Tomoko Uehara

List of Publications by Year
in descending order

57 papers	608 citations	759233 12 h-index	839539 18 g-index
57 all docs	57 docs citations	57 times ranked	1449 citing authors

#	Article	IF	Citations
1	Persistent Hyperplastic Primary Vitreous with Microphthalmia and Coloboma in a Patient with Okur-Chung Neurodevelopmental Syndrome. Molecular Syndromology, 2022, 13, 75-79.	0.8	3
2	A patient with compound heterozygosity of <scp>SMPD4</scp>: Another example of utility of exomeâ€based copy number analysis in autosomal recessive disorders. American Journal of Medical Genetics, Part A, 2022, 188, 613-617.	1.2	3
3	The p.<scp>Thr395Met</scp> missense variant of <scp>NFIA</scp> found in a patient with intellectual disability is a defective variant. American Journal of Medical Genetics, Part A, 2022, 188, 1184-1192.	1.2	5
4	Role of chimeric transcript formation in the pathogenesis of birth defects. Congenital Anomalies (discontinued), 2021, 61, 76-81.	0.6	11
5	Forkâ€shaped mandibular incisors as a novel phenotype of LRP5 â€associated disorder. American Journal of Medical Genetics, Part A, 2021, 185, 1544-1549.	1.2	1
6	Establishing intellectual disability as the key feature of patients with biallelic <scp>RNPC3</scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 1836-1840.	1.2	3
7	Clinical spectrum of individuals with de novo <scp>EBF3</scp> variants or deletions. American Journal of Medical Genetics, Part A, 2021, 185, 2913-2921.	1.2	7
8	Recurrent <scp>NFIA K125E</scp> substitution represents a lossâ€ofâ€function allele: Sensitive in vitro and in vivo assays forÂnontruncating alleles. American Journal of Medical Genetics, Part A, 2021, 185, 2084-2093.	1.2	9
9	Coloboma may be a shared feature in a spectrum of disorders caused by mutations in the <scp>WDR37â€PACS1â€PACS2</scp> axis. American Journal of Medical Genetics, Part A, 2021, 185, 884-888.	1.2	11
10	Biallelic loss of OTUD7A causes severe muscular hypotonia, intellectual disability, and seizures. American Journal of Medical Genetics, Part A, 2021, 185, 1182-1186.	1.2	14
11	The novel and recurrent variants in exon 31 of CREBBP in Japanese patients with Menkeâ€“Hennekam syndrome. American Journal of Medical Genetics, Part A, 2021, 188, 446.	1.2	5
12	Hereditary spastic paraplegia masqueraded by congenital melanocytic nevus syndrome: Dual pathogenesis of germline non-mosaicism and somatic mosaicism. European Journal of Medical Genetics, 2020, 63, 103803.	1.3	2
13	Pathogenesis of CDK8-associated disorder: two patients with novel CDK8 variants and in vitro and in vivo functional analyses of the variants. Scientific Reports, 2020, 10, 17575.	3.3	7
14	Learning disability and myoclonic epilepsy associated with apparently synonymous but spliceâ€disrupting <scp>JMJD1C</scp> variant that led to 21â€‰bp deletion of the transcript. American Journal of Medical Genetics, Part A, 2020, 182, 3064-3067.	1.2	0
15	Hypercoagulopathy Associated With Uniparental Disomy of Chromosome 2. Journal of Pediatric Hematology/Oncology, 2020, 42, 370-371.	0.6	0
16	Parallel detection of single nucleotide variants and copy number variants with exome analysis: Validation in a cohort of 700 undiagnosed patients. American Journal of Medical Genetics, Part A, 2020, 182, 2529-2532.	1.2	12
17	Protein elongation variant of <scp>PUF60</scp>: Milder phenotypic end of the Verheij syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2709-2714.	1.2	7
18	Consecutive medical exome analysis at a tertiary center: Diagnostic and healthâ€economic outcomes. American Journal of Medical Genetics, Part A, 2020, 182, 1601-1607.	1.2	10

