Tomonobu Hasegawa

List of Publications by Year
in descending order

231 papers	6,070 citations	81434 41 h-index	104191 69 g-index
233 all docs	233 docs citations	233 times ranked	7052 citing authors

#	Article	IF	Citations
1	Potential benefits of rapid genetic testing for germline WT1 in infants with bilateral renal tumors: A case report. Pediatric Blood and Cancer, 2022, 69, e29368.	0.8	0
2	Destructive thyroiditisâ€‰without autoantibodies in an infant. Pediatrics International, 2022, 64, .	0.2	1
3	Ultrasound finding of vaginal bleeding in infants with 21â€hydroxylase deficiency. Pediatrics International, 2022, 64, e14966.	0.2	1
4	Effectiveness of dihydrotestosterone ointment on glans penis size increment in a 5â€alphaâ€reductase type 2 deficiency patient. Pediatrics International, 2022, 64, e15079.	0.2	0
5	Cartilageâ€hair hypoplasia with Tâ€cell dysfunction. Pediatrics International, 2022, 64, e15080.	0.2	3
6	High-dose fludrocortisone therapy was transiently required in a female neonate with 21-hydroxylase deficiency. Clinical Pediatric Endocrinology, 2022, 31, 93-97.	0.4	1
7	The first adult case of cytochrome <scp>P450</scp> oxidoreductase deficiency with sufficient semen volume and sperm concentration. Congenital Anomalies (discontinued), 2022, 62, 136-137.	0.3	1
8	Reference values of inside leg length and inside leg length to stature ratio for Japanese children, 0â€“12 years of age. Annals of Human Biology, 2022, 49, 1-9.	0.4	1
9	The progression of saltâ€wasting and the body weight change during the first 2Âweeks of life in classical 21â€hydroxylase deficiency patients. Clinical Endocrinology, 2021, 94, 229-236.	1.2	8
10	A Case of Luscan-Lumish Syndrome: Possible Involvement of Enhanced GH Signaling. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 718-723.	1.8	5
11	Complete androgen insensitivity syndrome with accelerated onset of puberty due to a Sertoli cell tumor. Clinical Pediatric Endocrinology, 2021, 30, 99-104.	0.4	2
12	Novel STAR gene variant in a patient with classic lipoid congenital adrenal hyperplasia and combined pituitary hormone deficiency. Human Genome Variation, 2021, 8, 6.	0.4	0
13	Sequential imaging of hyperplastic callus formation in Osteogenesis Imperfecta type V: A case report and review of the literature. Journal of Orthopaedic Science, 2021, , .	0.5	0
14	The first survey about women doctors in the Japanese Society for Pediatric Endocrinology (JSPE). Clinical Pediatric Endocrinology, 2021, 30, 121-126.	0.4	0
15	Two girls with a neonatal screening-negative 21-hydroxylase deficiency requiring treatment with hydrocortisone for virilization in late childhood. Clinical Pediatric Endocrinology, 2021, 30, 143-148.	0.4	0
16	Clinical and Immunological Analyses of Ten Patients with MIRAGE Syndrome. Journal of Clinical Immunology, 2021, 41, 709-711.	2.0	4
17	Oral sodium phenylbutyrate for hyperammonemia associated with congenital portosystemic shunt: a case report. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 407-410.	0.4	1
18	Population-based waist circumference reference values in Japanese children (0â€“6 years): comparisons with Dutch, Swedish and Turkish preschool children. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 349-356.	0.4	6

#	Article	IF	Citations
19	Inactivation of a Frameshift TSH Receptor Variant Val711Phefs*18 is Due to Acquisition of a Hydrophobic Degron. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e265-e272.	1.8	0
20	A novel SOX10 variant in a Japanese girl with Waardenburg syndrome type 4C and Kallmann syndrome. Human Genome Variation, 2020, 7, 30.	0.4	2
21	Foetal virilisation caused by overproduction of non-aromatisable 11-oxygenated C19 steroids in maternal adrenal tumour. Human Reproduction, 2020, 35, 2609-2612.	0.4	8
22	Clinical Features of 57 Patients with Lipoid Congenital Adrenal Hyperplasia: Criteria for Nonclassic Form Revisited. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e3929-e3937.	1.8	16
23	Congenital Hypothyroidism Due to Truncating PAX8 Mutations: A Case Series and Molecular Function Studies. Journal of Clinical Endocrinology and Metabolism, 2020, 105, .	1.8	5
24	A novel mutation in the ACAN gene in a family with autosomal dominant short stature and intervertebral disc disease. Human Genome Variation, 2020, 7, 44.	0.4	7
25	Congenital lipoid adrenal hyperplasia: Immunohistochemical study of testosterone synthesis in Leydig cells. IJU Case Reports, 2020, 3, 53-56.	0.1	0
26	Infant with trisomy 13 who developed acute elevation of intraocular pressure and glaucoma. Congenital Anomalies (discontinued), 2020, 60, 151-152.	0.3	1
27	Role of NPR2 mutation in idiopathic short stature: Identification of two novel mutations. Molecular Genetics & Genomic Medicine, 2020, 8, e1146.	0.6	18
28	Reference values for salivary cortisol in healthy young infants by liquid chromatographyâ€“tandem mass spectrometry. Pediatrics International, 2020, 62, 785-788.	0.2	2
29	Clinical characteristics of cytochrome P450 oxidoreductase deficiency: a nationwide survey in Japan. Endocrine Journal, 2020, 67, 853-857.	0.7	5
30	Central precocious puberty in a boy with pseudohypoparathyroidism type Ia due to a novel GNAS mutation. Clinical Pediatric Endocrinology, 2020, 29, 89-90.	0.4	1
31	A novel NPR2 mutation (p.Arg388Gln) in a patient with acromesomelic dysplasia, type Maroteaux. Clinical Pediatric Endocrinology, 2020, 29, 99-103.	0.4	3
32	Relapsing 6q24-related transient neonatal diabetes mellitus with insulin resistance: A case report. Clinical Pediatric Endocrinology, 2020, 29, 179-182.	0.4	5
33	Testosterone priming increased growth hormone peak levels in the stimulation test and suppressed gonadotropin secretion in three Japanese adolescent boys. Clinical Pediatric Endocrinology, 2020, 29, 119-121.	0.4	3
34	Rapid Growth and Early Metastasis of Papillary Thyroid Carcinoma in an Adolescent Girl with Gravesâ€™ Disease. Hormone Research in Paediatrics, 2019, 91, 210-215.	0.8	3
35	Pubertal and Adult Testicular Functions in Nonclassic Lipoid Congenital Adrenal Hyperplasia: A Case Series and Review. Journal of the Endocrine Society, 2019, 3, 1367-1374.	0.1	12
36	National anthropometric reference values and growth curves for Japanese children: history and critical review. Annals of Human Biology, 2019, 46, 287-292.	0.4	3

