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

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
Masunaga Y, Inoue T, Yamoto K, Fujisawa Y, Sato Y, Kawashima-Sonoyama Y, Morisada N, Iijima K, Ohata Y, Namba N, Suzumura H, Kuribayashi R, Yamaguchi Y, Yoshihashi H, Fukami M, Saitsu H, Kagami M*, Ogata T	IGF2 Mutations: Report of Five Cases, Review of the Literature, and Comparison with H19/IGF2:IG-DMR Epimutations.	***J Clin Endocrinol Metab***	105（1）	dgz034	2020
Yamoto K, Saitsu H, Fujisawa Y, Kato F, Matsubara K, Fukami M, Kagami M, Ogata T	Coffin-Lowry syndrome in a girl with 46,XX,t(X;11)(p22;p15)dn: Identification of *RPS6KA3* disruption by whole genome sequencing.	***Clin Case Rep***	8（6）	1076-1080	2020
Yamazawa K*, Inoue T, Sakemi Y, Nakashima T, Yamashita H, Khono K, Fujita H, Enomoto K, Nakabayashi K, Hata K, Nakashima M, Matsunaga T, Nakamura A, Matsubara K, Ogata T, Kagami M	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.	***J Med Genet***			2020

Inoue T, Nakamura A, Iwahashi-Odano M, Tanase-Nakao K, Matsubara K, Nishioka J, Maruo Y, Hasegawa Y, Suzumura H, Sato S, Kobayashi Y, Murakami N, Nakabayashi K, Yamazawa K, Fuke T, Narumi S, Oka A, Ogata T, Fukami M, Kagami M	Contribution of gene mutations to Silver-Russell syndrome phenotype: multigene sequencing analysis in 92 etiology-unknown patients.	***Clin Epigenetics***	12(1)	86	2020
Hara-Isono K, Matsubara K, Mikami M, Arima T, Ogata T, Fukami M, Kagami M	Assisted reproductive technology represents a possible risk factor for development of epimutation-mediated imprinting disorders for mothers aged ≥ 30 years.	***Clin Epigenetics***	12(1)	111	2020
Masunaga Y, Fujisawa Y, Muramatsu M, Ono H, Inoue T, Fukami M, Kagami M, Saitsu H, Ogata T	Insulin resistant diabetes mellitus in SHORT syndrome: case report and literature review.	***Endocr J***	68(1)	111-117	2020
Hara-Isono K, Matsubara K, Fuke T, Yamazawa K, Satou K, Murakami N, Saitoh S, Nakabayashi K, Hata K, Ogata T, Fukami M, Kagami M	Genome-wide methylation analysis in Silver-Russell syndrome, Temple syndrome, and Prader-Willi syndrome.	***Clinical Epigenetics***	12(1)	159	2020
Omark J, Masunaga Y, Hannibal M, Shaw B, Fukami M, Kato F, Saitsu H, Kagami M, Ogata T	Kagami-Ogata syndrome in a patient with 46,XX,t(2;14)(q11.2;q32.2)mat disrupting MEG3.	***J Hum Genet***	66(4)	439-443	2021
Masunaga Y, Kagami M, Kato F, Usui T, Yonemoto T, Mishima K, Fukami M, Aoto, K, Saitsu H, **Ogata T**	Parthenogenetic mosaicism: generation via second polar body retention and unmasking of a likely causative PER2 variant for hypersomnia.	***Clin Epigenetics***			2021
Fuke T, Nakamura A, Inoue T, Kawashima S, Isono Hara K, Matsubara K, Sano S, Yamazawa K, Fukami M, Ogata T, Kagami M	Role of imprinting disorders in short children born SGA and Silver-Russell syndrome spectrum.	***J Clin Endocrinol Metab***	106(3)	802-813	2021
Konishi A, Ida S, Matsui F, Etani Y, **Kawai M.**	Male assignment in 5α-reductase type 2 deficiency with female external genitalia	***Pediatr Int.***		Mar 25. doi: 10.1111/ped.14447.	2021
Konishi A, Ida S, Shoji Y, Etani Y, **Kawai M**	Central hypothyroidism improves with age in very young children with Prader-Willi syndrome.	***Clin Endocrinol (Oxf).***	94	384-391.	2021

