別添4

研究成果の刊行に関する一覧表

書籍

令和元年度

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
芳賀信彦	タナトフォリック骨異形成症［指定難病275］	水澤英洋他	日本医師会雑誌 148(特別号1：指定難病ペディア2019)	日本医師会	日本	2019	S328
芳賀信彦	先天性骨系統疾患	井樋栄二、吉川秀樹、津村弘、田中栄、高木理彰、編集	標準整形外科学、第14版	医学書院	東京	2020	294-309
鬼頭浩史	骨・関節系統疾患	出家正隆他	TEXT整形外科学 改訂5版	南江堂	東京	2019	191-201
大幡泰久，窪田拓生，大薗恵一	低ホスファターゼ症	竹内 靖博，杉本 利嗣，成瀬 光栄	副甲状腺・骨代謝疾患診療マニュアル 改訂第2版	診断と治療社	日本	2019	197-200
大薗恵一	低ホスファターゼ症	牛島俊和他	遺伝子医学，10(1)	メディカルドゥ	日本	2020	92-103
大薗恵一，窪田拓生	軟骨無形成症診療ガイドラインについて教えてください	監修 田中敏章、田島敏広	成長障害診療Q&A	novo nordisk	日本	2020	なし
大薗恵一（司会）Lothar Seefried，鬼頭浩史，小山博之，福士純一，道上敏美	成人HPP患者の特徴と理解を深める	大薗恵一監修、道上敏美 鬼頭浩史 窪田拓生 澤井英明 仲野和彦 編集	HPP Frontier,3	メディカルレビュー社	日本	2020	5-11

窪田拓生	低ホスファターゼ症患者における臨床症状の発生頻度と年齢：系統的文献レビュー	大薗恵一監修、道上敏美 鬼頭浩史 窪田拓生 澤井英明 仲野和彦 編集	HPP Frontier, 3	メディカルレビュー社	日本	2020	44-45
山田崇弘，佐村修	８．性染色体数的異常	関沢明彦，佐村修，四元淳子　編著	周産期遺伝カウンセリングマニュアル　改訂3版	中外医学社	東京	2020	101-108

令和2年度

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，大薗恵一	Wnt シグナル・スクレロスチンと骨系統疾患	松本俊夫 中島友紀	抗スクレロスチン抗体編	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

令和3年度

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
窪田拓生	骨密度	日本小児内分泌学会編	小児内分泌学	診断と治療社	東京	2021	67-69
芳賀信彦	骨形成不全症	土屋弘行ほか	今日の整形外科治療指針	医学書院	東京	2021	267-269

鬼頭浩史	2型コラーゲングループおよび類似疾患概説	土屋弘行他編集	今日の整形外科治療指針第8版	医学書院	東京	2021	244
鬼頭浩史	先天性脊椎骨端異形成症	土屋弘行他編集	今日の整形外科治療指針第8版	医学書院	東京	2021	245
鬼頭浩史	Kniest骨異形成症	土屋弘行他編集	今日の整形外科治療指針第8版	医学書院	東京	2021	246
鬼頭浩史	Stickler症候群	土屋弘行他編集	今日の整形外科治療指針第8版	医学書院	東京	2021	247
鬼頭浩史	Schmid型骨幹端異形成症	土屋弘行他編集	今日の整形外科治療指針第8版	医学書院	東京	2021	252
大薗恵一	FGFR3（関連疾患：軟骨無形成症）		小児科診療第84巻（第11特大号）			2021	p305-307
大幡泰久	軟骨無形成症・低形成症		日本小児内分泌学会小児内分泌学　改訂第3版	診断と治療社	東京	2021	206-210
窪田拓生	骨密度	日本小児内分泌学会編	日本小児内分泌学会小児内分泌学　改訂第3版	診断と治療社	東京	2022	67-69
藤原誠	骨系統疾患	臨床遺伝専門医制度委員会 編	臨床遺伝専門医テキスト③　各論Ⅱ臨床遺伝学小児領域	診断と治療社	東京	2021	94-99
窪田拓生	軟骨異栄養症とGH治療		糖尿病・内分泌代謝科，53(2)			2021	187-193
道上敏美	低ホスファターゼ症	遠山正彌、馬場明道、土井健史編	薬剤師が知っておきたい病気と薬剤のはなし	金芳堂	京都	2021	219-224

