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	Congenital disorders of estrogen biosynthesis and action. Best Practice and Research in Clinical Endocrinology and Metabolism, 2022, 36, 101580.	4.7	9
2	Retrotransposition disrupting EBP in a girl and her mother with X-linked dominant chondrodysplasia punctata. Journal of Human Genetics, 2022, , .	2.3	2
3	A case of atypical congenital cytomegalovirus infection with intraventricular hemorrhage. Pediatrics International, 2022, 64, e14906.	0.5	0
4	A novel intronic <scp>PORCN</scp> variant creating an alternative splice acceptor site in a mother and her daughter with focal dermal hypoplasia. American Journal of Medical Genetics, Part A, 2022, 188, 1612-1617.	1.2	1
5	Genome sequencing and RNA sequencing of urinary cells reveal an intronic FBN1 variant causing aberrant splicing. Journal of Human Genetics, 2022, 67, 387-392.	2.3	7
6	Retinitis pigmentosa with optic neuropathy and COQ2 mutations: A case report. American Journal of Ophthalmology Case Reports, 2022, 25, 101298.	0.7	1
7	Intrauterine Hyponutrition Reduces Fetal Testosterone Production and Postnatal Sperm Count in the Mouse. Journal of the Endocrine Society, 2022, 6, bvac022.	0.2	1
8	ACAN biallelic variants in a girl with severe idiopathic short stature. Journal of Human Genetics, 2022, 67, 481-486.	2.3	1
9	Six yearsâ€™ accomplishment of the Initiative on Rare and Undiagnosed Diseases: nationwide project in Japan to discover causes, mechanisms, and cures. Journal of Human Genetics, 2022, 67, 505-513.	2.3	17
10	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	4.1	14
11	<scp>SHOX</scp> farâ€downstream deletion in a patient with nonsyndromic short stature. American Journal of Medical Genetics, Part A, 2022, 188, 2173-2177.	1.2	1
12	Maternal Uniparental Isodisomy of Chromosome 4 and 8 in Patients with Retinal Dystrophy: SRD5A3-Congenital Disorders of Glycosylation and RP1-Related Retinitis Pigmentosa. Genes, 2022, 13, 359.	2.4	4
13	Pathogenic Copy Number and Sequence Variants in Children Born SGA With Short Stature Without Imprinting Disorders. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e3121-e3133.	3.6	7
14	Loss of imprinting of the human-specific imprinted gene ZNF597 causes prenatal growth retardation and dysmorphic features: implications for phenotypic overlap with Silver-Russell syndrome. Journal of Medical Genetics, 2021, 58, 427-432.	3.2	6
15	Role of Imprinting Disorders in Short Children Born SGA and Silver-Russell Syndrome Spectrum. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 802-813.	3.6	16
16	Kagamiâ€“Ogata syndrome in a patient with 46,XX,t(2;14)(q11.2;q32.2)mat disrupting MEG3. Journal of Human Genetics, 2021, 66, 439-443.	2.3	1
17	Insulin resistant diabetes mellitus in SHORT syndrome: case report and literature review. Endocrine Journal, 2021, 68, 111-117.	1.6	7
18	Primary ovarian insufficiency in a female with phosphomannomutase-2 gene (PMM2) mutations for congenital disorder of glycosylation. Endocrine Journal, 2021, 68, 605-611.	1.6	4

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19	Long-term Effect of Aromatase Inhibition in Aromatase Excess Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 1491-1500.	3.6	1
20	NDNF variants are rare in patients with congenital hypogonadotropic hypogonadism. Human Genome Variation, 2021, 8, 5.	0.7	2
21	Biallelic CDK9 variants as a cause of a new multiple-malformation syndrome with retinal dystrophy mimicking the CHARGE syndrome. Journal of Human Genetics, 2021, 66, 1021-1027.	2.3	3
22	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. Frontiers in Cell and Developmental Biology, 2021, 9, 631428.	3.7	4
23	SOX10 Mutation Screening for 117 Patients with Kallmann Syndrome. Journal of the Endocrine Society, 2021, 5, bvab056.	0.2	3
24	Genetic and phenotypic analysis of 101 patients with developmental delay or intellectual disability using wholeâ€exome sequencing. Clinical Genetics, 2021, 100, 40-50.	2.0	17
