Koji Muroya

List of Publications by Year
in descending order

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115	4,164	172457	123424
papers	citations	29 h-index	61 g-index

117	117	117	4533
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Pseudoautosomal deletions encompassing a novel homeobox gene cause growth failure in idiopathic short stature and Turner syndrome. Nature Genetics, 1997, 16, 54-63.	21.4	867
2	SAMD9 mutations cause a novel multisystem disorder, MIRAGE syndrome, and are associated with loss of chromosome 7. Nature Genetics, 2016, 48, 792-797.	21.4	243
3	Burosumab versus conventional therapy in children with X-linked hypophosphataemia: a randomised, active-controlled, open-label, phase 3 trial. Lancet, The, 2019, 393, 2416-2427.	13.7	229
4	Skeletal Features and Growth Patterns in 14 Patients with Haploinsufficiency of SHOX: Implications for the Development of Turner Syndrome. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 4613-4621.	3.6	162
5	GATA3 abnormalities and the phenotypic spectrum of HDR syndrome. Journal of Medical Genetics, 2001, 38, 374-380.	3.2	151
6	PTPN11(Protein-Tyrosine Phosphatase, Nonreceptor-Type 11) Mutations in Seven Japanese Patients with Noonan Syndrome. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 3529-3533.	3.6	106
7	Transcription Factor Mutations and Congenital Hypothyroidism: Systematic Genetic Screening of a Population-Based Cohort of Japanese Patients. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 1981-1985.	3.6	105
8	A Member of a Gene Family on Xp22.3, VCX-A, Is Deleted in Patients with X-Linked Nonspecific Mental Retardation. American Journal of Human Genetics, 2000, 67, 563-573.	6.2	104
9	Heterozygous Orthodenticle Homeobox 2 Mutations Are Associated with Variable Pituitary Phenotype. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 756-764.	3.6	98
10	Turner Syndrome and Xp Deletions: Clinical and Molecular Studies in 47 Patients. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5498-5508.	3.6	92
11	Molecular Basis of Thyroid Dyshormonogenesis: Genetic Screening in Population-Based Japanese Patients. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1838-E1842.	3.6	82
12	TSHRMutations as a Cause of Congenital Hypothyroidism in Japan: A Population-Based Genetic Epidemiology Study. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 1317-1323.	3.6	80
13	The W258X mutation in SLC22A12 is the predominant cause of Japanese renal hypouricemia. Pediatric Nephrology, 2004, 19, 728-733.	1.7	79
14	OTX2 Mutation in a Patient with Anophthalmia, Short Stature, and Partial Growth Hormone Deficiency: Functional Studies Using the IRBP, HESX1, and POU1F1 Promoters. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 3697-3702.	3.6	76
15	Sex-Determining Gene(s) on Distal 9p: Clinical and Molecular Studies in Six Cases*. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 3094-3100.	3.6	75
16	Genetic evidence for a novel gene(s) involved in urogenital development on 10q26. Kidney International, 2000, 58, 2281-2290.	5.2	57
17	Molecular and Clinical Studies in 138 Japanese Patients with Silver-Russell Syndrome. PLoS ONE, 2013, 8, e60105.	2.5	55
18	Variability of biochemical and clinical phenotype in X-linked liver glycogenosis with mutations in the phosphorylase kinase PHKA2 gene. Human Genetics, 1998, 102, 423-429.	3.8	54

