Masanori Adachi

List of Publications by Year
in descending order

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82	1,830	257450 24	302126 39
papers	citations	h-index	g-index

84	84	84	2439
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	Urine Steroid Hormone Profile Analysis in Cytochrome P450 Oxidoreductase Deficiency: Implication for the Backdoor Pathway to Dihydrotestosterone. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 2643-2649.	3.6	144
3	Compound heterozygous mutations of cytochrome P450 oxidoreductase gene (POR) in two patients with Antleyâ€“Bixler syndrome. American Journal of Medical Genetics Part A, 2004, 128A, 333-339.	2.4	101
4	Cytochrome P450 Oxidoreductase Deficiency: Identification and Characterization of Biallelic Mutations and Genotype-Phenotype Correlations in 35 Japanese Patients. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 1723-1731.	3.6	99
5	Compound Heterozygous Mutations in the Â Subunit Gene of ENaC (1627delG and 1570-1GÂA) in One Sporadic Japanese Patient with a Systemic Form of Pseudohypoaldosteronism Type 1. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 9-12.	3.6	63
6	A Novel Missense Mutation of Mineralocorticoid Receptor Gene in One Japanese Family with a Renal Form of Pseudohypoaldosteronism Type 1. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 4690-4694.	3.6	60
7	Clinical practice guidelines for congenital hyperinsulinism. Clinical Pediatric Endocrinology, 2017, 26, 127-152.	0.8	48
8	Guidelines for Mass Screening of Congenital Hypothyroidism (2014 revision). Clinical Pediatric Endocrinology, 2015, 24, 107-133.	0.8	41
9	Next generation sequencing-based mutation screening of 86 patients with idiopathic short stature. Endocrine Journal, 2017, 64, 947-954.	1.6	41
10	Molecular and clinical analyses of Japanese patients with carbamoylphosphate synthetase 1 (CPS1) deficiency. Journal of Human Genetics, 2007, 52, 349-354.	2.3	39
11	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
12	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
13	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
14	Novel SLC12A1 (NKCC2) Mutations in Two Families with Bartter Syndrome Type 1. Endocrine Journal, 2007, 54, 1003-1007.	1.6	33
15	A Male Patient Presenting with Major Clinical Symptoms of Glucocorticoid Deficiency and Skeletal Dysplasia, showing a Steroid Pattern Compatible with 17.ALPHA.-Hydroxylase/ 17, 20-Lyase Deficiency, but without Obvious CYP 17 Gene Mutations.. Endocrine Journal, 1999, 46, 285-292.	1.6	30
16	Compound Heterozygous Mutations in the Î³ Subunit Gene of ENaC (1627delG and 1570-1Gâ†'A) in One Sporadic Japanese Patient with a Systemic Form of Pseudohypoaldosteronism Type 1. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 9-12.	3.6	29
17	POR R457H is a global founder mutation causing Antleyâ€“Bixler syndrome with autosomal recessive trait. American Journal of Medical Genetics, Part A, 2006, 140A, 633-635.	1.2	28
18	A Novel Mutation in the GATA3 Gene in a Family with HDR Syndrome (Hypoparathyroidism,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 67 Td Metabolism, 2006, 19, 87-92.	0.9	28