#	Article	IF	Citations
37	Present status of prophylactic thyroidectomy in pediatric multiple endocrine neoplasia 2: a nationwide survey in Japan 1997â€“2017. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 585-595.	0.4	5
38	Effects of pre- and post-pubertal dihydrotestosterone treatment on penile length in 5Î±-reductase type 2 deficiency. Endocrine Journal, 2019, 66, 837-842.	0.7	12
39	Acquired partial lipodystrophy with metabolic disease in children following hematopoietic stem cell transplantation: a report of two cases and a review of the literature. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 537-541.	0.4	9
40	A case report and literature review of monoallelic mutation of GHR. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 415-419.	0.4	2
41	Pubertal Development and Pregnancy Outcomes in 46,XX Patients With Nonclassic Lipoid Congenital Adrenal Hyperplasia. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 1866-1870.	1.8	11
42	MYRF haploinsufficiency causes 46,XY and 46,XX disorders of sex development: bioinformatics consideration. Human Molecular Genetics, 2019, 28, 2319-2329.	1.4	25
43	A Familial Case of a Whole Germline <i>CDC73</i> Deletion Discordant for Primary Hyperparathyroidism. Hormone Research in Paediatrics, 2019, 92, 56-63.	0.8	1
44	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.	1.8	15
45	Safety Outcomes During Pediatric GH Therapy: Final Results From the Prospective GeNeSIS Observational Program. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 379-389.	1.8	51
46	In Vivo Verification of the Pathophysiology of Lipoid Congenital Adrenal Hyperplasia in the Adrenal Cortex. Endocrinology, 2019, 160, 331-338.	1.4	3
47	Efficacy of denosumab therapy for osteoporosis-pseudoglioma syndrome with osteoporosis: a case report. Modern Rheumatology Case Reports, 2019, 3, 45-48.	0.3	2
48	Congenital pituitary hypoplasia model demonstrates hypothalamic OTX2 regulation of pituitary progenitor cells. Journal of Clinical Investigation, 2019, 130, 641-654.	3.9	43
49	WHO 2006 Child Growth Standards overestimate short stature and underestimate overweight in Japanese children. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 33-38.	0.4	9
50	Longitudinal serum and urine steroid metabolite profiling in a 46,XY infant with prenatally identified POR deficiency. Journal of Steroid Biochemistry and Molecular Biology, 2018, 178, 177-184.	1.2	6
51	Adrenocortical carcinoma characterized by gynecomastia: A case report. Clinical Pediatric Endocrinology, 2018, 27, 9-18.	0.4	6
52	Association between monoallelic TSHR mutations and congenital hypothyroidism: a statistical approach. European Journal of Endocrinology, 2018, 178, 137-144.	1.9	19
53	Methylome analysis of thyroid ectopy shows no disease-specific DNA methylation signature. Clinical Pediatric Endocrinology, 2018, 27, 235-238.	0.4	5
54	MIRAGE syndrome is a rare cause of 46,XY DSD born SGA without adrenal insufficiency. PLoS ONE, 2018, 13, e0206184.	1.1	22