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19	Endocrine and Radiological Studies in Patients with Molecularly Confirmed CHARGE Syndrome. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 920-924.	3.6	44
20	Next generation sequencing-based mutation screening of 86 patients with idiopathic short stature. Endocrine Journal, 2017, 64, 947-954.	1.6	41
21	Mass screening of newborns for congenital hypothyroidism of central origin by free thyroxine measurement of blood samples on filter paper. European Journal of Endocrinology, 2012, 166, 829-838.	3.7	39
22	CAG repeat length of the androgen receptor gene in Japanese males with cryptorchidism. Molecular Human Reproduction, 2000, 6, 973-975.	2.8	38
23	Rare pseudoautosomal copy-number variations involving SHOX and/or its flanking regions in individuals with and without short stature. Journal of Human Genetics, 2015, 60, 553-556.	2.3	37
24	Estrogen receptor alpha gene polymorphism is associated with idiopathic azoospermia. Fertility and Sterility, 2002, 78, 1341-1343.	1.0	36
25	Molecular basis of non-syndromic hypospadias: systematic mutation screening and genome-wide copy-number analysis of 62 patients. Human Reproduction, 2015, 30, 499-506.	0.9	36
26	Microphthalmia with linear skin defects syndrome in a mosaic female infant with monosomy for the Xp22 region: molecular analysis of the Xp22 breakpoint and the X-inactivation pattern. Human Genetics, 1998, 103, 51-56.	3.8	34
27	Discordant Genotype-Phenotype Correlation in Familial Hyperaldosteronism Type III with KCNJ5 Gene Mutation: A Patient Report and Review of the Literature. Hormone Research in Paediatrics, 2014, 82, 138-142.	1.8	34
28	Random X-inactivation in a girl with duplication Xp11.21-p21.3: Report of a patient and review of the literature. American Journal of Medical Genetics Part A, 1999, 86, 44-50.	2.4	33
29	Turner Syndrome and Xp Deletions: Clinical and Molecular Studies in 47 Patients. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5498-5508.	3.6	32
30	Long-Term 3,5,3â€²-Triiodothyroacetic Acid Therapy in a Child with Hyperthyroidism Caused by Thyroid Hormone Resistance: Pharmacological Study and Therapeutic Recommendations. Thyroid, 2012, 22, 1069-1075.	4.5	30
31	Abnormal Basiocciput Development in CHARGE Syndrome. American Journal of Neuroradiology, 2009, 30, 629-634.	2.4	29
32	GATA3 abnormalities in six patients with HDR syndrome. Endocrine Journal, 2011, 58, 117-121.	1.6	29
33	Nonclassic TSH Resistance:TSHRMutation Carriers with Discrepantly High Thyroidal Iodine Uptake. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1340-E1345.	3.6	29
34	Functional characterization of four novel PAX8 mutations causing congenital hypothyroidism: new evidence for haploinsufficiency as a disease mechanism. European Journal of Endocrinology, 2012, 167, 625-632.	3.7	28
35	Increased Na reabsorption via the Naâ€“Cl cotransporter in autosomal recessive pseudohypoaldosteronism. Clinical and Experimental Nephrology, 2010, 14, 228-232.	1.6	26
36	Gradual Loss of ACTH Due to a Novel Mutation in LHX4: Comprehensive Mutation Screening in Japanese Patients with Congenital Hypopituitarism. PLoS ONE, 2012, 7, e46008.	2.5	26

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37	Patient-Reported Outcomes from a Randomized, Active-Controlled, Open-Label, Phase 3 Trial of Burosumab Versus Conventional Therapy in Children with X-Linked Hypophosphatemia. Calcified Tissue International, 2021, 108, 622-633.	3.1	26
38	Systematic molecular analyses of SHOX in Japanese patients with idiopathic short stature and Leriâ€“Weill dyschondrosteosis. Journal of Human Genetics, 2016, 61, 585-591.	2.3	25
39	Partial lipodystrophy in patients who have undergone hematopoietic stem cell transplantation during childhood: an institutional cross-sectional survey. Clinical Pediatric Endocrinology, 2017, 26, 99-108.	0.8	25
40	Undermasculinized genitalia in a boy with an abnormally expanded CAG repeat length in the androgen receptor geneâ˜…. Clinical Endocrinology, 2001, 54, 835-838.	2.4	24
41	Micropenis and the AR Gene: Mutation and CAG Repeat-Length Analysis. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5372-5378.	3.6	24
42	Anorectal and urinary anomalies and aberrant retinoic acid metabolism in cytochrome P450 oxidoreductase deficiency. Molecular Genetics and Metabolism, 2010, 100, 269-273.	1.1	24
43	Combined Growth Hormone and Thyroid-Stimulating Hormone Deficiency in a Japanese Patient with a Novel Frameshift Mutation in IGSF1. Hormone Research in Paediatrics, 2015, 84, 349-354.	1.8	23
44	Mental retardation in a boy with an interstitial deletion at Xp22.3 involving STS, KAL1, and OA1: Implication for the MRX locus. , 1996, 64, 583-587.		21
45	Screening for mutations of the androgen receptor gene in patients with isolated cryptorchidism. Fertility and Sterility, 2001, 76, 834-836.	1.0	21
46	SHOX Nullizygosity and Haploinsufficiency in a Japanese Family: Implication for the Development of Turner Skeletal Features. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 1390-1394.	3.6	20
47	47,XXX male: A clinical and molecular study. American Journal of Medical Genetics Part A, 2001, 98, 353-356.	2.4	19
48	Neonatal case of classic maple syrup urine disease: Usefulness of <scp>¹Hâ€MRS</scp> in early diagnosis. Pediatrics International, 2014, 56, 112-115.	0.5	19
49	Association between monoallelic TSHR mutations and congenital hypothyroidism: a statistical approach. European Journal of Endocrinology, 2018, 178, 137-144.	3.7	19
50	Identification of Novel Low-Dose Bisphenol A Targets in Human Foreskin Fibroblast Cells Derived from Hypospadias Patients. PLoS ONE, 2012, 7, e36711.	2.5	17
51	Classic Bartter syndrome complicated with profound growth hormone deficiency: a case report. Journal of Medical Case Reports, 2013, 7, 283.	0.8	17
52	Clinical manifestations and enzymatic activities of mitochondrial respiratory chain complexes in Pearson marrow-pancreas syndrome with 3-methylglutaconic aciduria: a case report and literature review. European Journal of Pediatrics, 2015, 174, 1593-1602.	2.7	17
53	Mutation spectrum and phenotypic variation in nine patients with SOX2 abnormalities. Journal of Human Genetics, 2014, 59, 353-356.	2.3	16
54	Heterozygous defects in PAX6 gene and congenital hypopituitarism. European Journal of Endocrinology, 2015, 172, 37-45.	3.7	16

