

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
室月淳	日常診療で妊婦・家族ときちんと向き合うための基本がわかる	室月淳(著)	出生前診断と選択的中絶のケア	メディカ出版	東京	2021	
窪田拓生, 大 菌恵一	Wnt シグナル・スクレロシンと骨系統疾患	松本俊夫 中島友紀	抗スクレロシン抗体編	メディカルレビュー社	東京	2020	70-80
鬼頭浩史	骨形成不全症	水口雅他	今日の小児治療指針17版	医学書院	東京	2020	794

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
Mishima K, <u>Kitoh H</u> , Matsushita M, Nagata T, Nishida Y, Takahashi Y, Ishiguro N	Lower limb pain following allogeneic hematological stem cell transplantation in Japanese children	J Orthop Sci	25(4)	682-687	2020
Matsushita M, Mishima K, Yamashita S, Haga N, Fujiwara S, Ozono K, Kubota T, Kitaoka T, Ishiguro N, <u>Kitoh H</u>	Impact of fracture characteristics and disease-specific complications on health-related quality of life in osteogenesis imperfecta	J Bone Miner Metab	38(1)	109-116	2020
Kaneko H, <u>Kitoh H</u> , Mishima K, Matsushita M, Hattori T, Noritake K, Ishiguro N, Yoshihashi Y	Comparison of surgical and nonsurgical containment methods for patients with Legg-Calvé-Perthes disease of the onset ages between 6.0 and 8.0 years: Salter osteotomy versus non-weight-bearing hip flexion-abduction brace	J Pediatr Orthop B	29(6)	542-549	2020
Nagata T, Matsushita M, Mishima K, Kamiya Y, Kato K, Toyama M, Ogi T, Ishiguro N, <u>Kitoh H</u>	Severe achondroplasia due to <i>de novo</i> variants in the transmembrane domain of FGFR3 on the same allele: A case report	Mol Genet Genomic Med	8(3)	e1148	2020
<u>Kitoh H</u> , Matsushita M, Mishima K, Nagata T, Kamiya Y, Ueda K, Kuwatsuka Y, Morikawa H, Nakai Y, Ishiguro N	Pharmacokinetics and safety after once and twice a day doses of meclizine hydrochloride administered to children with achondroplasia	PLoS One	15(4)	e0229639	2020
Kaneko S, Matsushita M, Mishima K, Takegami Y, Imagama S, <u>Kitoh H</u>	Effect of periosteal reaction on longitudinal bone growth in a mouse model of achondroplasia	Bone Reports	13	100708	2020
<u>Kitoh H</u>	Clinical aspects and current therapeutic approaches for FOP	Biomedicines	8(9)	325	2020
Mishima K, Mizuno S, Matsushita M, Nagata T, Kamiya Y, <u>Kitoh H</u>	Legg-Calve-Perthes disease in a patient with Bardet-Biedl syndrome -A case report of a novel <i>MKKS/BBS6</i> mutation	Clin Case Rep	00	1-6	2020
Kaneko H, <u>Kitoh H</u> , Iwata K, Mishima K, Matsushita M, Hattori T	Gradual reduction using overhead traction for developmental dysplasia of the hip after walking age: 30-year retrospective study	Int J Pediatr Orthop	6(2)	12-17	2020
Sawamura K, Mishima K, Matsushita M, Kamiya Y, <u>Kitoh H</u>	Neglected unstable slipped capital femoral epiphysis: A case report	Acta Scientific Orthopaedics	3(12)	87-90	2020