25	Parthenogenetic mosaicism: generation via second polar body retention and unmasking of a likely causative PER2 variant for hypersomnia. Clinical Epigenetics, 2021, 13, 73.	4.1	4
26	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
27	ZNF445: a homozygous truncating variant in a patient with Temple syndrome and multilocus imprinting disturbance. Clinical Epigenetics, 2021, 13, 119.	4.1	9
28	Long-Term Effect of Aromatase Inhibition in Aromatase Excess Syndrome. Journal of the Endocrine Society, 2021, 5, A679-A679.	0.2	0
29	Global developmental delay, systemic dysmorphism and epilepsy in a patient with a de novo U2AF2 variant. Journal of Human Genetics, 2021, 66, 1185-1187.	2.3	7
30	Treatment approaches for congenital transverse limb deficiency: Data analysis from an epidemiological national survey in Japan. Journal of Orthopaedic Science, 2021, 26, 650-654.	1.1	6
31	Novel ALG12 variants and hydronephrosis in siblings with impaired N-glycosylation. Brain and Development, 2021, 43, 945-951.	1.1	1
32	IGF2 Mutations. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 116-125.	3.6	26
33	Exome reports A de novo GNB2 variant associated with global developmental delay, intellectual disability, and dysmorphic features. European Journal of Medical Genetics, 2020, 63, 103804.	1.3	15
34	De novo ZBTB7A variant in a patient with macrocephaly, intellectual disability, and sleep apnea: implications for the phenotypic development in 19p13.3 microdeletions. Journal of Human Genetics, 2020, 65, 181-186.	2.3	9
35	Erythrokeratodermia variabilis et progressiva with a rare GJB3 mutation. Journal of Dermatology, 2020, 47, e111-e113.	1.2	1
36	Random X chromosome inactivation in patients with Klinefelter syndrome. Molecular and Cellular Pediatrics, 2020, 7, 1.	1.8	10

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37	Assisted reproductive technology represents a possible risk factor for development of epimutation-mediated imprinting disorders for mothers aged â‰¥ 30â€‰years. Clinical Epigenetics, 2020, 12, 111.	4.1	11
38	Human Spermatogenesis Tolerates Massive Size Reduction of the Pseudoautosomal Region. Genome Biology and Evolution, 2020, 12, 1961-1964.	2.5	1
39	Genome-wide methylation analysis in Silverâ€“Russell syndrome, Temple syndrome, and Praderâ€“Willi syndrome. Clinical Epigenetics, 2020, 12, 159.	4.1	7
40	Nonsense-associated altered splicing of MAP3K1 in two siblings with 46,XY disorders of sex development. Scientific Reports, 2020, 10, 17375.	3.3	4
41	Case Report: Efficacy of Reduced Doses of Asfotase Alfa Replacement Therapy in an Infant With Hypophosphatasia Who Lacked Severe Clinical Symptoms. Frontiers in Endocrinology, 2020, 11, 590455.	3.5	1
42	TSC1 intragenic deletion transmitted from a mosaic father to two siblings with cardiac rhabdomyomas: Identification of two aberrant transcripts. European Journal of Medical Genetics, 2020, 63, 104060.	1.3	2
43	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
44	Coffinâ€Lowry syndrome in a girl with 46,XX,t(X;11)(p22;p15)dn: Identification of RPS6KA3 disruption by whole genome sequencing. Clinical Case Reports (discontinued), 2020, 8, 1076-1080.	0.5	4
45	Rare variant of the epigenetic regulator SMCHD1 in a patient with pituitary hormone deficiency. Scientific Reports, 2020, 10, 10985.	3.3	12
46	POLR3A variants in striatal involvement without diffuse hypomyelination. Brain and Development, 2020, 42, 363-368.	1.1	15
47	A de novo TOP2B variant associated with global developmental delay and autism spectrum disorder. Molecular Genetics & Genomic Medicine, 2020, 8, e1145.	1.2	10
48	Longâ€term observation of a Japanese mucolipidosis <scp>IV</scp> patient with a novel homozygous p.F313del variant of <scp>MCOLN1</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 1500-1505.	1.2	6
49	Long-term efficacy and safety of two doses of Norditropin^{Â®} (somatropin) in Noonan syndrome: a 4-year randomized, double-blind, multicenter trial in Japanese patients. Endocrine Journal, 2020, 67, 803-818.	1.6	16
50	Identification and functional characterization of a novel PAX8 mutation (p.His39Pro) causing familial thyroid hypoplasia. Clinical Pediatric Endocrinology, 2020, 29, 173-178.	0.8	5