#	Article	IF	Citations
55	A familial case of spondyloepiphyseal dysplasia tarda caused by a novel splice site mutation in <i>TRAPPC2</i>. Clinical Pediatric Endocrinology, 2018, 27, 193-196.	0.4	8
56	An association with hypopituitarism and 9q subtelomere deletion syndrome. Clinical Case Reports (discontinued), 2018, 6, 2371-2375.	0.2	1
57	Efficacy and Safety of Denosumab Therapy for Osteogenesis Imperfecta Patients with Osteoporosisâ€"Case Series. Journal of Clinical Medicine, 2018, 7, 479.	1.0	25
58	Comparison of serum 25-hydroxyvitamin D levels between radioimmunoassay and liquid chromatography-tandem mass spectrometry in infants and postpartum women. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 1105-1111.	0.4	8
59	Identification of compound heterozygous <i>TSHR</i> mutations (R109Q and R450H) in a patient with nonclassic TSH resistance and functional characterization of the mutant receptors. Clinical Pediatric Endocrinology, 2018, 27, 123-130.	0.4	5
60	A novel truncating mutation in MYH3 causes spondylocarpotarsal synostosis syndrome with basilar invagination. Journal of Human Genetics, 2018, 63, 1277-1281.	1.1	2
61	A pediatric case of pheochromocytoma without apparent hypertension associated with von Hippel-Lindau disease. Clinical Pediatric Endocrinology, 2018, 27, 87-93.	0.4	4
62	Trismus-pseudocamptodactyly syndrome with bilateral hypoplastic mandibular condyles and shallow mandibular fossa: A case report. Oral Science International, 2018, 15, 90-92.	0.3	1
63	A pediatric case of insulinoma and a novel MEN1 mutation: the efficacy of the combination therapy of diazoxide and cornstarch. Clinical Pediatric Endocrinology, 2018, 27, 197-199.	0.4	1
64	Fetal Goitrous Hypothyroidism and Polyhydramnios in a Patient with Compound Heterozygous <i>DUOXA2</i> Mutations. Hormone Research in Paediatrics, 2018, 90, 132-137.	0.8	6
65	A Novel Mutation in NKX2-1 Shows Dominant-Negative Effects Only in the Presence of PAX8. Thyroid, 2018, 28, 1071-1073.	2.4	7
66	Prevalence of central fatness in 1992â€"1994: 40% of Japanese boys 6â€"17 years. Endocrine Journal, 2018, 65, 213-220.	0.7	4
67	Responses to the Letter to the Editor â€œDoes growth-hormone treatment affect patients with and without a mitochondrial disorder differentially ?â€•(Vol. 27, No. 2, p. 107â€"108, 2018). Clinical Pediatric Endocrinology, 2018, 27, 201-202.	0.4	0
68	FGFR1 Analyses in Four Patients with Hypogonadotropic Hypogonadism with Split-Hand/Foot Malformation: Implications for the Promoter Region. Human Mutation, 2017, 38, 503-506.	1.1	6
69	Novel compound heterozygous mutations identified by whole exome sequencing in a Japanese patient with geroderma osteodysplastica. European Journal of Medical Genetics, 2017, 60, 635-638.	0.7	6
70	Three-Quarters Adrenalectomy for Infantile-Onset Cushing Syndrome due to Bilateral Adrenal Hyperplasia in McCune-Albright Syndrome. Hormone Research in Paediatrics, 2017, 88, 285-290.	0.8	5
71	Population Pharmacokinetics of Diazoxide in Children with Hyperinsulinemic Hypoglycemia. Hormone Research in Paediatrics, 2017, 88, 316-323.	0.8	16
72	A Novel Case of Somatic KCNJ5 Mutation in Pediatric-Onset Aldosterone-Producing Adenoma. Journal of the Endocrine Society, 2017, 1, 1056-1061.	0.1	3

#	Article	IF	Citations
73	Validation of auxological reference values for Japanese children with Noonan syndrome and comparison with growth in children with Turner syndrome. Clinical Pediatric Endocrinology, 2017, 26, 153-164.	0.4	4
74	Clinical practice guidelines for congenital hyperinsulinism. Clinical Pediatric Endocrinology, 2017, 26, 127-152.	0.4	48
75	Histological Changes in Autoimmune Hepatitis with Graves' Disease: A Child Case Report. Internal Medicine, 2017, 56, 2139-2143.	0.3	3
76	Incidence of diabetes mellitus and neoplasia in Japanese short-statured children treated with growth hormone in the Genetics and Neuroendocrinology of Short Stature International Study (GeNeSIS). Clinical Pediatric Endocrinology, 2017, 26, 229-241.	0.4	3
77	Genetic defects in pediatric-onset adrenal insufficiency in Japan. European Journal of Endocrinology, 2017, 177, 187-194.	1.9	33
78	A novel mutation of the THRB gene in a Japanese family with resistance to thyroid hormone. Clinical Pediatric Endocrinology, 2016, 25, 19-22.	0.4	3
79	Classic and non-classic 21-hydroxylase deficiency can be discriminated from P450 oxidoreductase deficiency in Japanese infants by urinary steroid metabolites. Clinical Pediatric Endocrinology, 2016, 25, 37-44.	0.4	11
80	A case of transient neonatal diabetes due to a novel mutation in <i>ABCC8</i>. Clinical Pediatric Endocrinology, 2016, 25, 139-141.	0.4	4
81	A patient with pseudohypoaldosteronism type II complicated by congenital hypopituitarism carrying a <i>KLHL3</i> mutation. Clinical Pediatric Endocrinology, 2016, 25, 127-134.	0.4	7
82	A novel dominant negative mutation in the intracellular domain of <scp>GHR</scp> is associated with growth hormone insensitivity. Clinical Endocrinology, 2016, 85, 669-671.	1.2	7
83	Feminizing Adrenocortical Carcinoma with Distinct Histopathological Findings. Internal Medicine, 2016, 55, 3301-3307.	0.3	4
84	SAMD9 mutations cause a novel multisystem disorder, MIRAGE syndrome, and are associated with loss of chromosome 7. Nature Genetics, 2016, 48, 792-797.	9.4	243
85	Gonadal macrophage infiltration in congenital lipoid adrenal hyperplasia. European Journal of Endocrinology, 2016, 175, 127-132.	1.9	9
86	A case of mature teratoma with a falsely high serum estradiol value measured with an immunoassay. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 737-9.	0.4	2
87	The Distribution and Cellular Lineages of XX and XY Cells in Gonads Associated with Ovotesticular Disorder of Sexual Development. Sexual Development, 2016, 10, 185-190.	1.1	1
88	Growth references for Japanese individuals with Noonan syndrome. Pediatric Research, 2016, 79, 543-548.	1.1	16
89	Criteria for radiologic diagnosis of hypochondroplasia in neonates. Pediatric Radiology, 2016, 46, 513-518.	1.1	11
90	Steroidogenic pathways involved in androgen biosynthesis in eumenorrheic women and patients with polycystic ovary syndrome. Journal of Steroid Biochemistry and Molecular Biology, 2016, 158, 31-37.	1.2	35

