

別紙 4

研究成果の刊行に関する一覧表レイアウト (参考)

書籍

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雑誌

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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
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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
Yamato K, Okamoto S, Fujisawa Y, Fukami M, Saitsu 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
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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

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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
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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
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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
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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
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
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>	6	7	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 (5)	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
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Uehara E, Hattori A, Shima H, Ishiguro A, Abe Y, <b>Ogata T</b> , Ogawa E, Fukami M*	Unbalanced Y;7 translocation between two low-similarity sequences leading to SRY-positive 45,X-testicular disorders of sex development.	<i>Cytogenet Genome Res</i>	158 (3)	115–120	2019
Ushijima K, Narumi S, <b>Ogata T</b> , Yokota I, Sugihara S, Kaname T, Horikawa Y, Matsubara Y, Fukami M*, Kawamura T	Japanese Study Group of Insulin Therapy for Childhood and Adolescent Diabetes.: <i>KLF11</i> ( <i>MODY7</i> ) variant in a family clinically diagnosed with early childhood-onset type 1B diabetes.	<i>Pediatr Diabetes</i>	20 (6)	712–719	2019
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Shimizu D, Sakamoto R, Yamoto K, Saitsu H, Fukami M, Nishimura G, <b>Ogata T*</b>	De novo AFF3 variant in a patient with mesomelic dysplasia with foot malformation.	<i>J Hum Genet</i>	64 (10)	1041–1044	2019
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