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19	Guidelines for diagnosis and treatment of 21-hydroxylase deficiency (2014 revision). Clinical Pediatric Endocrinology, 2015, 24, 77-105.	0.8	28
20	Clinical Practice Guidelines for Hypophosphatasia*. Clinical Pediatric Endocrinology, 2020, 29, 9-24.	0.8	28
21	Increased Na reabsorption via the Naâ€“Cl cotransporter in autosomal recessive pseudohypoaldosteronism. Clinical and Experimental Nephrology, 2010, 14, 228-232.	1.6	26
22	Hypothalamo-pituitary hypothyroidism detected by neonatal screening for congenital hypothyroidism using measurement of thyroidstimulating hormone and thyroxine. Acta Paediatrica, International Journal of Paediatrics, 2002, 91, 172-177.	1.5	26
23	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	26
24	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
25	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
26	Del(X)(p21.1) in a mother and two daughters: genotype-phenotype correlation of Turner features. Human Genetics, 2000, 106, 306-310.	3.8	24
27	Improved neutrophil function in a glycogen storage disease type 1b patient after liver transplantation. European Journal of Pediatrics, 2004, 163, 202-206.	2.7	24
28	Identification of novel RMRP mutations and specific founder haplotypes in Japanese patients with cartilage-hair hypoplasia. Journal of Human Genetics, 2006, 51, 706-710.	2.3	21
29	A Novel Missense Mutation of Mineralocorticoid Receptor Gene in One Japanese Family with a Renal Form of Pseudohypoaldosteronism Type 1. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 4690-4694.	3.6	21
30	Association between monoallelic TSHR mutations and congenital hypothyroidism: a statistical approach. European Journal of Endocrinology, 2018, 178, 137-144.	3.7	19
31	Clinical Practice Guidelines for Achondroplasia*. Clinical Pediatric Endocrinology, 2020, 29, 25-42.	0.8	19
32	SPONASTRIME dysplasia: Report on a female patient with severe skeletal changes. , 1996, 66, 429-432.		18
33	A missense mutation in domain III in HSPG2 in Schwartzâ€“Jampel syndrome compromises secretion of perlecan into the extracellular space. Neuromuscular Disorders, 2015, 25, 667-671.	0.6	18
34	P1148A in fibrillin-1 is not a mutation leading to Shprintzen-Goldberg syndrome. Human Mutation, 1997, 10, 326-327.	2.5	17
35	Classic Bartter syndrome complicated with profound growth hormone deficiency: a case report. Journal of Medical Case Reports, 2013, 7, 283.	0.8	17
36	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

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37	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
38	Final height and pubertal growth in Japanese patients with congenital hypothyroidism detected by neonatal screening. Acta Paediatrica, International Journal of Paediatrics, 2003, 92, 698-703.	1.5	15
39	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
40	Clinical performance of a novel chemiluminescent enzyme immunoassay for FGF23. Journal of Bone and Mineral Metabolism, 2021, 39, 1066-1075.	2.7	15
41	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
42	Growth impairment in individuals with citrin deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 501-508.	3.6	14
43	Prevalence of Obesity, Hyperlipemia and Insulin Resistance in Children with Suprasellar Brain Tumors. Clinical Pediatric Endocrinology, 2007, 16, 1-9.	0.8	14
44	Del(X)(p21.1) in a mother and two daughters: genotype-phenotype correlation of Turner features. Human Genetics, 2000, 106, 306-310.	3.8	12
45	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
46	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
47	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
48	Abnormal steroidogenesis in three patients with Antley-Bixler syndrome: Apparent decreased activity of 17alpha-hydroxylase, 17,20-lyase and 21-hydroxylase. Pediatrics International, 2004, 46, 583-589.	0.5	11
49	Ectopic Calcification as Discernible Manifestation in Neonates with Pseudohypoparathyroidism Type 1a. International Journal of Endocrinology, 2009, 2009, 1-3.	1.5	11
50	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
51	Clinical characteristics of septo-optic dysplasia accompanied by congenital central hypothyroidism in Japan. Clinical Pediatric Endocrinology, 2017, 26, 207-213.	0.8	11
52	Final height and pubertal growth in Japanese patients with congenital hypothyroidism detected by neonatal screening. Acta Paediatrica, International Journal of Paediatrics, 2003, 92, 698-703.	1.5	11
53	Monthly Urinary Gonadotropin and Ovarian Hormone Excretory Patterns in Normal Girls and Female Patients with Idiopathic Precocious Puberty. Pediatric Research, 1996, 40, 853-860.	2.3	11
54	Young-Simpson syndrome: Further delineation of a distinct syndrome with congenital hypothyroidism, congenital heart defects, facial dysmorphism, and mental retardation. , 1999, 84, 8-11.		10