#	Article	IF	Citations
91	Transient congenital hypothyroidism caused by compound heterozygous mutations affecting the NADPH-oxidase domain of DUOX2. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 363-71.	0.4	11
92	Potential utility of cinacalcet as a treatment for CDC73-related primary hyperparathyroidism: a case report. Clinical Pediatric Endocrinology, 2016, 25, 91-98.	0.4	1
93	Co-Administration of the CYP3A4 Inhibitor Diltiazem Counteracts Mitotane-Induced Clearance of Glucocorticoids and Antihypertensives in a Patient with Adrenocortical Carcinoma. AACE Clinical Case Reports, 2016, 2, e36-e40.	0.4	1
94	Elevated Levels of Plasma Immunoassayable Aldosterone in a Mild Form of 17 Alpha-Hydroxylase/17,20-lyase Deficiency Diagnosed at the Age of 50. AACE Clinical Case Reports, 2015, 1, e156-e160.	0.4	3
95	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.	0.4	14
96	Treatment situation of male hypogonadotropic hypogonadism in pediatrics and proposal of testosterone and gonadotropins replacement therapy protocols. Clinical Pediatric Endocrinology, 2015, 24, 37-49.	0.4	20
97	Frequencies of spontaneous breast development and spontaneous menarche in Turner syndrome in Japan. Clinical Pediatric Endocrinology, 2015, 24, 167-173.	0.4	43
98	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.	0.8	23
99	Heterozygous defects in PAX6 gene and congenital hypopituitarism. European Journal of Endocrinology, 2015, 172, 37-45.	1.9	16
100	C-Type Natriuretic Peptide Plasma Levels Are Elevated in Subjects With Achondroplasia, Hypochondroplasia, and Thanatophoric Dysplasia. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E355-E359.	1.8	29
101	Waist circumference centiles by age and sex for Japanese children based on the 1978â€“1981 cross-sectional national survey data. Annals of Human Biology, 2015, 42, 56-61.	0.4	11
102	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.	1.3	17
103	Trends in thin body stature among Japanese female adolescents, 2003â€“2012. Annals of Human Biology, 2015, 42, 533-537.	0.4	10
104	Distinguishing primary from secondary Î”⁴â€3â€oxosteroid 5Î²â€reductase (<scp>SRD</scp>5B1,) Tj ETQq0 0 0 rgBT /O	1.2	4
105	Human Chorionic Gonadotropin Stimulation Test in Prepubertal Children with Micropenis Can Accurately Predict Leydig Cell Function in Pubertal or Postpubertal Adolescents. Hormone Research in Paediatrics, 2015, 84, 305-310.	0.8	11
106	Pseudodominant inheritance in a family with nonautoimmune hypothyroidism due to biallelic <scp>DUOX</scp>2 mutations. Clinical Endocrinology, 2015, 83, 394-398.	1.2	10
107	Effect of Growth Hormone Treatment on Quality of Life in Japanese Children with Growth Hormone Deficiency: An Analysis from a Prospective Observational Study. Clinical Pediatric Endocrinology, 2014, 23, 83-92.	0.4	15
108	A Cross-Sectional Growth Reference and Chart of Stretched Penile Length for Japanese Boys Aged 0-7 Years. Hormone Research in Paediatrics, 2014, 82, 388-393.	0.8	14

