

## 業績 2019

- 内分泌 G 英文 18
1. Inoue T, Yagasaki H, Nishioka J, Nakamura A, Matsubara K, Narumi S, Nakabayashi K, Yamazawa K, Fuke T, Oka A, **Ogata T**, 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. *J Med Genet* 56 (6): 413–418, 2019. pii: jmedgenet-2018-105463. doi: 10.1136/jmedgenet-2018-105463.
  2. Suzuki E, Shima H, Kagami M, Soneda S, Tanaka T, Yatsuga S, Nishioka J, Oto Y, Kamiya T, Naiki Y, **Ogata T**, 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. *Hum Genome Var* 2019 Jan 21;6:7. doi: 10.1038/s41439-019-0039-9. PMID: 30675365
  3. Nakashima M, Tohyama J, Nakagawa E, Watanabe Y, Siew CG, Kwong CS, **Yamamoto K**, **Hiraide T**, **Fukuda T**, Kaname T, Nakabayashi K, Hata K, **Ogata T**, Saitsu H, Matsumoto N\*: Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures. *J Hum Genet* 64 (4): 313–322, 2019. doi: 10.1038/s10038-018-0559-z. PMID: 30655572 (神経)
  4. **Hiraide T**, **Ogata T**, Watanabe S, Nakashima M, **Fukuda T**, Saitsu H\*: Coexistence of a CAV3 mutation and a DMD deletion in a family with complex muscular diseases. *Brain Dev* 41 (5): 474–479, 2019. pii: S0387-7604(18)30594-1. doi: 10.1016/j.braindev.2019.01.005. PMID: 30723005 (神経)
  5. Hattori H, Hiura H, Kitamura A, Miyauchi N, Kobayashi N, Takahashi S, Okae H, Kyono K, Kagami M, **Ogata T**, Arima T\*: Association of four imprinting disorders and ART. *Clin Epigenetics* 11 (1): 21, 2019. doi: 10.1186/s13148-019-0623-3. PMID: 30732658
  6. 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, **Ogata T**\*: Germline-derived gain-of-function variants of Gsα-coding *GNAS* gene identified in nephrogenic syndrome of inappropriate antidiuresis. *J Am Soc Nephrol* 30 (5): 877–889, 2019. pii: ASN.2018121268. doi: 10.1681/ASN.2018121268. PMID: 30962325
  7. Matsubara K, Itoh M, Shimizu K, Saito S, Enomoto K, Nakabayashi K, Hata K, Kuroswa K, **Ogata T**, 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. *Clin Epigenetics*. 11 (1): 36, 2019. doi: 10.1186/s13148-019-0633-1. PMID: 30819260
  8. Kagami M, Yanagisawa A, Ota M, Matsuoka K, Nakamura A, Matsubara K, Nakabayashi K, Takada S, Fukami M, **Ogata T**\*: 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. *Clin Epigenetics* 11 (1): 42, 2019. doi: 10.1186/s13148-019-0640-2. PMID: 30846001
  9. **Matsushita R**\*, Nagasaki K, Ayabe T, Kinjo S, Haruna H, Ihara K, Hasegawa T, **Ogata T**, Ozono K, Minamitani K, Thyroid Committee of the Japanese Society for Pediatric Endocrinology: Present status of prophylactic thyroidectomy in pediatric multiple endocrine neoplasia 2: a nationwide survey in Japan 1997–2017. *J Pediatr Endocrinol Metab*. 2019;32(6):585–595. doi: 10.1515/j pem-2018-0444. PMID: 31150358
  10. 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, **Ogata T**, Matsumoto N\*: *MYRF* haploinsufficiency causes 46,XY and 46,XX disorders of sex development. *Hum Mol Genet.* 28 (14): 2319–2329, 2019 doi: 10.1093/hmg/ddz066. PMID: 30985895
