

## 研究成果の刊行に関する一覧表レイアウト（参考）

## 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

## 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
Suzuki E, Bo R, Sue K, Awano H, <b>Ogata T</b> , Narumi S, Kagami M, Sano S, Fukami M*	<i>De Novo</i> 50-bp <i>GNAS</i> ( <i>Gs-alpha</i> ) Intragenic Duplication in a Patient with Pseudohypoparathyroidism Type 1a	<i>Cytogenet Genome Res</i>	153 (3)	125–130	2018
Takasawa K, Gau M, Sutani A, Igarashi M, Ono M, Takemoto A, Takada S, Yamataka A, <b>Ogata T</b> , Morio T, Fukami M, Kashimada K*	Phenotypic variation in 46,XX disorders of sex development due to the NR5A1 p.R92W variant: a sibling case report and literature review	<i>Sex Dev</i>	11 (5–6)	284–288	2018
Haug MG, Brendehaug A, Houge G, Kagami M, <b>Ogata T</b> *	Mosaic UPD(14)pat in a Patient with Mild Features of Kagami-Ogata Syndrome	<i>Clin Case Rep</i>	6 (1)	91–95	2018
Ushijima K, Fukami M, Ayabe T, Narumi S, Okuno M, Nakamura A, Takahashi T, Ihara K, Ohkubo K, Tachikawa E, Nakayama S, Arai J, Kikuchi N, Kikuchi T, Kawamura T, Urakami T, Hata K, Nakabayashi K, Matsubara Y, Amemiya S, <b>Ogata T</b> , Yokota I, Sugihara S	The Japanese Study Group of Insulin Therapy for Childhood and Adolescent Diabetes. Comprehensive screening for monogenic diabetes in 89 Japanese children with insulin-requiring antibody-negative type 1 diabetes	<i>Pediatr Diabetes</i>	19 (2)	243–250	2018

Ohsako S*, Aiba T, Miyado M, Fukami M, <b>Ogata T</b> , Hayashi Y, Mizuno K, Kojima Y	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	<i>Arch Environ Contam Toxicol</i>	74 (2)	240–247	2018
Ozono K, <b>Ogata T</b> , Horikawa R, Matsubara Y, Ogata Y, Nishijima K, Yokoya S	Efficacy and safety of two doses of Norditropin® (somatropin) in short stature due to Noonan syndrome: a 2-year randomized, double-blind, multicenter trial in Japanese patients	<i>Endocr J</i>	65 (2)	159–174	2018
Yamamoto K, Okamoto S, Fujisawa Y, Fukami M, Saito H, <b>Ogata T</b>	<i>FGFR1</i> Disruption Identified by Whole Genome Sequencing in a Male With a Complex Chromosomal Rearrangement and Hypogonadotropic Hypogonadism	<i>Am J Med Genet A</i>	176 (1)	139–143	2018
Shima H, MD*, Koehler K*, Nomura Y, Sugimoto K, Satoh A, <b>Ogata T</b> , Fukami M, Schuelke M, Huebner A, Narum S	Two patients with MIRAGE syndrome lacking haematological features: role of somatic second-site reversion SAMD9 mutations	<i>J Med Genet</i>	55 (2)	81–85	2018
Okuno M, Ayabe T, Yokota I, Musya I, Shiga K, Kikuchi T, Kikuchi N, Ohtake A, Nakamura A, Nakabayashi K, Okamura K, Momozawa Y, Suzuki J, Urakami T, Kawamura T, Amemiya S, <b>Ogata T</b> , Sugihara S, Fukami M*	The Japanese Study Group of Insulin Therapy for Childhood and Adolescent Diabetes. Protein-Altering Variants of PTPN2 in Childhood-onset Type 1A Diabetes	<i>Diabet Med</i>	35 (3)	376–380	2018
Ono H, Numakura C, Homma K, Hasegawa T, Tsutsumi S, Kato F, Fujisawa Y, Fukami M, <b>Ogata T</b> *	Longitudinal Serum and Urine Steroid Metabolite Profiling in a 46,XY Infant with Prenatally Identified POR Deficiency	<i>J Steroid Biochem Mol Biol</i>	178	177–184	2018