#	Article	IF	Citations
109	Comprehensive Next-Generation Sequencing Analyses of Hypoparathyroidism: Identification of NovelGCM2Mutations. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E2421-E2428.	1.8	20
110	Steeper increases in body mass index during childhood correlate with blood pressure elevation in adolescence: a long-term follow-up study in a Japanese community. Hypertension Research, 2014, 37, 179-184.	1.5	18
111	Trends in thin body stature among Japanese male adolescents, 2003â€“2012. Annals of Human Biology, 2014, 41, 277-281.	0.4	11
112	Neonatal case of classic maple syrup urine disease: Usefulness of <scp>¹Hâ€MRS</scp> in early diagnosis. Pediatrics International, 2014, 56, 112-115.	0.2	19
113	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.	0.8	34
114	A Novel Mutation in SOX2 Causes Hypogonadotropic Hypogonadism with Mild Ocular Malformation. Hormone Research in Paediatrics, 2014, 81, 133-138.	0.8	15
115	Two Japanese familial cases of Caffey disease with and without the common COL1A1 mutation and normal bone density, and review of the literature. European Journal of Pediatrics, 2014, 173, 799-804.	1.3	11
116	Identification and Functional Characterization of Two Novel NPR2 Mutations in Japanese Patients With Short Stature. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E713-E718.	1.8	63
117	A novel mutation in SOX3 polyalanine tract: a case of kabuki syndrome with combined pituitary hormone deficiency harboring double mutations in MLL2 and SOX3. Pituitary, 2014, 17, 569-574.	1.6	17
118	<scp>IMAG</scp>e syndrome: clinical and genetic implications based on investigations in three Japanese patients. Clinical Endocrinology, 2014, 80, 706-713.	1.2	17
119	Deletions in the 3â€² Part of theNFIXGene Including a Recurrent Alu-Mediated Deletion of Exon 6 and 7 Account for Previously Unexplained Cases of Marshall-Smith Syndrome. Human Mutation, 2014, 35, 1092-1100.	1.1	26
120	Urinary steroid profiling: a powerful method for the diagnosis of abnormal steroidogenesis. Expert Review of Endocrinology and Metabolism, 2014, 9, 273-282.	1.2	6
121	The Contribution of Serine 194 Phosphorylation to Steroidogenic Acute Regulatory Protein Function. Molecular Endocrinology, 2014, 28, 1088-1096.	3.7	23
122	A 2.0ÂMb microdeletion in proximal chromosome 14q12, involving regulatory elements of FOXG1, with the coding region of FOXG1 being unaffected, results in severe developmental delay, microcephaly, and hypoplasia of the corpus callosum. European Journal of Medical Genetics, 2013, 56, 526-528.	0.7	16
123	Characteristic Testicular Histology Is Useful for the Identification of NR5A1 Gene Mutations in Prepubertal 46,XY Patients. Hormone Research in Paediatrics, 2013, 80, 119-128.	0.8	14
124	Heterozygous Mutations in Natriuretic Peptide Receptor-B (NPR2) Gene as a Cause of Short Stature in Patients Initially Classified as Idiopathic Short Stature. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1636-E1644.	1.8	111
125	A report of two novel <scp>NR</scp>5A1 mutation families: possible clinical phenotype of psychiatric symptoms of anxiety and/or depression. Clinical Endocrinology, 2013, 78, 957-965.	1.2	16
126	Measurement of reference intervals for urinary free adrenal steroid levels in Japanese newborn infants by using stable isotope dilution gas chromatography/mass spectrometry. Clinica Chimica Acta, 2013, 415, 302-305.	0.5	9

#	Article	IF	Citations
127	Backdoor pathway for dihydrotestosterone biosynthesis: Implications for normal and abnormal human sex development. Developmental Dynamics, 2013, 242, 320-329.	0.8	97
128	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
129	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.	0.5	11
130	Congenital hypothyroidism caused by a novel mutation of the dual oxidase 2 (DUOX2) gene. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 45-52.	0.4	22
131	Spousal Choice by Height in an Urban Middle-Class Japanese Population. Human Biology, 2013, 85, 619.	0.4	0
132	A 68-Year-Old Phenotypically Male Patient with 21-Hydroxylase Deficiency and Concomitant Adrenocortical Neoplasm Producing Testosterone and Cortisol. Tohoku Journal of Experimental Medicine, 2013, 231, 75-84.	0.5	9
133	Quantitative and Sensitive Detection of GNAS Mutations Causing McCune-Albright Syndrome with Next Generation Sequencing. PLoS ONE, 2013, 8, e60525.	1.1	41
134	Association Between Gravesâ€™ Disease and Renal Coloboma Syndrome: A Case Report. Clinical Pediatric Endocrinology, 2013, 22, 45-51.	0.4	1
135	Molecular and Clinical Studies in 138 Japanese Patients with Silver-Russell Syndrome. PLoS ONE, 2013, 8, e60105.	1.1	55
136	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.	1.9	28
137	A Genome-Wide Expression Profile of Adrenocortical Cells in Knockout Mice Lacking Steroidogenic Acute Regulatory Protein. Endocrinology, 2012, 153, 2714-2723.	1.4	18
138	PAPSS2mutations cause autosomal recessive brachyolmia. Journal of Medical Genetics, 2012, 49, 533-538.	1.5	44
139	A case of Rabson-Mendenhall syndrome with a novel mutation in the tyrosine kinase domain of the insulin receptor gene complicated by medullary sponge kidney. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 587-90.	0.4	11
140	Prenatal diagnosis of osteogenesis imperfecta type II by threeâ€dimensional computed tomography: The current state of fetal computed tomography. Congenital Anomalies (discontinued), 2012, 52, 203-206.	0.3	7
141	Two-Step Biochemical Differential Diagnosis of Classic 21-Hydroxylase Deficiency and Cytochrome P450 Oxidoreductase Deficiency in Japanese Infants by GC-MS Measurement of Urinary Pregnanetriolone/ Tetrahydroxycortisone Ratio and 11Î²-Hydroxyandrosterone. Clinical Chemistry, 2012, 58, 741-747.	1.5	27
142	Favorable Impact of Growth Hormone Treatment on Cholesterol Levels in Turner Syndrome. Clinical Pediatric Endocrinology, 2012, 21, 29-34.	0.4	5
143	A Novel Mutation in LEPRE1 That Eliminates Only the KDEL ER- Retrieval Sequence Causes Non-Lethal Osteogenesis Imperfecta. PLoS ONE, 2012, 7, e36809.	1.1	28
144	BMI z-score is the optimal measure of annual adiposity change in elementary school children. Annals of Human Biology, 2011, 38, 747-751.	0.4	57