11. Hosokawa Y, Higuchi S, Kawakita R, Hata I, Urakami T, Isojima T, Takasawa K, Matsu bara Y, Mizuno H, Maruo Y, Matsui K, Aizu K, Jinno K, Araki S, **Fujisawa Y**, Osugi K, Tono C, Takeshima Y, Yorifuji T\*: Pregnancy outcome of Japanese patients with gluc okinase-maturity-onset diabetes of the young. *J Diabetes Investig.* 10(6):1586-1589, 201 9 doi: 10.1111/jdi.13046. PMID: 30897270
  12. Uehara E, Hattori A, Shima H, Ishiguro A, Abe Y, **Ogata T**, Ogawa E, Fukami M\*: Unbalanced Y;7 translocation between two low-similarity sequences leading to SRY-positive 45,X-testicular disorders of sex development. *Cytogenet Genome Res* 158 (3): 115–120, 2019. doi: 10.1159/000501378. PMID: 31266029. Epub 2019 Jul 5.
  13. Ushijima K, Narumi S, **Ogata T**, Yokota I, Sugihara S, Kaname T, Horikawa Y, Matsub ara Y, Fukami M\*, Kawamura T; Japanese Study Group of Insulin Therapy for Childhoo d and Adolescent Diabetes: KLF11 (MODY7) variant in a family clinically diagnosed wi th early childhood-onset type 1B diabetes. *Pediatr Diabetes.* 20 (6): 712–719, 2019. doi: 10.1111/pedi.12868. PMID: 31124255. [Epub ahead of print]
  14. Yoshida T, Miyado M, Mikami M, Suzuki E, **Kinjo K**, Matsubara K, **Ogata T**, Akutsu H, Kagami M, Fukami M\*: Aneuploid rescue precedes X chromosome inactivation and i ncreases the incidence of its skewness by reducing the size of the embryonic progenitor c ell pool. *Hum Reprod.* 34 (9): 1762–1769. doi: 10.1093/humrep/dez117. PMID: 3139825 9. [Epub ahead of print] 2019 Sep 29
  15. **Yamoto K**, Saitsu H, Nishimura G, Kosaki R, Takayama S, Haga N, Tonoki H, Okumur a A, Horii E, Okamoto N, Suzumura H, Ikegawa S, **Kato F**, **Fujisawa Y**, **Nagata E**, Tak ada S, Fukami M, **Ogata T**\*: Comprehensive clinical and molecular studies in split hand /foot malformation: identification of two plausible candidate genes (LRP6 and UBA2). *E ur J Hum Genet.* 27 (12): 1845–1857, 2019. doi: 10.1038/s41431-019-0473-7. PMID: 31 332306. Epub 2019 Jul 22.
  16. **Ohishi A**, **Masunaga Y**, Iijima S, **Yamoto K**, **Kato F**, Fukami M, Saitsu H, **Ogata T**\*: De novo ZBTB7A variant in a patient with macrocephaly, intellectual disability, and slee p apnea: implications for the phenotypic development in 19p13.3 microdeletions. *J Hum Genet* 65 (2): 181–186, 2020. doi: 10.1038/s10038-019-0690-5. PMID: 31645653. (新生児)
  17. **Fukuda T**\*, **Hiraide T**, **Yamoto K**, Nakashima M, Kawai T, Yanagi K, **Ogata T**, Saits u H: Exome reports: A de novo GNB2 variant associated with global developmental dela y, intellectual disability, and dysmorphic features. *Eur J Med Genet* 2019 Nov 4:103804. doi: 10.1016/j.ejmg.2019.103804. PMID: 31698099. (神経)
  18. **Shimizu D**, Sakamoto R, **Yamoto K**, Saitsu H, Fukami M, Nishimura G, **Ogata T**\*: *De novo AFF3* variant in a patient with mesomelic dysplasia with foot malformation. *J Hum Genet* 64 (10):1041–1044, 2019. doi: 10.1038/s10038-019-0650-0. PMID: 31388108. (血液)