Nakashima M, Hiraide T, Yamoto K, Fukuda T, Kato M, Ikeda H, Sugie Y, Aoto K, Kaname T, Nakabayashi K, <b>Ogata T</b> , Matsumoto N, Saitsu H*	<i>De novo</i> variants in SETD1B are associated with intellectual disability, epilepsy and autism	<i>Hum Genet</i>	137 (1)	95–104	2018
Montalbano A, Juergensen L, Fukami M, Thiel, CT, Hauer NH, Roeth R, Weiss B, Naiki Y, <b>Ogata T</b> , Hassel D, Rappold GA*	Functional missense and splicing variants in the retinoic acid catabolizing enzyme CYP26C1 in idiopathic short stature	<i>Eur J Hum Genet</i>	26 (8)	1113–1120	2018
Nakamura A, Muroya K, Ogata-Kawata H, Nakabayashi K, Matsubara K, <b>Ogata T</b> , Kurosawa K, Fukami M, Kagami M*	A case of paternal uniparental isodisomy for chromosome 7 associated with overgrowth	<i>J Med Genet</i>	55 (8)	567–570	2018
Nakamura S, Kobori Y, Ueda Y, Tanaka Y, Ishikawa H, Yoshida A, Katsumi M, Saito K, Nakamura A, <b>Ogata T</b> , Okada H, Nakai H, Miyado M, Fukami M*	STX2 is a causative gene for nonobstructive azoospermia	<i>Hum Mutat</i>	39 (6)	830–833	2018
Ono H, Saitsu H, Horikawa R, Nakashima S, Ohkubo Y, Yanagi K, Nakabayashi K, Fukami M, Fujisawa Y, <b>Ogata T</b> *	Partial androgen insensitivity syndrome caused by a deep intronic mutation creating an alternative splice acceptor site of the AR gene	<i>Sci Rep</i>	8 (1)	2287	2018
Kawashima S, Nakamura A, Inoue T, Matsubara K, Horikawa R, Wakui K, Tkano K, Fukushima Y, Tatematsu T, Mizuno S, Tsubaki J, Kure S, Matsubara Y, <b>Ogata T</b> , Fukami M, Kagami M*	Maternal uniparental disomy for chromosome 20: physical and endocrinological characteristics of five patients	<i>J Clin Endocrinol Metab</i>	103 (6)	2083–2088	2018

Hernandez Mora JR, Tayama C, Sánchez-Delgado M, Monteagudo-Sánchez A, Hata K, <b>Ogata T</b> , Medrano J, Poo-Llanillo ME, Simón C, Moran S, Esteller M, Tenorio J, Pablo Lapunzina P, Kagami M, Monk D, Nakabayashi K*	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform	<i>Epigenomics</i>	10 (7)	941–954	2018
Kurata K, Hosono K, Hikoya A, Kato A, Saitsu H, Minoshima S, <b>Ogata T</b> , Hotta Y*	Clinical characteristics of a Japanese patient with Bardet-Biedl syndrome caused by BBS10 mutations	<i>Jpn J Ophthalmol A</i>	62 (4)	458–466	2018
Mano H, Fujiwara S, Takamura K, Kitoh H, Takayama S, <b>Ogata T</b> , Hashimoto S, Haga N*	Congenital limb deficiency in Japan: A cross-sectional nationwide survey on its epidemiology	<i>BMC Musculoskelet Disord</i>	19(1)	262	2018
Yoshida T, Matsuzaki T, Miyado M, Saito K, Iwasa T, Matsubara Y, <b>Ogata T</b> , Irahara M, Fukami M*	11-oxygenated C19 steroids as circulating androgens in women with polycystic ovary syndrome	<i>Endocr J</i>	65 (10)	979–990	2018
Inoue T, Yagasaki H, Nishioka J, Nakamura A, Matsubara K, Narumi S, Nakabayashi K, Yamazawa K, Fuke T, Oka A, <b>Ogata T</b> , Fukami M, Kagami M*	Molecular and clinical analyses of two patients with UPD(16)mat detected by screening 94 patients with Silver-Russell syndrome phenotype of unknown etiology	<i>J Med Genet</i>			2018
Shimizu D, Iwashima S, Sato K, Hayano S, Fukami M, Saitsu H, <b>Ogata T</b>	<i>GATA4</i> variant identified by whole exome sequencing in a Japanese family with atrial septal defect: implications for male sex development	<i>Clin Case Rep</i>	6 (11)	2229–2233	2018
Igarashi M, Mizuno K, Kon M, Narumi S, Kojima Y, Hayashi Y, <b>Ogata T</b> , Fukami M*	<i>GATA4</i> mutations are uncommon in patients with 46,X,Y disorders of sex development without heart anomaly	<i>Asian J Androl</i>	20 (6)	629–631	2018