51	De novo AFF3 variant in a patient with mesomelic dysplasia with foot malformation. Journal of Human Genetics, 2019, 64, 1041-1044.	2.3	6
52	Unbalanced Y;7 Translocation between Two Low-Similarity Sequences Leading to SRY-Positive 45,X Testicular Disorders of Sex Development. Cytogenetic and Genome Research, 2019, 158, 115-120.	1.1	2
53	Comprehensive clinical and molecular studies in split-hand/foot malformation: identification of two plausible candidate genes (LRP6 and UBA2). European Journal of Human Genetics, 2019, 27, 1845-1857.	2.8	11
54	(Epi)genetic defects of MKRN3 are rare in Asian patients with central precocious puberty. Human Genome Variation, 2019, 6, 7.	0.7	8

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55	KLF11 variant in a family clinically diagnosed with early childhoodâ€onset type 1B diabetes. Pediatric Diabetes, 2019, 20, 712-719.	2.9	18
56	Temple syndrome in a patient with variably methylated CpGs at the primary MEG3/DLK1:IG-DMR and severely hypomethylated CpGs at the secondary MEG3:TSS-DMR. Clinical Epigenetics, 2019, 11, 42.	4.1	11
57	Exploring the unique function of imprinting control centers in the PWS/AS-responsible region: finding from array-based methylation analysis in cases with variously sized microdeletions. Clinical Epigenetics, 2019, 11, 36.	4.1	7
58	Germline-Derived Gain-of-Function Variants of GsÎ±-Coding GNAS Gene Identified in Nephrogenic Syndrome of Inappropriate Antidiuresis. Journal of the American Society of Nephrology: JASN, 2019, 30, 877-889.	6.1	21
59	MYRF haploinsufficiency causes 46,XY and 46,XX disorders of sex development: bioinformatics consideration. Human Molecular Genetics, 2019, 28, 2319-2329.	2.9	25
60	Coexistence of a CAV3 mutation and a DMD deletion in a family with complex muscular diseases. Brain and Development, 2019, 41, 474-479.	1.1	6
61	Association of four imprinting disorders and ART. Clinical Epigenetics, 2019, 11, 21.	4.1	115
62	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
63	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
64	A case of paternal uniparental isodisomy for chromosome 7 associated with overgrowth. Journal of Medical Genetics, 2018, 55, 567-570.	3.2	13
65	Clinical characteristics of a Japanese patient with Bardet-Biedl syndrome caused by BBS10 mutations. Japanese Journal of Ophthalmology, 2018, 62, 458-466.	1.9	10
66	Mosaic upd(14)pat in a patient with mild features of Kagamiâ€“Ogata syndrome. Clinical Case Reports (discontinued), 2018, 6, 91-95.	0.5	12
67	Partial androgen insensitivity syndrome caused by a deep intronic mutation creating an alternative splice acceptor site of the AR gene. Scientific Reports, 2018, 8, 2287.	3.3	14
68	De novo variants in SETD1B are associated with intellectual disability, epilepsy and autism. Human Genetics, 2018, 137, 95-104.	3.8	60
69	(Epi)genotype-Phenotype Analysis in 69 Japanese Patients With Pseudohypoparathyroidism Type I. Journal of the Endocrine Society, 2018, 2, 9-23.	0.2	14
70	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.	2.5	6
71	Functional missense and splicing variants in the retinoic acid catabolizing enzyme CYP26C1 in idiopathic short stature. European Journal of Human Genetics, 2018, 26, 1113-1120.	2.8	10
72	STX2 is a causative gene for nonobstructive azoospermia. Human Mutation, 2018, 39, 830-833.	2.5	17

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73	Comprehensive screening for monogenic diabetes in 89 Japanese children with insulin-requiring antibody-negative type 1 diabetes. Pediatric Diabetes, 2018, 19, 243-250.	2.9	10
74	Gainâ€ofâ€function mutations in Gâ€proteinâ€“coupled receptor genes associated with human endocrine disorders. Clinical Endocrinology, 2018, 88, 351-359.	2.4	19
75	FGFR1 disruption identified by whole genome sequencing in a male with a complex chromosomal rearrangement and hypogonadotropic hypogonadism. American Journal of Medical Genetics, Part A, 2018, 176, 139-143.	1.2	1