#	Article	IF	Citations
145	Syndrome of inappropriate antiâ€diuretic hormone in Kawasaki disease. Pediatrics International, 2011, 53, 354-357.	0.2	15
146	Nonclassic TSH Resistance:TSHRMutation Carriers with Discrepantly High Thyroidal Iodine Uptake. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1340-E1345.	1.8	29
147	PAX8 Mutation Disturbing Thyroid Follicular Growth: A Case Report. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E2039-E2044.	1.8	14
148	Tracking of BMI in Japanese children from 6 to 18 years of age: Reference values for annual BMI incremental change and proposal for size of increment indicative of risk for obesity. Annals of Human Biology, 2011, 38, 146-149.	0.4	9
149	Molecular Basis of Thyroid Dyshormonogenesis: Genetic Screening in Population-Based Japanese Patients. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1838-E1842.	1.8	82
150	One Novel and Two Recurrent THRB Mutations Associated with Resistance to Thyroid Hormone: Structure-based Computational Mutation Prediction. Clinical Pediatric Endocrinology, 2010, 19, 91-99.	0.4	5
151	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.	1.8	105
152	The Effect of Intramuscular Testosterone Enanthate Treatment on Stretched Penile Length in Prepubertal Boys With Hypospadias. Urology, 2010, 76, 97-100.	0.5	30
153	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.	1.8	99
154	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.	1.8	80
155	Official Japanese reports significantly underestimate prevalence of overweight in school children: Inappropriate definition of standard weight and calculation of excess weight. Annals of Human Biology, 2009, 36, 139-145.	0.4	17
156	Measurement of Serum 17.ALPHA.-hydroxyprogesterone in Newborn Infants by Stable Isotope Dilution-Gas Chromatography/Mass Spectrometry. Clinical Pediatric Endocrinology, 2009, 18, 77-80.	0.4	4
157	Molecular and clinical findings and their correlations in Silver-Russell syndrome: implications for a positive role of IGF2 in growth determination and differential imprinting regulation of the IGF2â€“H19 domain in bodies and placentas. Journal of Molecular Medicine, 2008, 86, 1171-1181.	1.7	54
158	Complex Role of the Mitochondrial Targeting Signal in the Function of Steroidogenic Acute Regulatory Protein Revealed by Bacterial Artificial Chromosome Transgenesis in Vivo. Molecular Endocrinology, 2008, 22, 951-964.	3.7	51
159	De-novo balanced translocation between 7q31 and 10p14 in a girl with central precocious puberty, moderate mental retardation, and severe speech impairment. Clinical Dysmorphology, 2008, 17, 31-34.	0.1	14
160	Haplotype analysis of the estrogen receptor 1 gene in male genital and reproductive abnormalities. Human Reproduction, 2007, 22, 1279-1284.	0.4	59
161	Intact Kinase Homology Domain of Natriuretic Peptide Receptor-B Is Essential for Skeletal Development. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 4009-4014.	1.8	67
162	Prevalence and trends of underweight and BMI distribution changes in Japanese teenagers based on the 2001 National Survey data. Annals of Human Biology, 2007, 34, 354-361.	0.4	9

#	Article	IF	Citations
163	A Novel Mutation of Androgen Receptor Gene in Complete Androgen Insensitivity Syndrome. Clinical Pediatric Endocrinology, 2007, 16, 59-61.	0.4	0
164	Growth Failure in an Infant with Congenital Nephrogenic Diabetes Insipidus During Sodium Restriction. Clinical Pediatric Endocrinology, 2007, 16, 95-98.	0.4	0
165	Body mass index reference values (mean and SD) for Japanese children. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 1674-1676.	0.7	41
166	Age-dependent percentile for waist circumference for Japanese children based on the 1992â€“1994 cross-sectional national survey data. European Journal of Pediatrics, 2007, 166, 655-661.	1.3	41
167	The Circadian Variation of Cortisol Secretion in Patients with Anorexia Nervosa in Childhood and Adolescence after Recovery of Body Weight by Treatment Using Gas Chromatography/Mass Spectrometry in Selected Ion Monitoring. Clinical Pediatric Endocrinology, 2007, 16, 17-22.	0.4	0
168	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.	1.8	144
169	Phenotypic spectrum of CHARGE syndrome with CHD7 mutations. Journal of Pediatrics, 2006, 148, 410-414.	0.9	145
170	Growth-chart-based qualitative evaluation of height growth after hematopoietic stem cell transplantation. Pediatric Transplantation, 2006, 10, 26-31.	0.5	3
171	CXorf6 is a causative gene for hypospadias. Nature Genetics, 2006, 38, 1369-1371.	9.4	136
172	Analysis of the NSD1 promoter region in patients with a Sotos syndrome phenotype. Journal of Human Genetics, 2006, 51, 15-20.	1.1	3
173	Cytochrome P450 Oxidoreductase Deficiency in Three Patients Initially Regarded as Having 21-Hydroxylase Deficiency and/or Aromatase Deficiency: Diagnostic Value of Urine Steroid Hormone Analysis. Pediatric Research, 2006, 59, 276-280.	1.1	66
174	Congenital Hypothyroidism in Peters Plus Syndrome. Ophthalmic Genetics, 2006, 27, 67-69.	0.5	5
175	Standardized centile curves of body mass index for Japanese children and adolescents based on the 1978â€“1981 national survey data. Annals of Human Biology, 2006, 33, 444-453.	0.4	69
176	Mutational Analysis of Androgen Receptor (AR) Gene in 46, XY Patients with Ambiguous Genitalia and Normal Testosterone Secretion: Endocrinological Characteristics of Three Patients with AR Gene Mutations. Clinical Pediatric Endocrinology, 2006, 15, 151-162.	0.4	1
177	Gonadotrophin therapy in Kallmann syndrome caused by heterozygous mutations of the gene for fibroblast growth factor receptor 1: report of three families:Case report. Human Reproduction, 2005, 20, 2173-2178.	0.4	24
178	Cytochrome P450 Oxidoreductase Gene Mutations and Antley-Bixler Syndrome with Abnormal Genitalia and/or Impaired Steroidogenesis: Molecular and Clinical Studies in 10 Patients. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 414-426.	1.8	138
179	Association of Cryptorchidism with a Specific Haplotype of the Estrogen Receptor Î± Gene: Implication for the Susceptibility to Estrogenic Environmental Endocrine Disruptors. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 4716-4721.	1.8	68
180	Elevated Urine Pregnanetriolone Definitively Establishes the Diagnosis of Classical 21-Hydroxylase Deficiency in Term and Preterm Neonates. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 6087-6091.	1.8	37