Koizumi M, Ida S, Shoji Y, Nishimoto Y, Etani Y, **Kawai M**	Visceral adipose tissue resides within the reference range in children with Prader-Willi syndrome receiving nutritional intervention on a regular basis.	***Endocr J.***	28	1029-1037	2020
Itonaga T, Koga E, Nishigaki S, **Kawai M**, Sakakibara H, Hasegawa Y.	A retrospective multicenter study of bone mineral density in adolescents and adults with Turner syndrome in Japan.	***Endocr J***	67	1023-1028	2020
Oto Y, Murakami N, Matsubara K, Saima S, Ogata H, Ihara H, Nagai T, Matsubara T	Effects of growth hormone treatment on thyroid function in pediatric patients with Prader–Willi syndrome.	***Am J Med Genet Part A***		1-5	2020
Sato T, Ishii T, Yamaguchi Y, Ichihashi Y, Ochiai D, Asanuma H, Kuroda Tatsuo, Hasegawa T.	Prenatal genetic counseling to parents of fetuses suspected of having ambiguous genitalia.	***Front Pediatr***	8	569548	2021
Ushijima K, Ogawa Y, Terao M, Asakura Y, Muroya K, Hayashi M, Ishii T, Hasegawa T, Sekido R, Fukami M, Takada S, Narumi S.	Identification of the first promoter-specific gain-of-function *SOX9* missense variant (p. E50K) in a patient with 46,XX ovotesticular disorder of sex development.	***Am J Med Genet A***	7	56–59	2021
Miyoshi Y, Yorifuji T, Shimizu C, Nagasaki K, Kawai M, Ishiguro H, Okada S, Kanno J, Takubo N, **Muroya K**, Ito J, Horikawa R, Yokoya S, Ozono K	A nationwide questionnaire survey targeting Japanese pediatric endocrinologists regarding transitional care in childhood, adolescent, and young adult cancer survivors.	***Clin Pediatr Endocrinol***	29(2)	55-62	2020
Kinjo K, Nagasaki K, **Muroya K**, Suzuki E, Ishiwata K, Nakabayashi K, Hattori A, Nagao K, Nozawa RS, Obuse C, Miyado K, Ogata T, Fukami M, Miyado M.	Rare variant of the epigenetic regulator SMCHD1 in a patient with pituitary hormone deficiency.	***Sci Rep***	10(1)	10985	2020
Kawashima S, Yagi H, Hirano Y, Toki M, Izumi K, Dateki S, Namba N, Kamimaki T, **Muroya K**, Tanaka T, Fukami M, Kagami M	Japanese SHOX study group. Screening for imprinting disorders in 58 patients with clinically diagnosed idiopathic short stature.	***J Pediatr Endocrinol Metab***	33(10)	1335-1339	2020

Mizutani A, Sabu Y, Naoi S, Ito S, Nakano S, Minowa K, Mizuochi T, Ito K, Abukawa D, Kaji S, Sasaki M, **Muroya K**, Azuma Y, Watanabe S, Oya Y, Inomata Y, Fukuda A, Kasahara M, Inui A, Takikawa H, Kusuhara H, Bessho K, Suzuki M, Togawa T, Hayashi H	Assessment of Adenosine Triphosphatase Phospholipid Transporting 8B1 (ATP8B1) Function in Patients With Cholestasis With ATP8B1 Deficiency by Using Peripheral Blood Monocyte-Derived Macrophages.	***Hepatol Commun***	5(1)	52-62	2020
Iwahashi-Odano M, Nagasaki K, Fukami M, Nishioka J, Yatsuga S, Asakura Y, Adachi M, **Muroya K**, Hasegawa T, Narumi S	Congenital Hypothyroidism Due to Truncating PAX8 Mutations: A Case Series and Molecular Function Studies.	***J Clin Endocrinol Metab***	105(11)	dgaa584	2020
Adachi M, **Muroya K**, Hanakawa J, Asakura Y	Metreleptin worked in a diabetic woman with a history of hematopoietic stem cell transplantation (HSCT) during infancy: further support for the concept of 'HSCT-associated lipodystrophy'.	***Endocr J***	EJ20-0325		
Adachi M, Tajima T, Muroya K	Dietary potassium restriction attenuates urinary sodium wasting in the generalized form of pseudohypoaldosteronism type 1.	***CEN Case Rep***	9(2)	133-137	2020
Takizaki N, Tsurusaki Y, Katsumata K, Enomoto Y, Murakami H, **Muroya K**, Ishikawa H, Aida N, Nishimura G, Kurosawa K	Novel *CUL7* biallelic mutations alter the skeletal phenotype of 3M syndrome.	***Hum Genome Var***	7	1	2020
Padidela R, Whyte MP, Glorieux FH, Munns CF, Ward LM, Nilsson O, Portale AA, Simmons JH, Namba N, Cheong HI, Pitukcheewanont P, Sochett E, Högler W, **Muroya K**, Tanaka H, Gottesman GS, Biggin A, Perwad F, Williams A, Nixon A, Sun W, Chen A, Skrinar A, Imel EA	Patient-Reported Outcomes from a Randomized, Active-Controlled, Open-Label, Phase 3 Trial of Burosumab Versus Conventional Therapy in Children with X-Linked Hypophosphatemia.	***Calcif Tissue Int.***			2021