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55	Clinical and molecular studies in 15 females with ring X chromosomes: implications for r(X) formation and mental development. Human Genetics, 2000, 107, 433-439.	3.8	15
56	A Novel Mutation in SOX2 Causes Hypogonadotropic Hypogonadism with Mild Ocular Malformation. Hormone Research in Paediatrics, 2014, 81, 133-138.	1.8	15
57	Genetics of Congenital Isolated TSH Deficiency: Mutation Screening of the Known Causative Genes and a Literature Review. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 6229-6237.	3.6	15
58	PAX8 Mutation Disturbing Thyroid Follicular Growth: A Case Report. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E2039-E2044.	3.6	14
59	Combined pituitary hormone deficiency with unique pituitary dysplasia and morning glory syndrome related to a heterozygous <i>PROKR2</i> mutation. Clinical Pediatric Endocrinology, 2015, 24, 27-32.	0.8	14
60	Novel Splice Site Mutation in MAMLD1 in a Patient with Hypospadias. Sexual Development, 2015, 9, 130-135.	2.0	14
61	Prevalence of Obesity, Hyperlipemia and Insulin Resistance in Children with Suprasellar Brain Tumors. Clinical Pediatric Endocrinology, 2007, 16, 1-9.	0.8	14
62	Mother and daughter with 45,X/46,X,r(X)(p22.3q28) and mental retardation: Analysis of the x-inactivation patterns. American Journal of Medical Genetics Part A, 2000, 91, 267-272.	2.4	13
63	Individual Variation of the Genetic Response to Bisphenol A in Human Foreskin Fibroblast Cells Derived from Cryptorchidism and Hypospadias Patients. PLoS ONE, 2012, 7, e52756.	2.5	13
64	First case of a Japanese girl with Myhre syndrome due to a heterozygous SMAD4 mutation. American Journal of Medical Genetics, Part A, 2012, 158A, 1982-1986.	1.2	13
65	A case of paternal uniparental isodisomy for chromosome 7 associated with overgrowth. Journal of Medical Genetics, 2018, 55, 567-570.	3.2	13
66	The ratio of serum free triiodothyronine to free thyroxine in children: a retrospective database survey of healthy short individuals and patients with severe thyroid hypoplasia or central hypothyroidism. Thyroid Research, 2015, 8, 10.	1.5	12
67	Analysis of GBE1 mutations via protein expression studies in glycogen storage disease type IV: A report on a non-progressive form with a literature review. Molecular Genetics and Metabolism Reports, 2018, 17, 31-37.	1.1	12
68	Rare variant of the epigenetic regulator SMCHD1 in a patient with pituitary hormone deficiency. Scientific Reports, 2020, 10, 10985.	3.3	12
69	Abnormal adipose tissue distribution with unfavorable metabolic profile in five children following hematopoietic stem cell transplantation: a new etiology for acquired partial lipodystrophy. Clinical Pediatric Endocrinology, 2013, 22, 53-64.	0.8	12
70	Impaired urinary water excretion in a three-generation family. Pediatric Nephrology, 2001, 16, 820-822.	1.7	11
71	Ring chromosome 21 in a boy and a derivative chromosome 21 in the mother: Implication for ring chromosome formation. American Journal of Medical Genetics Part A, 2002, 110, 332-337.	2.4	11
72	Ectopic Calcification as Discernible Manifestation in Neonates with Pseudohypoparathyroidism Type 1a. International Journal of Endocrinology, 2009, 2009, 1-3.	1.5	11