#	Article	IF	Citations
181	Testicular Dysgenesis without Adrenal Insufficiency in a 46,XY Patient with a Heterozygous Inactive Mutation of Steroidogenic Factor-1. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 5930-5935.	1.8	98
182	Protein-Tyrosine Phosphatase, Nonreceptor Type 11 Mutation Analysis and Clinical Assessment in 45 Patients with Noonan Syndrome. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 3359-3364.	1.8	102
183	Clinical Assessment and Mutation Analysis of Kallmann Syndrome 1 (KAL1) and Fibroblast Growth Factor Receptor 1 (FGFR1, orKAL2) in Five Families and 18 Sporadic Patients. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 1079-1088.	1.8	208
184	Transient Hyper-17-OHPnemia Unrelated to Cross-Reactions with Residual Fetal Adrenal Cortex Products. Hormone Research in Paediatrics, 2004, 61, 242-245.	0.8	2
185	Multiple polypoid masses in the gastrointestinal tract in patient with Menkes disease on copper-histidinate therapy. European Journal of Pediatrics, 2004, 163, 745-746.	1.3	6
186	Genetic testing of glycogen storage disease type Ib in Japan: five novel G6PT1 mutations and a rapid detection method for a prevalent mutation W118R. Molecular Genetics and Metabolism, 2004, 81, 343-346.	0.5	12
187	Premature ovarian failure in a female with proximal symphalangism and Noggin mutation. Fertility and Sterility, 2004, 81, 1137-1139.	0.5	43
188	A Case of a Preterm Infant with 21-Hydroxylase Deficiency: Implications of the Biochemical Diagnosis with Urinary Pregnanetriolone by Gas Chromatography/Mass Spectrometry in Selected Ion Monitoring (GCMS-SIM). Clinical Pediatric Endocrinology, 2004, 13, 65-70.	0.4	4
189	Micropenis and the 5Î±-Reductase-2 (SRD5A2) Gene: Mutation and V89L Polymorphism Analysis in 81 Japanese Patients. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 3431-3436.	1.8	60
190	Longitudinal auxological study in a female with SHOX (short stature homeobox containing gene) haploinsufficiency and normal ovarian function. European Journal of Endocrinology, 2003, 149, 337-341.	1.9	10
191	Ischiospinal dysostosis with cystic kidney disease: report of two cases. Clinical Dysmorphology, 2003, 12, 101-104.	0.1	9
192	The Roles of Circulating High-Density Lipoproteins and Trophic Hormones in the Phenotype of Knockout Mice Lacking the Steroidogenic Acute Regulatory Protein. Molecular Endocrinology, 2002, 16, 2297-2309.	3.7	51
193	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.	1.8	106
194	Characterization of the aryl hydrocarbon receptor repressor gene and association of its Pro185Ala polymorphism with micropenis. Teratology, 2002, 65, 10-18.	1.8	44
195	A case of impairment of mitochondrial fatty acid beta-oxidation. Keio Journal of Medicine, 2002, 51, 100-6.	0.5	3
196	GATA3 abnormalities and the phenotypic spectrum of HDR syndrome. Journal of Medical Genetics, 2001, 38, 374-380.	1.5	151
197	Developmental Roles of the Steroidogenic Acute Regulatory Protein (StAR) as Revealed by StAR Knockout Mice. Molecular Endocrinology, 2000, 14, 1462-1471.	3.7	227
198	Molecular Cytogenetic Analysis of Eight Inversion Duplications of Human Chromosome 13q That Each Contain a Neocentromere. American Journal of Human Genetics, 2000, 66, 1794-1806.	2.6	112

