

研究成果の刊行に関する一覧表

書籍

著者氏名	論文タイトル名	書籍全体の編集者名	書籍名	出版社名	出版地	出版年	ページ
緒方勤	こどもの病気 遺伝について聞かれたら	松原洋一、呉繁夫、左合治彦(編)	性分化疾患	診断と治療社		2015	88-89
中島信一、 <u>緒方勤</u>	アンドロゲン不応症(精巢性女性化症)	吉川史隆、倉智智久、平松祐司(編)	産婦人科疾患 最新の治療	南江堂		2016	347-348
堀川玲子	思春期早発症	五十嵐隆	小児科診療ガイドライン第3版	総合医学社	東京	2016	442-446

雑誌

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Nomura R, Miyai K, Okada M, Kajiwara M, Ono M, <u>Ogata T</u> , et al.	A 45,X/46,XY DSD (Disorder of Sexual Development) case with an extremely uneven distribution of 46,XY cells between lymphocytes and gonads.	<i>Clin Pediatr Endocrinol</i>	24 (1)	11-14	2015
Kagami M, Mizuno S, Matsubara K, Nakabayashi K, Sano S, Fuke T, Fukami M, <u>Ogata T</u> *	Epimutations of the IG-DMR and the MEG3-DMR at the 14q32.2 imprinted region in two patients with Silver-Russell syndrome-compatible phenotype.	<i>Eur J Hum Genet</i>	23 (11)	1488-1498	2015
Nakashima S, Kato F, Kosho T, Nagasaki K, Kikuchi T, Kagami M, Fukami M, <u>Ogata T</u> *	Silver-Russell syndrome without body asymmetry in three patients with duplications of maternally derived chromosome 11p15 involving CDKN1C.	<i>J Hum Genet</i>	60 (2)	91-95,	2015
Miyatake S, Koshimizu E, Fujita A, Fukai R, Imagawa E, Ohba C, Kuki I, Makita Y, <u>Ogata T</u> , et al.	Detecting copy number variations in whole exome sequencing data using exome hidden markov model - an expectation of "exome-first" approach.	<i>J Hum Genet</i>	60 (4)	175-182	2015

Kagami M, Kurosawa K, Miyazaki O, Ishino F, Matsuoka K, <u>Ogata T*</u>	Comprehensive clinical studies in 34 patients with molecularly defined UPD(14)pat and related conditions (Kagami-Ogata syndrome).	<i>Eur J Hum Genet</i>	23 (11)	1488–1498	2015
Fujisawa Y, Napoli E, Wong S, Song G, Yamaguchi R, Matsui T, Nagasaki K, <u>Ogata T</u> , Giulivi C	Impact of a novel homozygous mutation in nicotinamide nucleotide transhydrogenase on mitochondrial DNA integrity in a case of familial glucocorticoid deficiency.	<i>BBA Clinical</i>	1 (3)	70–78	2015
Sano S, Iwata H, Matsubara K, Fukami M, Kagami M, <u>Ogata T</u>	Growth hormone deficiency in monozygotic twins with autosomal dominant pseudohypoparathyroidism type Ib.	<i>Endocr J</i>	62 (6)	523–529	2015
Choi JH, Balasubramanian R, Lee PH, Shaw ND, Hall JE, Plummer L, Buck CL, Kottler ML, Jarzabek K, Wołczynski S, Quinton R, Latronico AC, Dode C, <u>Ogata T</u> , et al.	Expanding the Spectrum of Founder Mutations Causing Isolated Gonadotropin-Releasing Hormone Deficiency.	<i>J Clin Endocrinol Metab</i>	100 (10)	E1378–1385	2015
Nagata E, Haga N, Ohtaka K, Fujisawa Y, Fukami M, Nishimura G, <u>Ogata T*</u>	Femoral-Tibial-Digital Malformations in a Boy with the Japanese Founder Triplication of <i>BHLHA9</i> .	<i>Am J Med Genet A</i>	167 (12)	3226–3228	2015
Ishii T, Matsuo N, Amano N, Hori N, Inokuchi M, Sasaki G, Kamimaki T, Anzo M, Tamai S, <u>Ogata T</u> , et al.	Human chorionic gonadotropin stimulation test in prepubertal children with micropenis can accurately predict Leydig cell function in pubertal or postpubertal adolescents.	<i>Horm Res Paediatr</i>	84 (5)	305–310	2015
Matsubara K, Kagami M, Nakabayashi K, Hata K, Fukami M, <u>Ogata T</u> , Yamazawa K*	Exploration of hydroxymethylation in Kagami-Ogata syndrome caused by hypermethylation of imprinting control regions.	<i>Clin Epigenet</i>	7(1)	90	2015
Matsubara K, Murakami N, Fukami M, Kagami M, Nagai T, <u>Ogata T*</u>	Risk assessment of medically assisted reproduction and advanced maternal ages in the development of Prader-Willi syndrome due to UPD(15)mat.	<i>Clin Genet</i>	89 (5)	614–619	2016