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55	Overall usefulness of newborn screening for congenital hypothyroidism by using free thyroxine measurement. Endocrine Journal, 2014, 61, 1025-1030.	1.6	9
56	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
57	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
58	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
59	Growth Hormone Response to GH-Releasing Peptide-2 in Children. Journal of Pediatric Endocrinology and Metabolism, 2010, 23, 473-80.	0.9	6
60	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
61	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
62	Foot anomalies in Antleyâ€“Bixler syndrome: three case reports. Journal of Pediatric Orthopaedics Part B, 2008, 17, 241-245.	0.6	4
63	Assessment of user-friendliness of the Norditropin FlexPro for pediatric patients treated with recombinant human growth hormone: results of an open-label user survey. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 1105-10.	0.9	4
64	Usefulness of pancreatic ultrasonography in the diagnosis of Shwachman-Bodian-Diamond syndrome. Acta Paediatrica, International Journal of Paediatrics, 2005, 94, 1686-1690.	1.5	3
65	Hyponatremia secondary to severe atopic dermatitis in early infancy. Pediatrics International, 2019, 61, 544-550.	0.5	3
66	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
67	Ruvalcaba syndrome revisited. American Journal of Medical Genetics, Part A, 2010, 152A, 1854-1857.	1.2	1
68	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
69	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
70	Hypothalamoâ€pituitary hypothyroidism detected by neonatal screening for congenital hypothyroidism using measurement of thyroidâ€stimulating hormone and thyroxine. Acta Paediatrica, International Journal of Paediatrics, 2002, 91, 172-177.	1.5	0
71	Unnoticed maternal Gravesâ€™ disease revealed by the babyâ€™s low free thyroxine in newborn screening: an underestimated condition supporting thyroid disease screening among pregnant women. Journal of Endocrinological Investigation, 2018, 41, 143-144.	3.3	0
72	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

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73	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
74	Quantification of serum thyroid hormones using tandem mass spectrometry in patients with Down syndrome. Biomedical Chromatography, 2021, , e5249.	1.7	0
75	Therapeutic Use of Oral Sodium Phosphate (Phosribbon^{Â®} Combination) Tj ETQq1 1 0.784314 rgBT /Overlock	0.8	0
76	Paradoxical Growth Hormone (GH) Response to Insulin Hypoglycemia in a Case of Diencephalic Syndrome. Clinical Pediatric Endocrinology, 1994, 3, 234-235.	0.8	0
77	Resumption of Puberty in Girls and Boys with Central Precocious Puberty After Withdrawal of Long-Term Therapy with LHRH-Analogue D-Ser-6-LHRH. Clinical Pediatric Endocrinology, 1994, 3, 45-54.	0.8	0
78	New Congenital Malformation Syndrome with Severe Short Stature, Craniosynostosis, and Generalized Osseous Dysplasia in Two Siblings; New Osseous Dysplasia in Two Siblings. Clinical Pediatric Endocrinology, 1994, 3, 83-92.	0.8	0
79	A Girl with Infant-onset Autoimmune Thyroiditis. Clinical Pediatric Endocrinology, 1995, 4, 119-120.	0.8	0
80	Optic Glioma accompanied with Somatic Overgrowth. Clinical Pediatric Endocrinology, 1996, 5, 66-67.	0.8	0
81	Cerebral Infarction in Three Infant Cases of Congenital Adrenal Hyperplasia. Clinical Pediatric Endocrinology, 1997, 6, 11-14.	0.8	0
82	â€œGrowth without Growth Hormoneâ€•in a Young Female with Remitted Langerhans Cell Histiocytosis: A Case Report. Clinical Pediatric Endocrinology, 1999, 8, 17-22.	0.8	0