Tsutomu Ogata

List of Publications by Year in descending order

364 papers	13,481 citations	28274 55 h-index	34986 98 g-index
366 all docs	366 docs citations	366 times ranked	13496 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	Mutation of ARX causes abnormal development of forebrain and testes in mice and X-linked lissencephaly with abnormal genitalia in humans. Nature Genetics, 2002, 32, 359-369.	21.4	647
3	Genomic imprinting at the mammalian Dlk1-Dio3 domain. Trends in Genetics, 2008, 24, 306-316.	6.7	362
4	Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124.	9.6	336
5	Genome-wide parent-of-origin DNA methylation analysis reveals the intricacies of human imprinting and suggests a germline methylation-independent mechanism of establishment. Genome Research, 2014, 24, 554-569.	5.5	311
6	Role of retrotransposon-derived imprinted gene, Rtl1, in the feto-maternal interface of mouse placenta. Nature Genetics, 2008, 40, 243-248.	21.4	300
7	Gain-of-Function Mutations in RIT1 Cause Noonan Syndrome, a RAS/MAPK Pathway Syndrome. American Journal of Human Genetics, 2013, 93, 173-180.	6.2	279
8	Deletions and epimutations affecting the human 14q32.2 imprinted region in individuals with paternal and maternal upd(14)-like phenotypes. Nature Genetics, 2008, 40, 237-242.	21.4	266
9	Turner syndrome and female sex chromosome aberrations: deduction of the principal factors involved in the development of clinical features. Human Genetics, 1995, 95, 607-29.	3.8	229
10	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.	3.6	208
11	Deletions of the Homeobox GeneSHOX(Short Stature Homeobox) Are an Important Cause of Growth Failure in Children with Short Stature. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 1402-1406.	3.6	188
12	The IG-DMR and the MEG3-DMR at Human Chromosome 14q32.2: Hierarchical Interaction and Distinct Functional Properties as Imprinting Control Centers. PLoS Genetics, 2010, 6, e1000992.	3.5	185
13	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
14	GATA3 abnormalities and the phenotypic spectrum of HDR syndrome. Journal of Medical Genetics, 2001, 38, 374-380.	3.2	151
15	Uniparental disomy and human disease: An overview. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2010, 154C, 329-334.	1.6	151
16	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
17	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.	3.6	138
18	CXorf6 is a causative gene for hypospadias. Nature Genetics, 2006, 38, 1369-1371.	21.4	136

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19	Characterization of DNA methylation errors in patients with imprinting disorders conceived by assisted reproduction technologies. Human Reproduction, 2012, 27, 2541-2548.	0.9	122
20	Fifty microdeletions among 112 cases of Sotos syndrome: Low copy repeats possibly mediate the common deletion. Human Mutation, 2003, 22, 378-387.	2.5	121
21	Silver-Russell syndrome in a girl born after in vitro fertilization: partial hypermethylation at the differentially methylated region of PEG1/MEST. Journal of Assisted Reproduction and Genetics, 2007, 24, 131-136.	2.5	121
22	Association of four imprinting disorders and ART. Clinical Epigenetics, 2019, 11, 21.	4.1	115
23	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
24	Imprinting of Human GRB10 and Its Mutations in Two Patients with Russell-Silver Syndrome. American Journal of Human Genetics, 2000, 67, 476-482.	6.2	104
25	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
26	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.	3.6	102
27	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
28	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.	3.6	98
29	Heterozygous Orthodenticle Homeobox 2 Mutations Are Associated with Variable Pituitary Phenotype. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 756-764.	3.6	98