76	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
77	Expression of Xenobiotic Biomarkers CYP1 Family in Preputial Tissue of Patients with Hypospadias and Phimosis and Its Association with DNA Methylation Level of SRD5A2 Minimal Promoter. Archives of Environmental Contamination and Toxicology, 2018, 74, 240-247.	4.1	5
78	GATA4 variant identified by wholeâ€exome sequencing in a Japanese family with atrial septal defect: Implications for male sex development. Clinical Case Reports (discontinued), 2018, 6, 2229-2233.	0.5	2
79	Congenital limb deficiency in Japan: a cross-sectional nationwide survey on its epidemiology. BMC Musculoskeletal Disorders, 2018, 19, 262.	1.9	19
80	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform. Epigenomics, 2018, 10, 941-954.	2.1	31
81	Efficacy and safety of two doses of Norditropin^{Â®} (somatropin) in short stature due to Noonan syndrome: a 2-year randomized, double-blind, multicenter trial in Japanese patients. Endocrine Journal, 2018, 65, 159-174.	1.6	17
82	11-oxygenated C19 steroids as circulating androgens in women with polycystic ovary syndrome. Endocrine Journal, 2018, 65, 979-990.	1.6	41
83	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
84	GATA4 mutations are uncommon in patients with 46,XY disorders of sex development without heart anomaly. Asian Journal of Andrology, 2018, 20, 629.	1.6	4
85	FGFR1 Analyses in Four Patients with Hypogonadotropic Hypogonadism with Split-Hand/Foot Malformation: Implications for the Promoter Region. Human Mutation, 2017, 38, 503-506.	2.5	6
86	De novoIGF2mutation on the paternal allele in a patient with Silverâ€“Russell syndrome and ectrodactyly. Human Mutation, 2017, 38, 953-958.	2.5	28
87	Nucleotide substitutions in <scp>CD</scp>101, the human homolog of a diabetes susceptibility gene in nonâ€obese diabetic mouse, in patients with type 1 diabetes. Journal of Diabetes Investigation, 2017, 8, 286-294.	2.4	4
88	Paradoxical gainâ€ofâ€function mutant of the Gâ€proteinâ€coupled receptor <scp>PROKR</scp>2 promotes early puberty. Journal of Cellular and Molecular Medicine, 2017, 21, 2623-2626.	3.6	24
89	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
90	Nomenclature of primary amenorrhea: A proposal document of the Japan Society of Obstetrics and Gynecology committee for the redefinition of primary amenorrhea. Journal of Obstetrics and Gynaecology Research, 2017, 43, 1738-1742.	1.3	2

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91	X-linked hypomyelination with spondylometaphyseal dysplasia (H-SMD) associated with mutations in AIFM1. Neurogenetics, 2017, 18, 185-194.	1.4	38
92	Temple syndrome: comprehensive molecular and clinical findings in 32 Japanese patients. Genetics in Medicine, 2017, 19, 1356-1366.	2.4	96
93	Genetic heterogeneity of patients with suspected Silver-Russell syndrome: genome-wide copy number analysis in 82 patients without imprinting defects. Clinical Epigenetics, 2017, 9, 52.	4.1	15
94	IdenticalNR5A1Missense Mutations in Two Unrelated 46,XX Individuals with Testicular Tissues. Human Mutation, 2017, 38, 39-42.	2.5	44
95	Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124.	9.6	336
96	Mutation analysis of FGFR1â€3 in 11 Japanese patients with syndromic craniosynostoses. American Journal of Medical Genetics, Part A, 2017, 173, 157-162.	1.2	7
97	Genome-wide multilocus imprinting disturbance analysis in Temple syndrome and Kagami-Ogata syndrome. Genetics in Medicine, 2017, 19, 476-482.	2.4	43
98	Phenotypic Variation in 46,XX Disorders of Sex Development due to the <i>NR5A1 </i>p.R92W Variant: A Sibling Case Report and Literature Review. Sexual Development, 2017, 11, 284-288.	2.0	9
99	A de novo 50-bp <i>GNAS</i> Intragenic Duplication in a Patient with Pseudohypoparathyroidism Type 1a. Cytogenetic and Genome Research, 2017, 153, 125-130.	1.1	1
100	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.	0.8	4
101	Knockout of Murine Mamld1 Impairs Testicular Growth and Daily Sperm Production but Permits Normal Postnatal Androgen Production and Fertility. International Journal of Molecular Sciences, 2017, 18, 1300.	4.1	13