Ushijima K, Ogawa Y, Terao M, Asakura Y, **Muroya K**, Hayashi M, Ishii T, Hasegawa T, Sekido R, Fukami M, Takada S, Narumi S	Identification of the first promoter-specific gain-of-function SOX9 missense variant (p.E50K) in a patient with 46,XX ovotesticular disorder of sex development.	***Am J Med Genet A***	185	1067-1075	2021
Masaaki Yamamoto, Genzo Iguchi, Hironori Bando, Keitaro Kanie, Ryoko Hidaka-Takeno, Hidenori Fukuoka, Yutaka Takahashi	Autoimmune Pituitary Disease: New Concepts With Clinical Implications.	***Endocrine reviews***	41(2)		2020
Yutaka Takahashi	MECHANISMS IN ENDOCRINOLOGY: Autoimmune hypopituitarism: novel mechanistic insights.	***European journal of endocrinology***	182 (4)	R59-R66	2020
Hiroshi Takagi, Shintaro Iwama, Yoshihisa Sugimura, Yutaka Takahashi, Yutaka Oki, Takashi Akamizu, Hiroshi Arima	Diagnosis and treatment of autoimmune and IgG4-related hypophysitis: clinical guidelines of the Japan Endocrine Society.	***Endocrine journal***	67(4)	373-378	2020
Kentaro Suda, Hidenori Fukuoka, Yuto Yamazaki, Katsumi Shigemura, Miki Mukai, Yukiko Odake, Ryusaku Matsumoto, Hironori Bando, Michiko Takahashi, Genzo Iguchi, Masato Fujisawa, Masahiro Oka, Katsuhiko Ono, Kazuo Chihara, Hironobu Sasano, Wataru Ogawa, Yutaka Takahashi	Cardiac Myxoma Caused by Fumarate Hydratase Gene Deletion in Patient With Cortisol-Secreting Adrenocortical Adenoma.	***The Journal of clinical endocrinology and metabolism***	105 (6)		2020
Hidenori Fukuoka, Katsumi Shigemura, Maki Kanzawa, Tomonori Kanda, Masaaki Yamamoto, Koichi Kitagawa, Mariko Sakamoto, Genzo Iguchi, Wataru Ogawa, Masato Fujisawa, Yutaka Takahashi	The impact of adrenal tumor multidisciplinary team meetings on clinical outcomes.	***Endocrine***	69(3)	519-525	2020