30	Backdoor pathway for dihydrotestosterone biosynthesis: Implications for normal and abnormal human sex development. Developmental Dynamics, 2013, 242, 320-329.	1.8	97
31	Temple syndrome: comprehensive molecular and clinical findings in 32 Japanese patients. Genetics in Medicine, 2017, 19, 1356-1366.	2.4	96
32	Kagamiâ€“Ogata syndrome: a clinically recognizable upd(14)pat and related disorder affecting the chromosome 14q32.2 imprinted region. Journal of Human Genetics, 2016, 61, 87-94.	2.3	95
33	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
34	A Track Record on SHOX: From Basic Research to Complex Models and Therapy. Endocrine Reviews, 2016, 37, 417-448.	20.1	87
35	Sex chromosome aberrations and stature: deduction of the principal factors involved in the determination of adult height. Human Genetics, 1993, 91, 551-62.	3.8	85
36	Comprehensive clinical studies in 34 patients with molecularly defined UPD(14)pat and related conditions (Kagamiâ€“Ogata syndrome). European Journal of Human Genetics, 2015, 23, 1488-1498.	2.8	85

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37	Compound Effects of Point Mutations Causing Campomelic Dysplasia/Autosomal Sex Reversal upon SOX9 Structure, Nuclear Transport, DNA Binding, and Transcriptional Activation. Journal of Biological Chemistry, 2001, 276, 27864-27872.	3.4	84
38	The W258X mutation in SLC22A12 is the predominant cause of Japanese renal hypouricemia. Pediatric Nephrology, 2004, 19, 728-733.	1.7	79
39	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
40	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
41	Spectrum of mutations and genotypeâ€“phenotype analysis in Noonan syndrome patients with RIT1 mutations. Human Genetics, 2016, 135, 209-222.	3.8	75
42	Mastermind-like Domain-containing 1 (MAMLD1 or CXorf6) Transactivates the Hes3 Promoter, Augments Testosterone Production, and Contains the SF1 Target Sequence. Journal of Biological Chemistry, 2008, 283, 5525-5532.	3.4	74
43	Prevalence and clinical features of Costello syndrome and cardioâ€facioâ€cutaneous syndrome in Japan: Findings from a nationwide epidemiological survey. American Journal of Medical Genetics, Part A, 2012, 158A, 1083-1094.	1.2	74
44	SHOX Haploinsufficiency as a Cause of Syndromic and Nonsyndromic Short Stature. Molecular Syndromology, 2016, 7, 3-11.	0.8	69
45	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.	3.6	68
46	Transactivation Function of an â¼4800-bp Evolutionarily Conserved Sequence at the SHOX 3â€² Region: Implication for the Downstream Enhancer. American Journal of Human Genetics, 2006, 78, 167-170.	6.2	67
47	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.	2.3	66
48	Segmental and full paternal isodisomy for chromosome 14 in three patients: Narrowing the critical region and implication for the clinical features. American Journal of Medical Genetics, Part A, 2005, 138A, 127-132.	1.2	64
49	Identification and Functional Analysis of Novel Human Growth Hormone Secretagogue Receptor (GHSR) Gene Mutations in Japanese Subjects with Short Stature. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E373-E378.	3.6	64
50	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.	3.6	63
51	Molecular Analysis of 9p Deletions Associated with XY Sex Reversal: Refining the Localization of a Sex-Determining Gene to the Tip of the Chromosome. American Journal of Human Genetics, 1998, 63, 905-908.	6.2	62
52	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.	3.6	60
53	Epimutations of the IG-DMR and the MEG3-DMR at the 14q32.2 imprinted region in two patients with Silverâ€“Russell Syndrome-compatible phenotype. European Journal of Human Genetics, 2015, 23, 1062-1067.	2.8	60