Savarirayan R, Tofts L, Irving M, Wilcox W, Bacino CA, Hoover-Fong J, Ullot Font R, Harmatz P, Rutsch F, Bober MB, Polgreen LE, Ginebreda I, Mohnike K, Charrow J, Hoernschmeyer D, <u>Ozono K</u> , Alanay Y, Arundel P, Kagami S, Yasui N, White KK, Saal HM, Leiva-Gea A, Luna-González F, Mochizuki H, Basel D, Porco DM, Jayaram K, Fischeleva E, Huntsman-Labed A, Day J	Once-daily, subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial	Lancet	396(10252)	684-692	2020
Matsuda N, Takasawa K, Ohata Y, Takishima S, Kubota T, Ishihara Y, Fujiwara M, Ogawa E, Morio T, Kashimada K, Ozono K	Potential pathological role of single nucleotide polymorphism (c.787T>C) in alkaline phosphatase (ALPL) for the phenotypes of hypophosphatasia	Endocr J	67(12)	1227-1232	2020
Takeyari S, Kubota T, Ohata Y, Fujiwara M, Kitaoka T, Taga Y, Mizuno K, Ozono K	4-phenylbutyric acid enhances the mineralization of osteogenesis imperfecta iPSC-derived osteoblasts	J Biol Chem	296	100027	2020
Fujisawa Y, Kitaoka T, Ono H, Nakashima S, Ozono K, Ogata T	Case Report: Efficacy of Reduced Doses of Asfotase Alfa Replacement Therapy in an Infant with Hypophosphatasia Who Lacked Severe Clinical Symptoms	Front Endocrinol (Lausanne)	18 December 2020	doi.org/10.3389/fendo.2020.590455	2020
Io S, Watanabe A, Yamada S, Mandai M, <u>Yamada T</u>	Perinatal benign hypophosphatasia antenatally diagnosed through measurements of parental serum alkaline phosphatase and ultrasonography	Congenit Anom (Kyoto)	2020 May 11	doi: 10.1111/cga.12374. Epub 2020 Jun 16. PMID 32390219	2020
Kawasaki H, <u>Yamada T*</u> , Wada T, Kosugi S	Current status and legal/ethical problems in the research use of the tissues of aborted human fetuses in Japan	Congenit Anom (Kyoto)	60	166-174	2020
Tsuchiya M, <u>Yamada T*</u> , Akaishi R, Hamanoue H, Hirasawa A, Hyodo M, Imoto I, Kosho T, Kurosawa K, Murakami H, Nakatani K, Nomura F, Sasaki A, Shimizu K, Tamai M, Umemura H, Watanabe A, Yoshida A, Yoshihashi H, Yotsumoto J, Kosugi S	Attitudes toward and current status of disclosure of secondary findings from next-generation sequencing: A nation-wide survey of clinical genetics professionals in Japan	J Hum Genet	65	1045-1053	2020