102	Individual Clinically Diagnosed with CHARGE Syndrome but with a Mutation in KMT2D, a Gene Associated with Kabuki Syndrome: A Case Report. Frontiers in Genetics, 2017, 8, 210.	2.3	18
103	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
104	Childbirth and fertility preservation in childhood and adolescent cancer patients: a second national survey of Japanese pediatric endocrinologists. Clinical Pediatric Endocrinology, 2017, 26, 81-88.	0.8	5
105	Next generation sequencing-based mutation screening of 86 patients with idiopathic short stature. Endocrine Journal, 2017, 64, 947-954.	1.6	41
106	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.	0.8	11
107	Gonadal function, fertility, and reproductive medicine in childhood and adolescent cancer patients: a national survey of Japanese pediatric endocrinologists. Clinical Pediatric Endocrinology, 2016, 25, 45-57.	0.8	13
108	Complex Genomic Rearrangement Within theGNASRegion Associated With Familial Pseudohypoparathyroidism Type 1b. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2623-2627.	3.6	25

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109	Blood allopregnanolone levels in women with polycystic ovary syndrome. Clinical Endocrinology, 2016, 85, 151-152.	2.4	2
110	Complex X-Chromosomal Rearrangements in Two Women with Ovarian Dysfunction: Implications of Chromothripsis/Chromoanasynthesis-Dependent and -Independent Origins of Complex Genomic Alterations. Cytogenetic and Genome Research, 2016, 150, 86-92.	1.1	19
111	The p.R92W variant of NR5A1/Nr5a1 induces testicular development of 46,XX gonads in humans, but not in mice: phenotypic comparison of human patients and mutation-induced mice. Biology of Sex Differences, 2016, 7, 56.	4.1	19
112	Genotypeâ€“phenotype correlation of PAX6 gene mutations in aniridia. Human Genome Variation, 2016, 3, 15052.	0.7	55
113	Copy Number Variations of the Azoospermia Factor Region and SRY Are Not Associated with the Risk of Hypospadias. Sexual Development, 2016, 10, 12-15.	2.0	2
114	SHOX Haploinsufficiency as a Cause of Syndromic and Nonsyndromic Short Stature. Molecular Syndromology, 2016, 7, 3-11.	0.8	69
115	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
116	Beckwithâ€“Wiedemann syndrome and pseudohypoparathyroidism type Ib in a patient with multilocus imprinting disturbance: a female-dominant phenomenon?. Journal of Human Genetics, 2016, 61, 765-769.	2.3	21
117	Silverâ€“Russell syndrome in a patient with somatic mosaicism for upd(11)mat identified by buccal cell analysis. American Journal of Medical Genetics, Part A, 2016, 170, 1938-1941.	1.2	20
118	NR0B1 Frameshift Mutation in a Boy with Idiopathic Central Precocious Puberty. Sexual Development, 2016, 10, 205-209.	2.0	8
119	Long-term clinical course in three patients with MAMLD1 mutations. Endocrine Journal, 2016, 63, 835-839.	1.6	6
120	Identification of monogenic gene mutations in Japanese subjects diagnosed with type 1B diabetes between >5 and 15.1 years of age. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 1047-54.	0.9	5
121	A Chronic Graft-versus-host Disease Case after Improvement of Basedow's Disease developed after Allogeneic Bone Marrow Transplantation. Journal of Hematopoietic Cell Transplantation, 2016, 5, 13-17.	0.1	0
122	Retinoic acid catabolizing enzyme CYP 26C1 is a genetic modifier in SHOX deficiency. EMBO Molecular Medicine, 2016, 8, 1455-1469.	6.9	23
123	Novel HPS6 mutations identified by whole-exome sequencing in two Japanese sisters with suspected ocular albinism. Journal of Human Genetics, 2016, 61, 839-842.	2.3	11
124	A Track Record on SHOX: From Basic Research to Complex Models and Therapy. Endocrine Reviews, 2016, 37, 417-448.	20.1	87
125	Risk assessment of medically assisted reproduction and advanced maternal ages in the development of Praderâ€“Willi syndrome due to UPD(15)mat. Clinical Genetics, 2016, 89, 614-619.	2.0	8
126	Spectrum of mutations and genotypeâ€“phenotype analysis in Noonan syndrome patients with RIT1 mutations. Human Genetics, 2016, 135, 209-222.	3.8	75