54	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism. Human Genetics, 2018, 137, 95-104.	3.8	60

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55	Haplotype analysis of the estrogen receptor 1 gene in male genital and reproductive abnormalities. Human Reproduction, 2007, 22, 1279-1284.	0.9	59
56	Identification of the mouse paternally expressed imprinted gene Zdbf2 on chromosome 1 and its imprinted human homolog ZDBF2 on chromosome 2. Genomics, 2009, 93, 461-472.	2.9	59
57	Molecular mechanisms regulating phenotypic outcome in paternal and maternal uniparental disomy for chromosome 14. Epigenetics, 2008, 3, 181-187.	2.7	58
58	Genetic evidence for a novel gene(s) involved in urogenital development on 10q26. Kidney International, 2000, 58, 2281-2290.	5.2	57
59	Methylation screening of reciprocal genome-wide UPDs identifies novel human-specific imprinted genesâ€. Human Molecular Genetics, 2011, 20, 3188-3197.	2.9	55
60	Genotypeâ€“phenotype correlation of PAX6 gene mutations in aniridia. Human Genome Variation, 2016, 3, 15052.	0.7	55
61	Safety and efficacy of treatment with asfotase alfa in patients with hypophosphatasia: Results from a Japanese clinical trial. Clinical Endocrinology, 2017, 87, 10-19.	2.4	55
62	Molecular and Clinical Studies in 138 Japanese Patients with Silver-Russell Syndrome. PLoS ONE, 2013, 8, e60105.	2.5	55
63	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.	3.9	54
64	Detecting copy-number variations in whole-exome sequencing data using the eXome Hidden Markov Model: an â€˜exome-firstâ€™ approach. Journal of Human Genetics, 2015, 60, 175-182.	2.3	54
65	Mutation and Gene Copy Number Analyses of Six Pituitary Transcription Factor Genes in 71 Patients with Combined Pituitary Hormone Deficiency: Identification of a Single Patient withLHX4Deletion. Journal of Clinical Endocrinology and Metabolism, 2010, 95, 4043-4047.	3.6	53
66	Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures. Journal of Human Genetics, 2019, 64, 313-322.	2.3	51
67	TBX1 Mutation Identified by Exome Sequencing in a Japanese Family with 22q11.2 Deletion Syndrome-Like Craniofacial Features and Hypocalcemia. PLoS ONE, 2014, 9, e91598.	2.5	49
68	Two patients with MIRAGE syndrome lacking haematological features: role of somatic second-site reversion SAMD9 mutations. Journal of Medical Genetics, 2018, 55, 81-85.	3.2	49
69	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	49
70	Hypogonadotropic hypogonadism in an adult female with a heterozygous hypomorphic mutation of SOX2. European Journal of Endocrinology, 2007, 156, 167-171.	3.7	47
71	Maternal age effect on the development of Praderâ€“Willi syndrome resulting from upd(15)mat through meiosis 1 errors. Journal of Human Genetics, 2011, 56, 566-571.	2.3	47
72	Maternal Uniparental Disomy 14 Syndrome Demonstrates Prader-Willi Syndrome-Like Phenotype. Journal of Pediatrics, 2009, 155, 900-903.e1.	1.8	46

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73	Association of cryptorchidism with Gly146Ala polymorphism in the gene for steroidogenic factor-1. Fertility and Sterility, 2006, 85, 787-790.	1.0	45
74	Kallmann Syndrome Phenotype in a Female Patient with CHARGE Syndrome and CHD7 Mutation. Endocrine Journal, 2006, 53, 741-743.	1.6	45
75	Comprehensive and quantitative multilocus methylation analysis reveals the susceptibility of specific imprinted differentially methylated regions to aberrant methylation in Beckwithâ€“Wiedemann syndrome with epimutations. Genetics in Medicine, 2014, 16, 903-912.	2.4	45
76	Characterization of the aryl hydrocarbon receptor repressor gene and association of its Pro185Ala polymorphism with micropenis. Teratology, 2002, 65, 10-18.	1.6	44