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73	Diabetes Mellitus in a Japanese Girl with HDR Syndrome and GATA3 Mutation. Endocrine Journal, 2010, 57, 171-174.	1.6	11
74	Novel compound heterozygous mutations of the growth hormone-releasing hormone receptor gene in a case of isolated growth hormone deficiency. Growth Hormone and IGF Research, 2013, 23, 89-97.	1.1	11
75	Sporadic paraganglioma caused by de novo SDHB mutations in a 6-year-old girl. European Journal of Pediatrics, 2016, 175, 137-141.	2.7	11
76	Assessment of ATP8B1 Deficiency in Pediatric Patients With Cholestasis Using Peripheral Blood Monocyte-Derived Macrophages. EBioMedicine, 2018, 27, 187-199.	6.1	11
77	Gonadoblastoma, mixed germ cell tumor, and Y chromosomal genotype: Molecular analysis in four patients. , 1999, 25, 40-45.		10
78	A nationwide questionnaire survey targeting Japanese pediatric endocrinologists regarding transitional care in childhood, adolescent, and young adult cancer survivors. Clinical Pediatric Endocrinology, 2020, 29, 55-62.	0.8	10
79	Micropenis and the AR Gene: Mutation and CAG Repeat-Length Analysis. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5372-5378.	3.6	10
80	Female carriers of Xp22.3 deletion including MRX locus. American Journal of Medical Genetics Part A, 1999, 84, 384-385.	2.4	9
81	Androgen Receptor Gene and Male Genital Anomaly. Archives of Andrology, 2002, 48, 461-466.	1.0	9
82	Overall usefulness of newborn screening for congenital hypothyroidism by using free thyroxine measurement. Endocrine Journal, 2014, 61, 1025-1030.	1.6	9
83	Gonadal macrophage infiltration in congenital lipoid adrenal hyperplasia. European Journal of Endocrinology, 2016, 175, 127-132.	3.7	9
84	Safety and Efficacy of Burosumab in Pediatric Patients With X-Linked Hypophosphatemia: A Phase 3/4 Open-Label Trial. Journal of the Endocrine Society, 2022, 6, bvac021.	0.2	9
85	Dietary potassium restriction attenuates urinary sodium wasting in the generalized form of pseudohypoaldosteronism type 1. CEN Case Reports, 2020, 9, 133-137.	0.9	8
86	Assessment of Adenosine Triphosphatase Phospholipid Transporting 8B1 (ATP8B1) Function in Patients With Cholestasis With ATP8B1 Deficiency by Using Peripheral Blood Monocyteâ€Đerived Macrophages. Hepatology Communications, 2021, 5, 52-62.	4.3	8
87	Refinement of the locus for X-linked recessive chondrodysplasia punctata. Human Genetics, 1995, 95, 577-80.	3.8	7
88	Screening for Alagille Syndrome Mutations in the JAG1 and NOTCH2 Genes Using Denaturing High-Performance Liquid Chromatography. Genetic Testing and Molecular Biomarkers, 2007, 11, 216-227.	1.7	7
89	Somatically Acquired Isodicentric Y and Mosaic Loss of Chromosome Y in a Boy with Hypospadias. Cytogenetic and Genome Research, 2018, 154, 122-125.	1.1	7
90	Spontaneous virilization around puberty in <i>NR5A1</i>-related 46,XY sex reversal: additional case and a literature review. Endocrine Journal, 2018, 65, 1187-1192.	1.6	7

