of Medical Genetics, Part A, 2017, 173, 2690-2696.	0.7	7
53	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
54	A case of early onset epileptic encephalopathy with de novo mutation in SLC35A2: Clinical features and treatment for epilepsy. Brain and Development, 2017, 39, 256-260.	0.6	27

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55	The smallest de novo 20q11.2 microdeletion causing intellectual disability and dysmorphic features. Human Genome Variation, 2017, 4, 17050.	0.4	3
56	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	0.7	15
57	Tattonâ€“Brownâ€“Rahman syndrome due to 2p23 microdeletion. American Journal of Medical Genetics, Part A, 2016, 170, 1339-1342.	0.7	23
58	A novel TUBB3 mutation in a sporadic patient with asymmetric cortical dysplasia. American Journal of Medical Genetics, Part A, 2016, 170, 1076-1079.	0.7	19
59	Challenges in detecting genomic copy number aberrations using next-generation sequencing data and the eXome Hidden Markov Model: a clinical exome-first diagnostic approach. Human Genome Variation, 2016, 3, 16025.	0.4	38
60	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. Journal of Human Genetics, 2016, 61, 835-838.	1.1	27
61	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. American Journal of Human Genetics, 2016, 99, 950-961.	2.6	51
62	A novel PIGN mutation and prenatal diagnosis of inherited glycosylphosphatidylinositol deficiency. American Journal of Medical Genetics, Part A, 2016, 170, 183-188.	0.7	25
63	De novo KCNH1 mutations in four patients with syndromic developmental delay, hypotonia and seizures. Journal of Human Genetics, 2016, 61, 381-387.	1.1	38
64	Deep intronic GPR143 mutation in a Japanese family with ocular albinism. Scientific Reports, 2015, 5, 11334.	1.6	26
65	Truncating mutation in NFIA causes brain malformation and urinary tract defects. Human Genome Variation, 2015, 2, 15007.	0.4	24
66	Mutations in PIGL in a patient with Mabry syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 777-785.	0.7	30
67	A clinical study of patients with pericentromeric deletion and duplication within 16p12.2â€“p11.2. American Journal of Medical Genetics, Part A, 2014, 164, 213-219.	0.7	4
68	PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy. Neurogenetics, 2014, 15, 85-92.	0.7	57
69	De novo SOX11 mutations cause Coffinâ€“Siris syndrome. Nature Communications, 2014, 5, 4011.	5.8	118
70	Early manifestations of BPAN in a pediatric patient. American Journal of Medical Genetics, Part A, 2014, 164, 3095-3099.	0.7	30
71	KIF1A mutation in a patient with progressive neurodegeneration. Journal of Human Genetics, 2014, 59, 639-641.	1.1	53
72	Microarray and FISHâ€based genotypeâ€“phenotype analysis of 22 Japanese patients with Wolfâ€“Hirschhorn syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 597-609.	0.7	26

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73	Microdeletions of 5.5â€‰Mb (4q13.2â€“q13.3) and 4.1â€‰Mb (7p15.3â€–p21.1) associated with a saethreâ€“chotzenâ€like phenotype, severe intellectual disability, and autism. American Journal of Medical Genetics, Part A, 2013, 161, 2078-2083.	0.7	12
74	De Novo Mutations inSLC35A2Encoding a UDP-Galactose Transporter Cause Early-Onset Epileptic Encephalopathy. Human Mutation, 2013, 34, 1708-1714.	1.1	85
75	Clinical correlations of mutations affecting six components of the <scp>SWI</scp>/<scp>SNF</scp> complex: Detailed description of 21 patients and a review of the literature. American Journal of Medical Genetics, Part A, 2013, 161, 1221-1237.	0.7	91
76	109â€‰kb deletion of chromosome 4p16.3 in a patient with mild phenotype of Wolfâ€“Hirschhorn syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 1465-1469.	0.7	19
77	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
78	Pelizaeus-Merzbacher disease caused by a duplication-inverted triplication-duplication in chromosomal segments including the PLP1 region. European Journal of Medical Genetics, 2012, 55, 400-403.	0.7	28
79	Subtelomeric deletions of 1q43q44 and severe brain impairment associated with delayed myelination. Journal of Human Genetics, 2012, 57, 593-600.	1.1	20
80	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. Nature Genetics, 2012, 44, 376-378.	9.4	435
81	Novel intragenic duplications and mutations of CASK in patients with mental retardation and microcephaly with pontine and cerebellar hypoplasia (MICPCH). Human Genetics, 2012, 131, 99-110.	1.8	40
82	A de novo deletion of 20q11.2â€“q12 in a boy presenting with abnormal hands and feet, retinal dysplasia, and intractable feeding difficulty. American Journal of Medical Genetics, Part A, 2011, 155, 409-414.	0.7	10
83	Submicroscopic deletion in 7q31 encompassing CADPS2 and TSPAN12 in a child with autism spectrum disorder and PHPV. American Journal of Medical Genetics, Part A, 2011, 155, 1568-1573.	0.7	24
84	Submicroscopic deletion of 12q13 including HOXC gene cluster with skeletal anomalies and global developmental delay. American Journal of Medical Genetics, Part A, 2011, 155, 2997-3001.	0.7	10
85	Coâ€occurrence of Praderâ€“Willi and Sotos syndromes. American Journal of Medical Genetics, Part A, 2010, 152A, 2103-2109.	0.7	5
86	22q13 microduplication in two patients with common clinical manifestations: A recognizable syndrome?. American Journal of Medical Genetics, Part A, 2007, 143A, 2804-2809.	0.7	40
87	Molecular mechanisms and neuroimaging criteria for severe L1 syndrome with X-linked hydrocephalus. Journal of Neurosurgery: Pediatrics, 2006, 105, 403-412.	0.8	38
88	Germline KRAS and BRAF mutations in cardio-facio-cutaneous syndrome. Nature Genetics, 2006, 38, 294-296.	9.4	517
89	Hydrocephalus and Hirschsprungâ€™s disease with a mutation of L1CAM. Journal of Human Genetics, 2004, 49, 334-337.	1.1	60
90	Preferential Paternal Origin of Microdeletions Caused by Prezygotic Chromosome or Chromatid Rearrangements in Sotos Syndrome. American Journal of Human Genetics, 2003, 72, 1331-1337.	2.6	48

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91	New MCA/MR syndrome with generalized hypotonia, congenital hydronephrosis, and characteristic face. American Journal of Medical Genetics Part A, 1997, 68, 347-349.	2.4	9