77	Monozygotic female twins discordant for Silverâ€“Russell syndrome and hypomethylation of the H19-DMR. Journal of Human Genetics, 2008, 53, 950-955.	2.3	44
78	Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793.	2.8	44
79	IdenticalNR5A1Missense Mutations in Two Unrelated 46,XX Individuals with Testicular Tissues. Human Mutation, 2017, 38, 39-42.	2.5	44
80	Premature ovarian failure in a female with proximal symphalangism and Noggin mutation. Fertility and Sterility, 2004, 81, 1137-1139.	1.0	43
81	A novel loss-of-function mutation in OTX2 in a patient with anophthalmia and isolated growth hormone deficiency. Human Genetics, 2010, 127, 721-729.	3.8	43
82	Genome-wide multilocus imprinting disturbance analysis in Temple syndrome and Kagami-Ogata syndrome. Genetics in Medicine, 2017, 19, 476-482.	2.4	43
83	Next generation sequencing-based mutation screening of 86 patients with idiopathic short stature. Endocrine Journal, 2017, 64, 947-954.	1.6	41
84	11-oxygenated C19 steroids as circulating androgens in women with polycystic ovary syndrome. Endocrine Journal, 2018, 65, 979-990.	1.6	41
85	Genome-wide copy number analysis and systematic mutation screening in 58 patients with hypogonadotropic hypogonadism. Fertility and Sterility, 2014, 102, 1130-1136.e3.	1.0	40
86	Abnormal urethra formation in mouse models of Split-hand/split-foot malformation type 1 and type 4. European Journal of Human Genetics, 2008, 16, 36-44.	2.8	39
87	Short Stature Homeobox-Containing Gene Duplication on the der(X) Chromosome in a Female with 45,X/46,X, der(X), Gonadal Dysgenesis, and Tall Stature1. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 2927-2930.	3.6	38
88	CAG repeat length of the androgen receptor gene in Japanese males with cryptorchidism. Molecular Human Reproduction, 2000, 6, 973-975.	2.8	38
89	Female Patient Showing Hypohidrotic Ectodermal Dysplasia and Immunodeficiency (HED-ID). American Journal of Human Genetics, 2001, 69, 664-665.	6.2	38
90	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	1.4	38

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91	Testicular volume in Japanese boys up to the age of 15 years. European Journal of Pediatrics, 2000, 159, 843-845.	2.7	37
92	Relative frequency of underlying genetic causes for the development of UPD(14)pat-like phenotype. European Journal of Human Genetics, 2012, 20, 928-932.	2.8	37
93	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
94	Estrogen receptor alpha gene polymorphism is associated with idiopathic azoospermia. Fertility and Sterility, 2002, 78, 1341-1343.	1.0	36
95	X-linked lissencephaly with ambiguous genitalia: Delineation of further case. American Journal of Medical Genetics Part A, 2000, 94, 174-176.	2.4	35
96	Statural Growth in 31 Japanese Patients with SHOX Haploinsufficiency: Support for a Disadvantageous Effect of Gonadal Estrogens. Endocrine Journal, 2004, 51, 197-200.	1.6	35
97	Association of variants in genes involved in environmental chemical metabolism and risk of cryptorchidism and hypospadias. Journal of Human Genetics, 2012, 57, 434-441.	2.3	35
98	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.	2.5	35
99	Genetic screening of Wnt signaling factors in advanced retinopathy of prematurity. Molecular Vision, 2010, 16, 2572-7.	1.1	35
100	Paternal uniparental disomy 14 and related disorders. Epigenetics, 2012, 7, 1142-1150.	2.7	34
101	Growth Hormone and Gonadotropin-Releasing Hormone Analog Therapy in Haploinsufficiency of SHOX.. Endocrine Journal, 2001, 48, 317-322.	1.6	33
102	Novel mutation ofTBX3 in a Japanese family with Ulnar-Mammary syndrome: Implication for impaired sex development. American Journal of Medical Genetics Part A, 2002, 110, 365-369.	2.4	33
103	Skeletal Deformity Associated with <i>SHOX</i> Deficiency. Clinical Pediatric Endocrinology, 2014, 23, 65-72.	0.8	33