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91	DNA Methylation Status of SHOX-Flanking CpG Islands in Healthy Individuals and Short Stature Patients with Pseudoautosomal Copy Number Variations. Cytogenetic and Genome Research, 2019, 158, 56-62.	1.1	7
92	Novel CUL7 biallelic mutations alter the skeletal phenotype of 3M syndrome. Human Genome Variation, 2020, 7, 1.	0.7	7
93	Metreleptin worked in a diabetic woman with a history of hematopoietic stem cell transplantation (HSCT) during infancy: further support for the concept of â€˜HSCT-associated lipodystrophyâ€™. Endocrine Journal, 2021, 68, 399-407.	1.6	7
94	Structural analysis of a rare rearranged Y chromosome and its bearing on genotype-phenotype correlation. , 2000, 92, 256-259.		6
95	Growth Hormone Response to GH-Releasing Peptide-2 in Children. Journal of Pediatric Endocrinology and Metabolism, 2010, 23, 473-80.	0.9	6
96	Long-term clinical course in three patients with MAMLD1 mutations. Endocrine Journal, 2016, 63, 835-839.	1.6	6
97	Noonan syndrome: Genotype analysis of the Noonan syndrome critical region at chromosome 12q in a three-generation family. , 1998, 79, 153-154.		5
98	Diaphyseal medullary stenosis with malignant fibrous histiocytoma: Further evidence for loss of heterozygosity involving 9p21-22 in tumor tissue. Genes Chromosomes and Cancer, 2002, 33, 326-328.	2.8	5
99	Unfavorable lipoprotein profile in childhood cancer survivors with suprasellar brain tumorsâ€”a high Apo B level and increased small dense LDL-cholesterol. Child's Nervous System, 2009, 25, 669-675.	1.1	5
100	Congenital Hypothyroidism Due to Truncating PAX8 Mutations: A Case Series and Molecular Function Studies. Journal of Clinical Endocrinology and Metabolism, 2020, 105, .	3.6	5
101	A patient with Silver-Russell syndrome with multilocus imprinting disturbance, and Schimke immuno-osseous dysplasia unmasked by uniparental isodisomy of chromosome 2. Journal of Human Genetics, 2021, 66, 1121-1126.	2.3	4
102	SHOX farâ€downstream copyâ€number variations involving cisâ€regulatory nucleotide variants in two sisters with Leriâ€Weill dyschondrosteosis. American Journal of Medical Genetics, Part A, 2019, 179, 1778-1782.	1.2	3
103	A patient with Pendred syndrome whose goiter progressed with normal serum thyrotropin and iodine organification. American Journal of Medical Genetics, Part A, 2010, 152A, 1793-1797.	1.2	2
104	Identification of the first promoterâ€specific gainâ€ofâ€function SOX9 missense variant (p. E50K) in a patient with 46, XX ovotesticular disorder of sex development. American Journal of Medical Genetics, Part A, 2021, 185, 1067-1075.	1.2	2
105	GWAS of thyroid dysgenesis identifies a risk locus at 2q33.3 linked to regulation of Wnt signaling. Human Molecular Genetics, 2022, 31, 3967-3974.	2.9	2
106	Ruvalcaba syndrome revisited. American Journal of Medical Genetics, Part A, 2010, 152A, 1854-1857.	1.2	1
107	Polyostotic osteolysis and hypophosphatemic rickets with elevated serum fibroblast growth factor 23: A case report. American Journal of Medical Genetics, Part A, 2015, 167, 2430-2434.	1.2	1
108	Additional report on Morenoâ€Nishimuraâ€Schmidt overgrowth syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2834-2837.	1.2	1

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109	Potential utility of cinacalcet as a treatment for CDC73-related primary hyperparathyroidism: a case report. Clinical Pediatric Endocrinology, 2016, 25, 91-98.	0.8	1
110	Fluctuation of blood glucose levels in an infant with an ileostomy on continuous glucose monitoring: A case report. Clinical Pediatric Endocrinology, 2018, 27, 39-43.	0.8	0
111	Severe in utero under-virilization in a 46,XY patient with Silver-Russell syndrome with 11p15 loss of methylation. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 191-196.	0.9	0
112	Very longâ€chain acylâ€CoA dehydrogenase deficiency: No developmental delay after cardiopulmonary arrest. Pediatrics International, 2021, 63, 992-994.	0.5	0
113	A case of hypoparathyroidism, sensorineural deafness, and renal dysplasia syndrome with kidney failure and recurrent pancreatitis: Questions. Pediatric Nephrology, 2021, 36, 4067-4069.	1.7	0
114	Quantification of serum thyroid hormones using tandem mass spectrometry in patients with Down syndrome. Biomedical Chromatography, 2021, , e5249.	1.7	0
115	Screening for imprinting disorders in 58 patients with clinically diagnosed idiopathic short stature. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 1335-1339.	0.9	0