● 免疫・アレルギーG 英文4(+1症例報告) 和文3

1. Palmar hyperlinearity in early childhood atopic dermatitis is associated with filaggrin mut ation and sensitization to egg. Fukuie T, **Yasuoka R**, Fujiyama T, Sakabe JI, Taguchi T, Tokura Y. *Pediatr Dermatol.* 2019 Mar;36(2):213-218.

2. Palmar hyperlinearity in early childhood atopic dermatitis is associated with filaggrin mutation and sensitization to egg. Fukuie T, **Yasuoka R**, Fujiyama T, Sakabe JI, Taguchi T, Tokura Y. *Pediatr Dermatol*. 2019 Mar;36(2):213-218. Pharmacokinetics of mycophenolate mofetil in juvenile patients with autoimmune diseases. Nakaseko H, Iwata N, **Yasuoka R**, Kohagura T, Abe N, Kawabe S, Mori M. *Mod Rheumatol*. 2019 Nov;29(6):1002-1006.
3. Expanding clinical spectrum of autosomal dominant pyrin-associated autoinflammatory disorder caused by the heterozygous MEFV p.Thr577Asn variant. Nakaseko H, Iwata N, Izawa K, Shibata H, Yasuoka R, Kohagura T, Abe N, Kawabe S, Ishikomori R. *Rheumatology (Oxford)*. 2019 Jan 1;58(1):182-184. (症例報告)
4. Clinical subsets of juvenile dermatomyositis classified by myositis-specific autoantibodies: Experience at a single center in Japan. Iwata N, Nakaseko H, Kohagura T, **Yasuoka R**, Abe N, Kawabe S, Sugiura S, Muro Y. *Mod Rheumatol*. 2019 Sep;29(5):802-807
5. Risk factors for hypersensitivity reactions to tocilizumab introduction in systemic juvenile idiopathic arthritis. **Yasuoka R**, Iwata N, Abe N, Kohagura T, Nakaseko H, Shimizu M, Kawabe S. *Mod Rheumatol*. 2019; 29: 324-7
6. 加藤由希子, 夏目統, 久保田綾乃, 幸田昌樹, 坂井聰, 松永真由美, 犬塚祐介, 田口智英. 小児食物アレルギー患者をもつ家族の震災時対策に関する実態調査. 日本小児臨床アレルギー学会誌 2019; 17: 11-18.
7. 犬塚祐介, 夏目統, 松永真由美, 加藤由希子, 褒田晃央, 溝口優子, 田口智英. 当院での食物アレルギーに対する栄養食事指導の評価. 小児科臨床 2019; 72 (7): 863-70
8. 高橋 仁美(国立成育医療研究センター臨床研究センター), 夏目 統, 成田 雅美, 山本 貴和子, 権島 重憲, 中村 秀文, 佐古 まゆみ, 大矢 幸弘. 臨床研究参加の意思決定に影響する因子の検討 乳児アトピー性皮膚炎患児の保護者を対象とした研究参加同意/不同意の意思決定要因の質問紙調査を実施して. 臨床薬理 2019; 50: 259-64

● 循環器 G 英文 1 (うち症例報告 1) 和文 0 (うち症例報告 0 )

1. Seki K, Iwashima S, Uchiyama H, Ohishi A, Ishikawa T. Successful Management of Pulmonary Arterial Hypertension by Monitoring N-Terminal Pro-B-Type Natriuretic Peptide Serum Levels in a Preterm Infant With Chronic Lung Disease: A Case Report. *AJP Rep* 9(2):e133-7, 2019.

● 神経 G 英文 11

1. Yokoi K, Nakajima Y, Ohye T, Inagaki H, Wada Y, **Fukuda T**, Sugie H, Yuasa I, Ito T, Kurahashi H. Disruption of the Responsible Gene in a Phosphoglucomutase 1 Deficiency Patient by Homozygous Chromosomal Inversion. *JIMD Rep*. 2019;43:85-90. doi: 10.1007/8904\_2018\_108. Epub 2018 May 1. PMID: 29752652; PMCID: PMC6323009.
2. Itamura S, Okanishi T, Arai Y, Nishimura M, Baba S, Ichikawa N, Hirayama Y, Ishihara N, **Hiraide T**, Ishigaki H, **Fukuda T**, Otsuki Y, Enoki H, Fujimoto A. Three Cases of Hemiconvulsion-Hemiplegia-Epilepsy Syndrome With Focal Cortical Dysplasia Type IIId. *Front Neurol*. 2019 Nov 20;10:1233. doi: 10.3389/fneur.2019.01233. PMID: 31824410; PMCID: PMC6879674.
3. Nakashima M, Tohyama J, Nakagawa E, Watanabe Y, Siew CG, Kwong CS, Yamoto K, **Hiraide T**, **Fukuda T**, Kaname T, Nakabayashi K, Hata K, **Ogata T**, Saitsu H, Matsumoto N. Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures. *J Hum Genet*. 2019 Apr;64(4):313-322. doi: 10.1038/s10038-018-0559-z. Epub 2019 Jan 17. PMID: 30655572.
4. **Fukuda T**, **Hiraide T**, Yamoto K, Nakashima M, Kawai T, Yanagi K, **Ogata T**, Saitsu H. Exome reports A de novo *GNB2* variant associated with global developmental delay, intellectual disability, and dysmorphic features. *Eur J Med Genet*. 2020 Apr;63(4):103804. doi: 10.1016/j.ejmg.2019.103804. Epu