Maki Kanzawa, Hidenori Fukuoka, Akane Yamamoto, Kentaro Suda, Katsumi Shigemura, Shigeo Hara, Naoko Imagawa, Ryuko Tsukamoto, Yayoi Aoyama, Yasuhiro Nakamura, Masato Fujisawa, Wataru Ogawa, Yutaka Takahashi, Tomoo Itoh	Adrenal Corticomedullary Mixed Tumor Associated With the FGFR4-G388R Variant.	***Journal of the Endocrine Society***	4(9)	bvaa101	2020
Tomoko Yamada, Hidenori Fukuoka, Yusei Hosokawa, Yukiko Odake, Kenichi Yoshida, Ryusaku Matsumoto, Hironori Bando, Yuko Okada, Yushi Hirota, Genzo Iguchi, Wataru Ogawa, Yutaka Takahashi	Patients with pheochromocytoma exhibit low aldosterone renin ratio-preliminary reports.	***BMC endocrine disorders***	20(1)	140	2020
Fumio Otsuka, Yutaka Takahashi, Shigeyuki Tahara, Yoshihisa Ogawa, Michael Højby Rasmussen, Koji Takano	Similar safety and efficacy in previously treated adults with growth hormone deficiency randomized to once-weekly somapacitan or daily growth hormone.	***Clinical endocrinology***	93(5)	620-628	2020
Hidenori Fukuoka, Hiroki Shichi, Masaaki Yamamoto, Yutaka Takahashi	The Mechanisms Underlying Autonomous Adrenocorticotropic Hormone Secretion in Cushing's Disease.	***International journal of molecular sciences***	21 (23)		2020
Yasunori Fujita, Hironori Bando, Genzo Iguchi, Keiji Iida, Hitoshi Nishizawa, Keitaro Kanie, Kenichi Yoshida, Ryusaku Matsumoto, Kentaro Suda, Hidenori Fukuoka, Wataru Ogawa, Yutaka Takahashi	Clinical Heterogeneity of Acquired Idiopathic Isolated Adrenocorticotropic Hormone Deficiency.	***Frontiers in endocrinology***	12	578802-578802	2021
Ken Takeshima, Yaqiong Li, Kennichi Kakudo, Mitsuyoshi Hirokawa, Eijun Nishihara, Akira Shimatsu, Yutaka Takahashi, Takashi Akamizu	Proposal of diagnostic criteria for IgG4-related thyroid disease.	***Endocrine journal***	68(1)	1-6	2021

asuo Imanishi, Nobuaki Ito, Yumie Rhee, Yasuhiro Takeuchi, Chan Soo Shin, Yutaka Takahashi, Hiroki Onuma, Masahiro Kojima, Masanori Kanematsu, Hironori Kanda, Yoshiki Seino, Seiji Fukumoto	Interim Analysis of a Phase 2 Open-Label Trial Assessing Burosumab Efficacy and Safety in Patients With Tumor-Induced Osteomalacia.	***Journal of bone and mineral research***	36(2)	262-270	2021
Keitaro Kanie, Genzo Iguchi, Megumi Inuzuka, Kentaro Sakaki, Hironori Bando, Shin Urai, Hiroki Shichi, Yasunori Fujita, Ryusaku Matsumoto, Kentaro Suda, Masaaki Yamamoto, Hidenori Fukuoka, Takao Taniguchi, Wataru Ogawa, Yutaka Takahashi	Two Cases of anti-PIT-1 Hypophysitis Exhibited as a Form of Paraneoplastic Syndrome not Associated With Thymoma.	***Journal of the Endocrine Society***	5(3)	bvaa194	2021
Ryusaku Matsumoto, Yutaka Takahashi	Human pituitary development and application of iPSCs for pituitary disease.	***Cellular and molecular life sciences***	78(5)	2069-2079	2021
Kentaro Suda, Hidenori Fukuoka, Genzo Iguchi, Keitaro Kanie, Yasunori Fujita, Yukiko Odake, Ryusaku Matsumoto, Hironori Bando, Hiroki Ito, Michiko Takahashi, Kazuo Chihara, Hiroshi Nagai, Satoshi Narumi, Tomonobu Hasegawa, Wataru Ogawa, Yutaka Takahashi	A Case of Luscan-Lumish Syndrome: Possible Involvement of Enhanced GH Signaling.	***The Journal of clinical endocrinology and metabolism***	106 (3)	718-723	2021
Akira Shimatsu, Akinobu Nakamura, Yutaka Takahashi, Shingo Fujio, Fumitoshi Satoh, Shigeyuki Tahara, Hiroshi Nishioka, Koji Takano, Miho Yamashita, Hiroshi Arima, Atsushi Tominaga, Shohei Tateishi, Yusaku Matsushita	Preoperative and long-term efficacy and safety of lanreotide autogel in patients with thyrotropin-secreting pituitary adenoma: a multicenter, single-arm, phase 3 study in Japan.	***Endocrine journal***			2021