Matsushita R, Isojima T, Takaya R, Satake E, Yamaguchi R, Kitsuda K, Nagata E, Sano S, Nakanishi T, Nakagawa Y, Ohzeki T, <u>Ogata T</u> , Fujisawa Y*	Development of waist circumference percentiles for Japanese children and an examination of their screening utility for childhood metabolic syndrome.	<i>BMC Public Health</i>	15	1121	2015
Yaota M, Niihori T, Mizuno S, Okamoto N, Hayashi S, Watanabe A, Yokozawa M, Santos F, Garcia S, <u>Ogata T</u> , Aoki Y*, et al	Spectrum of mutations and genotype-phenotype analysis in Noonan syndrome patients with <i>RIT1</i> mutations.	<i>Hum Genet</i>	135 (2)	209–222	2016
Saito K, Matsuzaki T, Iwasa T, Miyado M, Saito H, Hasegawa T, Homma K, Inoue E, Kubota T, Irahara M, <u>Ogata T</u> , Fukami M*	Multiple Androgen Biosynthesis Pathways Are Operating in Women with Polycystic Ovary Syndrome.	<i>J Steroid Biochem Mol Biol</i>	158	31–37	2016
Fujisawa Y, Sakaguchi K, Ono H, Yamaguchi R, Kato F, Kagami M, Fukami M, <u>Ogata T</u> *	Combined steroidogenic characters of fetal adrenal and Leydig cells in childhood adrenocortical carcinoma.	<i>J Steroid Biochem Mol Biol</i>	159	86–93	2016
Saito K, Matsuzaki T, Iwasa T, Miyado M, Saito H, Kubota T, Irahara M, <u>Ogata T</u> , Fukami M*	Blood allopregnanolone levels in women with polycystic ovary syndrome.	<i>Clin Endocrinol</i>	(accepted)		
Miyamichi D, Asahina M, Nakajima J, Sato M, Hosono K, Nomura T, Negishi T, Miyake N, Hotta Y, <u>Ogata T</u> , Matsumoto N*	Novel <i>HPS6</i> mutations identified by whole-exome sequencing in two Japanese sisters with suspected ocular albinism.	<i>J Hum Genet</i>	(accepted)		
Luk H-M, Lo F-M I, Sano S, Matsbara K, Nakamura A, <u>Ogata T</u> *, Kagami M*	Silver-Russell syndrome in a patient with somatic mosaicism for upd(11)mat identified by buccal cell analysis.	<i>Am J Med Genet A</i>	(accepted)		
Sano S, Nagasaki K, Kikuchi T, Nakabayashi K, Hata K, Fukami M, Kagami M, <u>Ogata T</u> *	Beckwith-Wiedemann syndrome and pseudohypoparathyroidism type Ib in a patient with multilocus methylation defects: a female-dominant phenomenon?	<i>J Hum Genet</i>	(accepted)		
<u>Ogata T</u> *, Kagami M	Kagami-Ogata syndrome: a clinically recognizable upd(14)pat and related disorder affecting the chromosome 14q32.2 imprinted region.	<i>J Hum Genet</i>	61 (2)	87–94	2016
Fukami M*. Seki A, <u>Ogata T</u>	<i>SHOX</i> Haploinsufficiency as a Cause of Syndromic and Non-Syndromic Short Stature.	<i>Mol Syndromol</i>	(accepted)		

緒方勤	インプリンティング疾患発症機序について .	脳と発達	47 (2)	17-22	2015
緒方勤	身長性の差 .	チャイルドヘルス	18 (1)	41-45	2015
緒方勤	性の分化および生殖発生遺伝学	産婦人科の実際	64 (3)	391-396	2015
緒方勤	ヒトインプリンティング異常症発症機序 : Kagami-Ogata syndrome / Temple syndrome を主として .	ホルモンと臨床	61 (8)	69-78	2015
緒方勤	生殖補助医療と小児科とのかわり .	小児科臨床	68 (7)	13-20	2015
宮戸真美、宮戸健二、緒方勤、深見真紀	MAMLD1 : 胎生期精巣におけるステロイドホルモン産生の新規調節因子	本生殖内分泌学会雑誌	20	19-24	2015
緒方勤、深見真紀	遺伝子解析と遺伝カウンセリング	産婦人科の実際 (いま、性分化とその異常を考える)	64 (10)	1301-1307	2015
小野裕之、緒方勤	軟骨毛髪低形成症	日本臨床	36 (別冊免疫症候群、第2版)	228-230	2016
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長谷川真理、位田忍他8名	卵精巣性性分化疾患12例の臨床的検討	日本小児科学会雑誌	119	566-572	2015
位田忍	Turner 症候群	小児内科	47	1760-1764	2015