- b 2019 Nov 4. PMID: 31698099.
5. Ago Y, Sugie H, **Fukuda T**, Otsuka H, Sasai H, Nakama M, Abdelkreem E, Fukao T. A rare *PHKA2* variant (p.G991A) identified in a patient with ketotic hypoglycemia. *JIMD Rep*. 2019 May 28;48(1):1-5-18. doi: 10.1002/jmd2.12041. PMID: 31392108; PMCID: PMC6606978.
  6. **Hiraide T, Ogata T**, Watanabe S, Nakashima M, **Fukuda T**, Saitsu H. Coexistence of a *CAV3* mutation and a DMD deletion in a family with complex muscular diseases. *Brain Dev*. 2019 May;41(5):474-479. doi:10.1016/j.braindev.2019.01.005. Epub 2019 Feb 2. PMID: 30723005.
  7. Miyamoto S, Aoto K, **Hiraide T**, Nakashima M, Takabayashi S, Saitsu H. Nanopore sequencing reveals a structural alteration of mirror-image duplicated genes in a genome-editing mouse line. *Congenit Anom (Kyoto)*. 2020 Jul;60(4):120-125. doi: 10.1111/cga.12364. Epub 2019 Dec 25. PMID: 31837184.
  8. **Hiraide T**, Hattori A, Ieda D, Hori I, Saitoh S, Nakashima M, Saitsu H. De novo variants in *SETD1B* cause intellectual disability, autism spectrum disorder, and epilepsy with myoclonic absences. *Epilepsia Open*. 2019 May 24;4(3):476-481. doi: 10.1002/epi4.12339. PMID: 31440728; PMCID: PMC6698685.
  9. Nakashima M, Tohyama J, Nakagawa E, Watanabe Y, Siew CG, Kwong CS, Yamoto K, **Hiraide T, Fukuda T**, Kaname T, Nakabayashi K, Hata K, **Ogata T**, Saitsu H, Matsumoto N. Identification of de novo *CSNK2A1* and *CSNK2B* variants in cases of global developmental delay with seizures. *J Hum Genet*. 2019 Apr;64(4):313-322. doi: 10.1038/s10038-018-0559-z. Epub 2019 Jan 17. PMID: 30655572.
  10. **Hiraide T**, Kaba Yasui H, Kato M, Nakashima M, Saitsu H. A de novo variant in *RAC3* causes severe global developmental delay and a middle interhemispheric variant of holoprosencephaly. *J Hum Genet*. 2019 Nov;64(11):1127-1132. doi: 10.1038/s10038-019-0656-7. Epub 2019 Aug 16. PMID: 31420595.
  11. Miyamoto S, Nakashima M, Ohashi T, **Hiraide T**, Kurosawa K, Yamamoto T, Takanashi J, Osaka H, Inoue K, Miyazaki T, Wada Y, Okamoto N, Saitsu H. A case of de novo splice site variant in *SLC35A2* showing developmental delays, spastic paraplegia, and delayed myelination. *Mol Genet Genomic Med*. 2019 Aug;7(8):e814. doi: 10.1002/mgg3.814. Epub 2019 Jun 23. PMID: 31231989; PMCID: PMC6687661.

● 新生児 G 英文 5 (うち症例報告 1 循環器グループと重複 1) 和文 0

1. **Iijima S.** Current knowledge of transient hypothyroxinemia of prematurity: to treat or not to treat? *J Matern Fetal Neonatal Med* 2019; 32:2591-2597.
2. Shibasaki J, Mukai T, Tsuda K, Takeuchi A, Ioroi T, Sano H, Yutaka N, Takahashi A, Sobajima H, Tamura M, Hosono S, Nabetani M, Iwata O, **Baby Cooling Registry of Japan Collaboration Team**. Outcomes related to 10-min Apgar scores of zero in Japan. *Arch Dis Child Fetal Neonatal Ed* 105(1):64-68, 2020.
3. **Iijima S.** Current knowledge about the in utero and peripartum management of fetal goiter associated with maternal Graves' disease. *Eur J Obstet Gynecol Reprod Biol X* 2019; 3: 100027.
4. **Iijima S.** Late-onset glucocorticoid-responsive circulatory collapse in premature infants. *Pediatr Neonatol* 60: 603-610, 2019.
5. Seki K, Iwashima S, Uchiyama H, **Ohishi A**, Ishikawa T. Successful management of pulmonary arterial hypertension by monitoring N-terminal pro-B-type natriuretic peptide serum levels in a preterm infant with chronic lung disease: a case report. *AJP Rep* 2019; 9: e133-e137.