Suzuki E, Shima H, Kagami M, Soneda S, Tanaka T, Yatsuga S, Nishioka J, Oto Y, Kamiya T, Naiki Y, <b>Ogata T</b> , Fujisawa Y, Nakamura A, Kawashima S, Morikawa S, Horikawa R, Sano S, Fukami M*	(Epi)genetic defects of MKRN3 are rare in Asian patients with central precocious puberty	<i>Hum Genome Var</i>			2019
Nakashima M, Tohyama J, Nakagawa E, Watanabe Y, Siew CG, Kwong CS, Yamoto K, Hiraide T, Fukuda T, Kaname T, Nakabayashi K, Hata K, <b>Ogata T</b> , Saitsu H, Matsumoto N*	Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures	<i>J Hum Genet</i>	64 (4)	313–322	2019
Hiraide T, <b>Ogata T</b> , Watanabe S, Nakashima M, Fukuda T, Saitsu H*	Coexistence of a CAV3 mutation and a DMD deletion in a family with complex muscular diseases	<i>Brain Dev</i>	41 (?)	474–479	2019
Hattori H, Hiura H, Kitamura A, Miyauchi N, Kobayashi N, Takahashi S, Okae H, Kyono K, Kagami M, <b>Ogata T</b> , Arima T*	Association of four imprinting disorders and ART	<i>Clin Epigenetics</i>	11 (1)	21	2019
Miyado M, Fukami M, Takada S, Terao M, Nakabayashi K, Hata K, Matsubara Y, Tanaka Y, Sasaki G, Nagasaki K, Shiina M, Ogata K, Masunaga Y, Saitsu H, <b>Ogata T</b> *	Germline-derived gain-of-function variants of Gs $\alpha$ -coding GNAS gene identified in nephrogenic syndrome of inappropriate antidiuresis	<i>J Am Soc Nephrol</i>			2019
Matsubara K, Itoh M, Shimizu K, Saito S, Enomoto K, Nakabayashi K, Hata K, Kurosawa K, <b>Ogata T</b> , Fukami M, Kagami M	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	<i>Clin Epigenetics</i>	11 (1)	36	2019

Kagami M, Yanagisawa A, Ota M, Matsuoka K, Nakamura A, Matsubara K, Nakabayashi K, Takada S, Fukami M, <b>Ogata T*</b>	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	<i>Clin Epigenetics</i>	11 (1)	42	2019
Matsushita R*, Nagasaki K, Ayabe T, Kinjo S, Haruna H, Ihara K, Hasegawa T, <b>Ogata T</b> , Ozono K, Minamitani K	thyroid Committee of the Japanese Society for Pediatric Endocrinology: Early Calcitonin Level-Based Thyroidectomy May Reduce Postoperative Complications and Improve Prognosis in MEN2	<i>J Pediatr Endocrinol Metab</i>			
Hamanaka K, Takata A, Uchiyama Y, Miyatake S, Miyake N, Mitsuhashi S, Iwama K, Fujita A, Imagawa E, Alkanaq AN, Koshimizu E, Azuma Y, Nakashima M, Mizuguchi T, Saitsu H, Yuka Wada, Minami S, Katoh-Fukui Y, Masunaga Y, Fukami M, Hasegawa T, <b>Ogata T</b> , Matsumoto N*	<i>MYRF</i> haploinsufficiency causes 46,XY and 46,XX disorders of sex development	<i>Hum Mol Genet</i>			2019
Fukami M*, Suzuki E, Igarashi M, Miyado M, <b>Ogata T</b>	Gain-of-Function Mutations in G-protein Coupled Receptor Genes Associated with Human Endocrine Disorders	<i>Clin Endocrinol</i>	88 (3)	351–359	2018