鹿島田健一	Newborn screening for congenital adrenal hyperplasia in Tokyo, Japan from 1989 to 2013: a retrospective population-based study.	BMC Pediatrics	15	209.	2015
鹿島田健一	Clinical, biochemical, and genetic features of non-classical 21-hydroxylase deficiency in Japanese children.	Endocr Journal	62	277-82	2015
鹿島田健一	TALEN-Mediated Gene Disruption on Y Chromosome Reveals Critical Role of EIF2S3Y in Mouse Spermatogenesis.	Stem Cells Development.	24	1164-70	2015
鹿島田健一	卵巣発生において卵巣特異的転写因子 FOXL2 は WT1 による Sfl の発現を抑制する	日本生殖内分泌学会雑誌	20	47-51	2015
鹿島田健一	先天性副腎過形成(CAH)精査機関での診断のポイント ~17-OHP 高値をとる CAH の病型診断と遺伝学的検査について~	日本マススクリーニング学会誌	25	251-259	2015
Izumi Y, Musha I, Suzuki E, Iso M, Jinno T, Horikawa R, Amemiya S, Ogata T, <u>Fukami M</u> , Ohtake A	Hypogonadotropic hypogonadism in a female patient previously diagnosed as having Waardenburg syndrome due to a <i>SOX10</i> mutation.	Endocrine	49(2)	553-556	2015
Igarashi M, Wada Y, Kojima Y, Miyado M, Nakamura M, Muroya K, Mizuno K, Hayashi Y, Nonomura K, Kohri K, Ogata T, <u>Fukami M</u>	Novel Splice Site Mutation in MAMLD1 in a Patient with Hypospadias.	Sex Dev	9(3)	130-135	2015
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Ruiz-Arana IL, Hübner A, Cetingdag C, Krude H, Grüters A, <u>Fukami M</u> , Biebermann H, Köhler B	A Novel Hemizygous Mutation of MAMLD1 in a Patient with 46,XY Complete Gonadal Dysgenesis.	Sex Dev	9(2)	80-85	2015

Yagi H, Takagi M, Kon M, Igarashi M, <u>Fukami M</u> , Hasegawa Y	Fertility preservation in a family with a novel NR5A1 mutation.	Endocr J	62(3)	289–295	2015
Okuno M, Ogata T, Nakabayashi K, Urakami T, <u>Fukami M</u> , Nagasaki K	Endocrinopathies in a Boy with Cryptic Copy-number Variations on 4q, 7q, and Xp. Hum Genome Variat.	Hum Genome Variat [Epub ahead of print]			
Katoh-Fukui Y, Igarashi M, Nagasaki K, Horikawa R, Nagai T, Tsuchiya T, Suzuki E, Miyado M, Hata K, Nakabayashi K, Hayashi K, Matsubara Y, Baba T, Morohashi K, Igarashi A, Ogata T, Takada S, <u>Fukami M</u>	Testicular Dysgenesis/Regression without Campomelic Dysplasia in Patients Carrying Missense Mutations and Upstream Deletion of SOX9.	Mol Genet Genom Med	3(6)	550–557	2015
Miyado M, Miyado K, Katsumi M, Saito K, Nakamura A, Shihara D, Ogata T, <u>Fukami M</u>	Parturition failure in mice lacking Maml1.	Sci Rep	5	14705	2015
Kon M, Saito K, Mitsui T, Miyado M, Igarashi M, Moriya K, Nonomura K, Shinohara N, Ogata T, <u>Fukami M</u>	Copy-Number Variations of the Azoospermia Factor Region or SRY Are Not Associated with the Risk of Hypospadias.	Sex Dev [Epub ahead of print]			
Kon M, <u>Fukami M</u>	Submicroscopic copy-number variations associated with 46,XY disorders of sex development.	Mol Cell Pediatr	2(1)	7	2015
<u>深見真紀</u>	停留精巣とホルモン環境	小児外科	47(8)	793–795	2015
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宮戸真美, 宮戸健二, 緒方勤, <u>深見真紀</u>	MAMLD1: 胎生期精巣におけるステロイドホルモン産生の新規調節因子	日本生殖内分泌学会雑誌	20	19–23	2015
<u>深見真紀</u>	Sohval-Soffer 症候群	小児科診療増刊号 79 小児の症候群 診断と治療社	79		2015
<u>深見真紀</u>	Turner 症候群	小児疾患診療のための病態生理 2	2		2015