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127	Growth references for Japanese individuals with Noonan syndrome. Pediatric Research, 2016, 79, 543-548.	2.3	16
128	Combined steroidogenic characters of fetal adrenal and Leydig cells in childhood adrenocortical carcinoma. Journal of Steroid Biochemistry and Molecular Biology, 2016, 159, 86-93.	2.5	12
129	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
130	Novel RAB3GAP1 compound heterozygous mutations in Japanese siblings with Warburg Micro syndrome. Brain and Development, 2016, 38, 337-340.	1.1	13
131	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
132	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
133	Chromosome 6q24 methylation defects are uncommon in childhood-onset non-autoimmune diabetes mellitus patients born appropriate- or large-for-gestational age. Clinical Pediatric Endocrinology, 2016, 25, 99-102.	0.8	3
134	Testicular dysgenesis/regression without campomelic dysplasia in patients carrying missense mutations and upstream deletion of SOX 9. Molecular Genetics & Genomic Medicine, 2015, 3, 550-557.	1.2	19
135	Growth hormone deficiency in monozygotic twins with autosomal dominant pseudohypoparathyroidism type Ib. Endocrine Journal, 2015, 62, 523-529.	1.6	14
136	Parturition failure in mice lacking Mamld1. Scientific Reports, 2015, 5, 14705.	3.3	13
137	Endocrinopathies in a boy with cryptic copy-number variations on 4q, 7q and Xp. Human Genome Variation, 2015, 2, 15020.	0.7	0
138	Development of waist circumference percentiles for Japanese children and an examination of their screening utility for childhood metabolic syndrome: a population-based cross-sectional study. BMC Public Health, 2015, 15, 1121.	2.9	10
139	Exploration of hydroxymethylation in Kagami-Ogata syndrome caused by hypermethylation of imprinting control regions. Clinical Epigenetics, 2015, 7, 90.	4.1	10
140	Femoralâ€tibialâ€digital malformations in a boy with the Japanese founder triplication of BHLHA9. American Journal of Medical Genetics, Part A, 2015, 167, 3226-3228.	1.2	4
141	A 45,X/46,XY DSD (Disorder of Sexual Development) case with an extremely uneven distribution of 46,XY cells between lymphocytes and gonads. Clinical Pediatric Endocrinology, 2015, 24, 11-14.	0.8	6
142	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
143	SOX3 Overdosage Permits Normal Sex Development in Females with Random X Inactivation. Sexual Development, 2015, 9, 125-129.	2.0	6
144	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

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145	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
146	Copy-number variations in Y-chromosomal azoospermia factor regions identified by multiplex ligation-dependent probe amplification. Journal of Human Genetics, 2015, 60, 127-131.	2.3	18
147	Hypogonadotropic hypogonadism in a female patient previously diagnosed as having waardenburg syndrome due to a sox10 mutation. Endocrine, 2015, 49, 553-556.	2.3	16
148	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
149	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
150	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
151	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
152	Novel Splice Site Mutation in MAMLD1 in a Patient with Hypospadias. Sexual Development, 2015, 9, 130-135.	2.0	14
153	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.	1.8	11
154	Hemodynamic assessment in a child with renovascular hypertension using timeâ€resolved threeâ€dimensional cine phaseâ€contrast MRI. Journal of Magnetic Resonance Imaging, 2015, 41, 165-168.	3.4	9
155	Silverâ€“Russell syndrome without body asymmetry in three patients with duplications of maternally derived chromosome 11p15 involving CDKN1C. Journal of Human Genetics, 2015, 60, 91-95.	2.3	21
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157	Skeletal Deformity Associated with <i>SHOX</i> Deficiency. Clinical Pediatric Endocrinology, 2014, 23, 65-72.	0.8	33
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