Kawasaki H, <u>Yamada T*</u> , Takahashi Y, Nakayama T, Wada T, Kosugi S	Epidemiology of birth defects in very low birth weight infants in Japan	J Pediatr	2020 Jul 4:S0022-3476(20)30855-6	doi: 10.1016/j.jpeds.2020.07.012. Online ahead of print. PMID:32634406	2020
Hayashi H, Tanishima S, Fujii K, Mori R, Okada C, Yanagita E, Shibata Y, Matsuoka R, Amano T, Yamada T, Yabe I, Kinoshita I, Komatsu Y, Akita H, Nishihara H	Clinical impact of a cancer genomic profiling test using an in-house comprehensive targeted sequencing system	Cancer Sci	2020 Aug 8	doi: 10.1111/cas.14608. Epub ahead of print. PMID: 32772458	2020
Kawasaki H, <u>Yamada T*</u> , Takahashi Y, Nakayama T, Wada T, Kosugi S, The Neonatal Research Network of Japan	The short-term mortality and morbidity of very low birth weight infants with trisomy 18 or trisomy 13 in Japan	J Hum Genet	66	273-285, 2020. PMID: 32943740	2020
Kondo T, Matsubara J, Pham Nguyen, Quy PN, Fukuyama K, Nomura M, Funakoshi T, Doi K, Sakamori Y, Yoshioka M, Yokoyama A, Tamaoki M, Kou T, Hirohashi K, Yamada A, Yamamoto Y, Minamiguchi S, Nishigaki M, <u>Yamada T</u> , Kanai M, Matsumoto S, Muto M	Comprehensive genomic profiling for patients with chemotherapy-naïve advanced cancer	Cancer Sci	2020 Oct 2	doi: 10.1111/cas.14674. Epub ahead of print. PMID: 33007138	2020
Suzumori N, Sekizawa A, Takeda E, Samura O, Sasaki A, Akaishi R, Wada S, Hamanoue H, Hirahara F, Sawai H, Nakamura H, <u>Yamada T</u> , Miura K, Masuzaki H, Nakayama S, Kamei Y, Namba A, Murotsuki J, Yamaguchi M, Tairaku S, Maeda K, Kaji T, Okamoto Y, Endo M, Ogawa M, Kasai Y, Ichizuka K, Yamada N, Ida A, Miharu N, Kawaguchi S, Hasuo Y, Okazaki T, Ichikawa M, Izumi S, Kuno N, Yotsumoto J, Nishiyama M, Shirato N, Hirose T, Sago H	Retrospective details of false-positive and false-negative results in noninvasive prenatal testing for fetal trisomy 21, 18 and 13	Eur J Obstet Gynecol Reprod Biol	2020 Oct 27;256:75-81	doi: 10.1016/j.ejogrb.2020.10.050. Epub ahead of print. PMID: 33171421	2020
Umazume T, <u>Yamada T</u> , Furuta I, Iwano H, Morikawa M, Watari H, Minakami H	Morphofunctional Cardiac Changes in Singleton and Twin Pregnancies: A Longitudinal Cohort Study	BMC Pregnancy and Childbirth	in press	in press	2020
佐々木佑菜, <u>山田崇弘</u> , 小杉眞司	ビスホスホネート製剤導入が骨形成不全症罹患児の両親に与えた影響の調査：質的研究の統合	周産期医学	in press	in press	2020

Nagaoka S, Yamaguchi-Kabata Y, Murotsuki J, et al:	Estimation of the carrier frequencies and proportions of potential patients by detecting causative gene variants associated with autosomal recessive bone dysplasia using a whole-genome reference panel of Japanese individuals	Hum Genome Var	2021 Jan 15;8(1):2	doi: 10.1038/s41439-020-0133-7	2021
Kato M, Michigami T, Tachikawa K, Kato M, Yabe I, Shimizu T, Asaka T, Kitagawa Y, Atsumi T	Novel mutation in the ALPL gene with a dominant negative effect in a Japanese family	<i>J Bone Miner Metab,</i>	2021, Apr 5	doi: 10.1007/s00774-021-01219-0. Online ahead of print	2021
芳賀信彦	小児希少疾患のリハビリテーション診療（教育講座）	Jpn J Rehabil Med	57(4)	334-339	2020.4
芳賀信彦	成人後を見据えた小児骨系統疾患の診療	日整会誌	94(4)	248-254	2020.4
Matsuoka M, Tsukamoto S, Orihara Y, Kawamura R, Kuratani M, Haga N, Ikebuchi K, Katagiri T*	Design of primers for direct sequencing of nine coding exons in the human ACVR1 gene	Bone	138	115469	2020
Pignolo RJ, Cheung K, Kile S, Fitzpatrick MA, De Cunto C, Al Mukaddam M, Hsiao EC, Baujat G, Delai P, Eekhoff EMW, Di Rocco M, Grunwald Z, Haga N, Keen R, Levi B, Morhart R, Scott C, Sherman A, Zhang K, Kaplan FS	Self-reported baseline phenotypes from the International Fibrodysplasia Ossificans Progressiva (FOP) Association Global Registry	Bone	134	115274	2020
宮寄 治	第56回日本小児放射線学会学術集会“新時代の小児診療，360度の評価をめざして”より胎児と新生児の骨疾患：診断の決め手となるkey findingの指摘	日本小児放射線学会誌	37	25-33	2021