104	Epimutation (hypomethylation) affecting the chromosome 14q32.2 imprinted region in a girl with upd(14)mat-like phenotype. European Journal of Human Genetics, 2008, 16, 1019-1023.	2.8	32
105	<i>MAMLD1 (CXorf6):</i> A New Gene Involved in Hypospadias. Hormone Research in Paediatrics, 2009, 71, 245-252.	1.8	32
106	PRKAR1AMutation Affecting cAMP-Mediated G Protein-Coupled Receptor Signaling in a Patient with Acrodysostosis and Hormone Resistance. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1808-E1813.	3.6	32
107	Maternal Uniparental Disomy for Chromosome 20: Physical and Endocrinological Characteristics of Five Patients. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2083-2088.	3.6	32
108	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform. Epigenomics, 2018, 10, 941-954.	2.1	31

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109	SHOX: pseudoautosomalhomeobox containing gene for short stature and dyschondrosteosis. Growth Hormone and IGF Research, 1999, 9, 53-58.	1.1	30
110	Microdeletion in theSHOX 3â€² region associated with skeletal phenotypes of Langer mesomelic dysplasia in a 45,X/46,X,r(X) infant and Leri-Weill dyschondrosteosis in her 46,XX mother: Implication for theSHOX enhancer. American Journal of Medical Genetics, Part A, 2005, 137A, 72-76.	1.2	30
111	Nomenclature for alleles of the cytochrome P450 oxidoreductase gene. Pharmacogenetics and Genomics, 2009, 19, 565-566.	1.5	30
112	Aromatase Excess Syndrome: Identification of Cryptic Duplications and Deletions Leading to Gain of Function ofCYP19A1and Assessment of Phenotypic Determinants. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1035-E1043.	3.6	30
113	Cytochrome <scp>P450</scp> oxidoreductase deficiency: Rare congenital disorder leading to skeletal malformations and steroidogenic defects. Pediatrics International, 2014, 56, 805-808.	0.5	30
114	MAMLD1 and 46,XY Disorders of Sex Development. Seminars in Reproductive Medicine, 2012, 30, 410-416.	1.1	29
115	Human glutathione Sâ€transferase A (GSTA) family genes are regulated by steroidogenic factor 1 (SFâ€1) and are involved in steroidogenesis. FASEB Journal, 2013, 27, 3198-3208.	0.5	29
116	Contribution of gene mutations to Silver-Russell syndrome phenotype: multigene sequencing analysis in 92 etiology-unknown patients. Clinical Epigenetics, 2020, 12, 86.	4.1	29
117	Mamld1 Knockdown Reduces Testosterone Production and Cyp17a1 Expression in Mouse Leydig Tumor Cells. PLoS ONE, 2011, 6, e19123.	2.5	28
118	De novoIGF2mutation on the paternal allele in a patient with Silverâ€“Russell syndrome and ectrodactyly. Human Mutation, 2017, 38, 953-958.	2.5	28
119	Kallmann Syndrome: Somatic and Germline Mutations of the Fibroblast Growth Factor Receptor 1 Gene in a Mother and the Son. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 1415-1418.	3.6	27
120	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.	3.2	27
121	PTPN11 mutations and genotype-phenotype correlations in Noonan and LEOPARD syndromes. Pediatric Endocrinology Reviews, 2005, 2, 669-74.	1.2	27
122	HLA-class II and class I genotypes among Japanese children with Type 1A diabetes and their families. Pediatric Diabetes, 2012, 13, 33-44.	2.9	26
123	IGF2 Mutations. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 116-125.	3.6	26
124	Association of Severe Micropenis with Gly146Ala Polymorphism in the Gene for Steroidogenic Factor-1. Endocrine Journal, 2005, 52, 445-448.	1.6	25
125	Number of CGG repeats in the FMR1 gene of Japanese patients with primary ovarian insufficiency. Fertility and Sterility, 2011, 96, 1170-1174.	1.0	25
126	Androgenetic/biparental mosaicism in a girl with Beckwithâ€“Wiedemann syndrome-like and upd(14)pat-like phenotypes. Journal of Human Genetics, 2011, 56, 91-93.	2.3	25