#	Article	IF	Citations
199	Developmental and Hormonal Regulation of Murine Scavenger Receptor, Class B, Type 1. Molecular Endocrinology, 1999, 13, 1460-1473.	3.7	73
200	Four novel mutations of the Fanconi anemia group A gene (FAA) in Japanese patients. Journal of Human Genetics, 1999, 44, 48-51.	1.1	14
201	Joint laxity, vitreoretinal degeneration, facial abnormalities, and generalized skeletal alterations: A new syndrome?. Journal of Human Genetics, 1998, 43, 191-194.	1.1	2
202	Insulin-like growth factor binding protein-4 accumulation is negatively correlated with growth rate in TM-3 cells. Growth Hormone and IGF Research, 1998, 8, 277-282.	0.5	4
203	Mutation of the Type X Collagen Gene (COL10A1) Causes Spondylometaphyseal Dysplasia. American Journal of Human Genetics, 1998, 63, 1659-1662.	2.6	48
204	Hypergonadotropic Hypogonadism in a 3-Year-Old Girl with Blepharophimosis, Ptosis, and Epicanthus inversus Syndrome. Hormone Research in Paediatrics, 1998, 50, 190-192.	0.8	3
205	High Ratios of Free to Total Insulin-Like Growth Factor-I in Early Infancy. Journal of Clinical Endocrinology and Metabolism, 1997, 82, 156-158.	1.8	32
206	Measurements of Serum MÃ¼llerian Inhibiting Substance in the Evaluation of Children with Nonpalpable Gonads. New England Journal of Medicine, 1997, 336, 1480-1486.	13.9	175
207	Transition from latent to overt hypoparathyroidism in a child with CATCH 22. European Journal of Pediatrics, 1996, 155, 425-426.	1.3	2
208	Plasma free insulin-like growth factor I concentrations in growth hormone deficiency in children and adolescents. European Journal of Endocrinology, 1996, 134, 184-189.	1.9	50
209	Short Term Growth Hormone Treatment for A Girl with Osteodysplastic Primordial Dwarfism Type II. Clinical Pediatric Endocrinology, 1996, 5, 139-141.	0.4	0
210	Coarctation of the aorta and renal hypoplasia in a boy with Turner/Noonan surface anomalies and a 46,XY karyotype: a clinical model for the possible impairment of a putative lymphogenic gene(s) for Turner somatic stigmata. Human Genetics, 1996, 97, 564-567.	1.8	1
211	Clinical information on serum IGFBP-3 levels and IGFBP-3 proteolytic activity in childhood. Progress in Growth Factor Research, 1995, 6, 457-463.	1.7	3
212	Multiple Hormonal Regulation of IGF Binding Protein (IGFBP) -4 in TM-3 Cells. Clinical Pediatric Endocrinology, 1994, 3, 179-180.	0.4	2
213	Physical mapping of the split hand/split foot locus on chromosome 7 and implication in syndromic ectrodactyly. Human Molecular Genetics, 1994, 3, 1345-1354.	1.4	125
214	Single amino acid substitution (840Arg â†' His) in the hormone-binding domain of the androgen receptor leads to incomplete androgen insensitivity syndrome associated with a thermolabile androgen receptor. European Journal of Endocrinology, 1994, 130, 569-574.	1.9	28
215	Clinical utility of insulin-like growth factor binding protein-3 in the evaluation and treatment of short children with suspected growth hormone deficiency. European Journal of Endocrinology, 1994, 131, 27-32.	1.9	54
216	Currarino triad (anorectal malformation, sacral bony abnormality and presacral mass) with partial trisomy of chromosomes 13q and 20p. Clinical Genetics, 1994, 45, 272-273.	1.0	23

#	Article	IF	Citations
217	Clinical Utility of Newly Developed Plasma Free Insulin-like Growth Factor I (IGF-I) Measurements by Immunoradiometric Assay (IRMA) -Preliminary Results in Growth Hormone Deficiency (GHD) Adults and Pregnant Women. Clinical Pediatric Endocrinology, 1994, 3, 160-161.	0.4	3
218	Ratio of False Positive Results of Growth Hormone Stimulation Tests (Arginine and Insulin). Clinical Pediatric Endocrinology, 1993, 2, 65-67.	0.4	6
219	The breakpoints of the EEC syndrome (ectrodactyly, ectodermal dysplasia and cleft lip/palate) confirmed to 7q11.21 and 9p12 by fluorescence in situ hybridization. Clinical Genetics, 1993, 44, 50-50.	1.0	21
220	A Patient with Short Stature with Normal Growth Hormone Provocative Tests and Low Physiological 24 hr Growth Hormone Concentration. Clinical Pediatric Endocrinology, 1993, 2, 33-38.	0.4	3
221	Syndrome of Inappropriate Antidiuretic Hormone Secretion due to Osmoreceptor Resetting in a Patient with Juvenile Rheumatoid Arthritis Associated with Pleural Effusion. Clinical Pediatric Endocrinology, 1993, 2, 17-20.	0.4	0
222	Insulin-like Growth Factor Binding Protein-3 (IGFBP-3) Measurements in the Diagnosis of Growth Hormone Insufficiency and Comparison with IGF-I Measurements. Clinical Pediatric Endocrinology, 1993, 2, 31-37.	0.4	2
223	Western Ligand Blot Assay for Human Growth Hormone-Dependent Insulin-Like Growth Factor Binding Protein (IGFBP-3): The Serum Levels in Patients with Classical Growth Hormone Deficiency.. Endocrinologia Japonica, 1992, 39, 121-127.	0.5	10
224	Gradual Progress of ACTH Deficiency in a Child with Panhypopituartarism Associated with Pituitary Stalk Transection. Endocrinologia Japonica, 1992, 39, 165-167.	0.5	7
225	Spontaneous Growth Hormone Secretion in Healthy Prepubertal Children of Normal Stature.. Endocrinologia Japonica, 1992, 39, 9-12.	0.5	5
226	Usefulness and Limitation of Measurement of Insulin-Like Growth Factor Binding Protein-3(IGFBP-3) for Diagnosis of Growth Hormone Deficiency.. Endocrinologia Japonica, 1992, 39, 585-591.	0.5	33
227	An Unusual Variant of Chromosome 16 in Three Generations. Pediatrics International, 1992, 34, 166-168.	0.2	5
228	Infantile polymyositis: A case report. Brain and Development, 1992, 14, 167-169.	0.6	11
229	Partial Growth Hormone Deficiency with Pituitary Stalk Transection.. Endocrinologia Japonica, 1991, 38, 573-575.	0.5	2
230	EEC syndrome (ectrodactyly, ectodermal dysplasia and cleft lip/palate) with a balanced reciprocal translocation between 7q11.21 and 9p12 (or 7p11.2 and 9q12) in three generations. Clinical Genetics, 1991, 40, 202-206.	1.0	44
231	Triphasic AVP Secretion in Encephalopathy.. Endocrinologia Japonica, 1990, 37, 171-175.	0.5	2