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19	Shortfall of exome analysis for diagnosis of <scp>Shwachmanâ€Diamond</scp> syndrome: Mismapping due to the pseudogene <scp>SBDSP1</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 1631-1636.	1.2	11
20	Diagnostic utility of integrated analysis of exome and transcriptome: Successful diagnosis of Auâ€Kline syndrome in a patient with submucous cleft palate, scaphocephaly, and intellectual disabilities. Molecular Genetics & Genomic Medicine, 2020, 8, e1364.	1.2	6
21	Blended phenotype of AP4E1 deficiency and Angelman syndrome caused by paternal isodisomy of chromosome 15. Brain and Development, 2020, 42, 289-292.	1.1	6
22	Biallelic Mutations in the LSR Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis. Journal of Pediatrics, 2020, 221, 251-254.	1.8	11
23	A paradoxical thrombogenic mutation in factor II at the target site of arthropod bleeding toxin. European Journal of Medical Genetics, 2019, 62, 93-95.	1.3	5
24	IFT172 as the 19th gene causative of oralâ€facialâ€digital syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2510-2513.	1.2	9
25	A case of autism spectrum disorder with cleft lip and palate carrying a mutation in exon 8 of AUTS2. Clinical Case Reports (discontinued), 2019, 7, 2059-2063.	0.5	5
26	De novo NSF mutations cause early infantile epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2019, 6, 2334-2339.	3.7	10
27	CNOT2 haploinsufficiency causes a neurodevelopmental disorder with characteristic facial features. American Journal of Medical Genetics, Part A, 2019, 179, 2506-2509.	1.2	9
28	Severe Noonan syndrome phenotype associated with a germline Q71R MRAS variant: a recurrent substitution in RAS homologs in various cancers. American Journal of Medical Genetics, Part A, 2019, 179, 1628-1630.	1.2	13
29	Noninvasive diagnosis of TRIT1â€related mitochondrial disorder by measuring i⁶A37 and ms²i⁶A37 modifications in tRNAs from blood and urine samples. American Journal of Medical Genetics, Part A, 2019, 179, 1609-1614.	1.2	6
30	Pathogenetic basis of Takenouchi-Kosaki syndrome: Electron microscopy study using platelets in patients and functional studies in a Caenorhabditis elegans model. Scientific Reports, 2019, 9, 4418.	3.3	16
31	SATB2â€associated syndrome in patients from Japan: Linguistic profiles. American Journal of Medical Genetics, Part A, 2019, 179, 896-899.	1.2	11
32	CNOT2 as the critical gene for phenotypes of 12q15 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 659-662.	1.2	8
33	Cover Image, Volume 179A, Number 6, June 2019. American Journal of Medical Genetics, Part A, 2019, 179, i.	1.2	0
34	Further evidence of a causal association between AGO1, a critical regulator of microRNA formation, and intellectual disability/autism spectrum disorder. European Journal of Medical Genetics, 2019, 62, 103537.	1.3	16
35	Schuursâ€Hoeijmakers syndrome in two patients from Japan. American Journal of Medical Genetics, Part A, 2019, 179, 341-343.	1.2	16
36	Three patients with DeSantoâ€Shinawi syndrome: Further phenotypic delineation. American Journal of Medical Genetics, Part A, 2018, 176, 1335-1340.	1.2	12

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37	Growth pattern of Rahman syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 712-714.	1.2	14
38	Novel BICD2 mutation in a Japanese family with autosomal dominant lower extremity-predominant spinal muscular atrophy-2. Brain and Development, 2018, 40, 343-347.	1.1	8
39	Biallelic mutations in NALCN: Expanding the genotypic and phenotypic spectra of IHPRF1. American Journal of Medical Genetics, Part A, 2018, 176, 431-437.	1.2	16
40	Redefining the phenotypic spectrum of de novo heterozygous CDK13 variants: Three patients without cardiac defects. European Journal of Medical Genetics, 2018, 61, 243-247.	1.3	14
41	Ablepharon and craniosynostosis in a patient with a localized TWIST1 basic domain substitution. American Journal of Medical Genetics, Part A, 2018, 176, 2777-2780.	1.2	7
42	Haploinsufficiency of NCOR1 associated with autism spectrum disorder, scoliosis, and abnormal palatogenesis. American Journal of Medical Genetics, Part A, 2018, 176, 2466-2469.	1.2	5
43	Expanding Phenotype of Nephronophthisis-Related Ciliopathy: an Elderly Patient with Homozygous RPGRIP1L Mutation. Nephron, 2018, 140, 74-78.	1.8	2
44	Systemic lupus erythematosus in a patient with Noonan syndromeâ€like disorder with loose anagen hair 1: More than a chance association. American Journal of Medical Genetics, Part A, 2018, 176, 1662-1666.	1.2	16
45	Further evidence that a blepharophimosis syndrome phenotype is associated with a specific class of mutation in the ADNP gene. American Journal of Medical Genetics, Part A, 2017, 173, 1631-1634.	1.2	10
46	Cover Image, Volume 173A, Number 5, May 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
47	Diagnostic use of computational retrotransposon detection: Successful definition of pathogenetic mechanism in a ciliopathy phenotype. , 2017, 173, 1353-1357.		9
48	Preaxial polydactyly in an individual with Wiedemann-Steiner syndrome caused by a novel nonsense mutation in KMT2A. , 2017, 173, 2821-2825.		11
49	Truncating mutation in CSNK2B and myoclonic epilepsy. Human Mutation, 2017, 38, 1611-1612.	2.5	18
50	Coâ€occurrence of Sturgeâ€“Weber syndrome and Klippelâ€“Trenaunayâ€“Weber syndrome phenotype: Consideration of the historical aspect. American Journal of Medical Genetics, Part A, 2017, 173, 2831-2833.	1.2	6
51	Expansion of the phenotype of Kosaki overgrowth syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2422-2427.	1.2	27
52	Establishing SON in 21q22.11 as a cause a new syndromic form of intellectual disability: Possible contribution to Braddockâ€“Carey syndrome phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2587-2590.	1.2	35
53	Further evidence of a mutation in CDC42 as a cause of a recognizable syndromic form of thrombocytopenia. American Journal of Medical Genetics, Part A, 2016, 170, 852-855.	1.2	49
54	Hirschsprung disease as a yet undescribed phenotype in a patient with ARID1B mutation. American Journal of Medical Genetics, Part A, 2016, 170, 3249-3252.	1.2	20

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55	Cover Image, Volume 170A, Number 12, December 2016. , 2016, 170, i-i.		0
56	The Tumor Suppressor BCL7B Functions in the Wnt Signaling Pathway. PLoS Genetics, 2015, 11, e1004921.	3.5	33
57	H+/myo-inositol transporter genes, hmit-1.1 and hmit-1.2, have roles in the osmoprotective response in Caenorhabditis elegans. Biochemical and Biophysical Research Communications, 2011, 410, 471-477.	2.1	28