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127	Mamld1 Deficiency Significantly Reduces mRNA Expression Levels of Multiple Genes Expressed in Mouse Fetal Leydig Cells but Permits Normal Genital and Reproductive Development. Endocrinology, 2012, 153, 6033-6040.	2.8	25
128	Complex Genomic Rearrangement Within theGNASRegion Associated With Familial Pseudohypoparathyroidism Type 1b. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2623-2627.	3.6	25
129	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
130	Efficacy and safety of octreotide for the treatment of congenital hyperinsulinism: a prospective, open-label clinical trial and an observational study in Japan using a nationwide registry. Endocrine Journal, 2017, 64, 867-880.	1.6	25
131	MYRF haploinsufficiency causes 46,XY and 46,XX disorders of sex development: bioinformatics consideration. Human Molecular Genetics, 2019, 28, 2319-2329.	2.9	25
132	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
133	Micropenis and the AR Gene: Mutation and CAG Repeat-Length Analysis. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5372-5378.	3.6	24
134	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.	0.9	24
135	Determination of biotin (vitamin H) by the high-performance affinity chromatography with a trypsin-treated avidin-bound column. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2008, 869, 93-100.	2.3	24
136	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
137	Identification ofINSandKCNJ11gene mutations in type 1B diabetes in Japanese children with onset of diabetes before 5 yr ofÂage. Pediatric Diabetes, 2013, 14, 112-120.	2.9	24
138	Understanding the pathological manifestations of aromatase excess syndrome: lessons for clinical diagnosis. Expert Review of Endocrinology and Metabolism, 2014, 9, 397-409.	2.4	24
139	Impact of a novel homozygous mutation in nicotinamide nucleotide transhydrogenase on mitochondrial DNA integrity in a case of familial glucocorticoid deficiency. BBA Clinical, 2015, 3, 70-78.	4.1	24
140	Loss-of-Function SOX10 Mutation in a Patient with Kallmann Syndrome, Hearing Loss, and Iris Hypopigmentation. Hormone Research in Paediatrics, 2015, 84, 212-216.	1.8	24
141	Paradoxical gainâ€ofâ€function mutant of the Gâ€protein–coupled receptor <scp>PROKR</scp>2 promotes early puberty. Journal of Cellular and Molecular Medicine, 2017, 21, 2623-2626.	3.6	24
142	Association of Micropenis with Pro185Ala Polymorphism of the Gene for Aryl Hydrocarbon Receptor Repressor Involved in Dioxin Signaling. Endocrine Journal, 2005, 52, 83-88.	1.6	23
143	Hepatoblastoma in a Noonan syndrome patient with a PTPN11 mutation. Pediatric Blood and Cancer, 2008, 50, 1274-1276.	1.5	23
144	Genomic Basis of Aromatase Excess Syndrome: Recombination- and Replication-Mediated Rearrangements Leading to CYP19A1 Overexpression. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E2013-E2021.	3.6	23

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145	Retinoic acid catabolizing enzyme CYP 26C1 is a genetic modifier in SHOX deficiency. EMBO Molecular Medicine, 2016, 8, 1455-1469.	6.9	23
146	Molecular and clinical analyses of two patients with UPD(16)mat detected by screening 94 patients with Silver-Russell syndrome phenotype of unknown aetiology. Journal of Medical Genetics, 2019, 56, 413-418.	3.2	23
147	Expanding the Spectrum of Founder Mutations Causing Isolated Gonadotropin-Releasing Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1378-E1385.	3.6	22
148	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
149	Screening for mutations of the androgen receptor gene in patients with isolated cryptorchidism. Fertility and Sterility, 2001, 76, 834-836.	1.0	21
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