● 血液・腫瘍 G 英文 3 (うち症例報告 2) 和文 1 (うち症例報告 0 )

1. **Shimizu D**, Sakamoto R, **Yamato K**, Saitsu H, Fukami M, Nishimura G and **Ogata T**. De novo AFF3 variant in a patient with mesomelic dysplasia with foot malformation. *Journal of Human Genetics*. 2019;64(10): 1041-1044.

2. Fukuchi K, Tatsuno K, Sakaguchi K, Sano S, Sasaki T, Aoki S, Kubo A and Tokura Y. Novel gene mutations in Chediak-Higashi syndrome with hyperpigmentation. *Journal of Dermatology*. 2019;46(11):e416-e8.
3. 坂口 公祥. 小児白血病における薬剤感受性試験. *BIO Clinica*. 2019;34(6):614-20.

● 腎臓 G 英文 1 和文 9

1. Hibino S, Uemura O, Uchida H, Majima H, Yamaguchi R, Tanaka K, Kawaguchi A, Yamakawa S, Fujita N: Solute Clearance and Fluid Removal: Large-Dose Cyclic Tidal Peritoneal Dialysis. *Ther Apher Dial*. 2019 Apr;23(2):180-186. doi: 10.1111/1744-9987.12765. Epub 2018 Oct 24. PMID: 30259676
2. 加賀田敬郎, 北形綾一, 西村竜哉, 内田博之, 田中一樹, 日比野聰, 藤田直也 : 乳幼児期に腹膜透析を開始した小児の栄養と成長. *腎と透析* 87 別冊 腹膜透析 2019, 182–184, 2019
3. 田中一樹, 北形綾一, 西村竜哉, 加賀田敬郎, 内田博之, 日比野聰, 藤田直也 : 小児腹膜透析患者における 24 時間自由行動下血圧測定の検討. *日本小児 PD・HD 研究会雑誌* 31: 104-106, 2019
4. 西村竜哉, 北形綾一, 加賀田敬郎, 内田博之, 田中一樹, 日比野聰, 藤田直也 : 腹膜透析カテーテル関連感染症予防に対するクロルヘキシジンでの出口部消毒の有用性. *日本小児 P D・HD 研究会雑誌* 31: 98-100, 2019
5. 日比野聰, 北形綾一, 西村竜哉, 加賀田敬郎, 内田博之, 田中一樹, 藤田直也 : ネフローゼ症候群の体液貯留時におけるトルバズタンの利尿効果. *日本小児体液研究会誌* 11: 61-66, 2019
6. 北形綾一, 西村竜哉, 加賀田敬郎, 内田博之, 田中一樹, 日比野聰, 長野智那, 野津寛大, 飯島一誠, 武田朝美, 藤田直也 : 初回腎生検で膜性増殖性糸球体腎炎 III 型様病理像を呈し、WT1 遺伝子 exon9 領域に新規ミスセンス変異を同定した女児例. *日本小児腎不全学会雑誌* 39: 172-175, 2019
7. 北形綾一, 西村竜哉, 加賀田敬郎, 内田博之, 田中一樹, 日比野聰, 伊藤健太, 藤田直也 : *Chryseobacterium indologenes* による腹膜透析関連腹膜炎の 1 女児例. *腎と透析* 87 別冊 腹膜透析 2019, 109-111, 2019
8. 内田 博之, 北形 綾一, 西村 竜哉, 加賀田 敬郎, 田中 一樹, 日比野 聰, 藤田 直也 : 小児腎疾患患者に対する 24 時間自動血圧測定の有用性. *日本小児高血圧研究会誌*(1344-0217)16巻1号 Page9-13(2019. 07)
9. 伊藤 創太郎, 山川 聰, 日比野 聰, 河口 亜津彩, 山口 玲子, 真島 久和, 内田 博之, 藤田 直也 : 小児ネフローゼ症候群におけるシクロスボリンの液剤とカプセル剤の薬物動態の変化. *日本小児腎臓病学会雑誌*(0915-2245)32巻1号 Page24-30(2019. 04)
10. 田中 一樹, 松田 百代, 西村 竜哉, 加賀田 敬郎, 内田 博之, 日比野 聰, 藤田 直也, 上原 央久, 鈴木 裕子, 吉野 薫 : VUR の手術適応について異なる選択をした CKD の 3 例. *日本逆流性腎症フォーラム記録集*(2435-323X)26回 Page38-40(2019. 01)