雑誌

令和元年度

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
芳賀信彦	小児骨系統疾患・全身疾患の診かた	Loco Cure	5(3) (特集：小児運動器疾患の診かた)	232-236	2019
芳賀信彦	骨関節疾患の移行期医療	J Clin Rehabil	28 (特集・小児慢性疾患の移行期医療とリハビリテーション)	1274-1279	2019
Matsushita M, Kitoh H, Mishima K, Yamashita S, Haga N, Fujiwara S, Ozono K, Kubota T, Kitaoka T, Ishiguro N	Physical, mental and social problems of adolescent and adult patients with achondroplasia	Calc Tissue Int	104(4)	364-372	2019
Nakahara Y, Kitoh H, Nakashima Y, Toguchida J, Haga N	Longitudinal study of the activities of daily living and quality of life in Japanese patients with fibrodysplasia ossificans progressiva	Disabil Rehabil	41(6)	699-704	2019
Haga N, Nakashima Y, Kitoh H, Kamizono J, Katagiri T, Saijo H, Tsuakamoto S, Shinoda Y, Sawada R, Nakahara Y	Fibrodysplasia ossificans progressiva: Review and research activities in Japan	Pediatr Int	62	3-13	2020
Matsushita M, Mishima K, Yamashita S, Haga N, Fujiwara S, Ozono K, Kubota T, Kitaoka T, Ishiguro N, Kitoh H	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
Nagata T, Matsushita M, Mishima K, Kamiya Y, Kato K, Toyama M, Ogi T, Ishiguro N, Kitoh H	Severe achondroplasia due to *de novo* variants in the transmembrane domain of FGFR3 on the same allele: A case report	Mol Genet Genomic Med	8(3)	e1148	2020
Kitoh H, 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

Mishima K, Kitoh H, 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	(Epub ahead of print)	(Epub ahead of print)	
Kaneko H, Kitoh H, 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	(Epub ahead of print)	(Epub ahead of print)	
Michigami T, Ohata Y, Fujiwara M, Mochizuki H, Adachi M, Kitaoka T, Kubota T, Sawai H, Namba N, Hasegawa K, Fujiwara I, Ozono K	Clinical Practice Guidelines for Hypophosphatasia.	Clin Pediatr Endocrinol	29(1)	9- 24	2020
Michigami T, Tachikawa K, Yamazaki M, Kawai M, Kubota T, Ozono K	Hypophosphatasia in Japan: *ALPL* mutation analysis in 98 unrelated patients	Calcif Tissue Int	106(3)	221- 231	2020
Rassie K, Dray M, Michigami T, Cundy T	Bisphosphonate use and fractures in adults with hypophosphtasia	JBMR Plus	3(10)	e10223	2019
Ishiguro T, Sugiyama Y, Ueda K, Muramatsu Y, Tsuda H, Kotani T, Michigami T, Tachikawa K, Akiyama T, Hayakawa M	Findings of amplitude-integrated electroencephalogram recordings and serum vitamin B6 metabolites in perinatal lethal hypophosphatasia during enzyme replacement therapy	Brain Dev	41(8)	721-725	2019
Okawa R, Kokomoto K, Kitaoka T, Kubota T, Watanabe A, Taketani T, Michigami T, Ozono K, Nakano K	Japanese nationwide survey of hypophosphtasia reveals prominent differences in genetic and dental findings between odonto and non-odonto types	PLoS One	14(10)	e0222931	2019
Yamamoto K, Kawai M, Yamazaki M, Tachikawa K, Kubota T, Ozono K, Michigami T	CREB activation in hypertrophic chondrocytes is involved in the skeletal overgrowth in epiphyseal chondrodysplasia Miura type caused by activating mutations of natriuretic peptide receptor B	Hum Mol Genet	28(7)	1183-1198	2019

Ohata Y, Takeyari S, Nakano Y, Kitaoka T, Nakayama H, Bizaoui V, Yamamoto K, Miyata K, Yamamoto K, Fujiwara M, Kubota T, Michigami T, Yamamoto K, Yamamoto T, Namba N, Ebina K, Yoshikawa H, Ozono K	Comprehensive genetic analyses using targeted next-generation sequencing and genotype-phenotype correlations in 53 Japanese patients with osteogenesis imperfecta	Osteoporos Int	30(11)	2333-2342	2019
Nakano C, Kitabatake Y, Takeyari S, Ohata Y, Kubota T, Taketani K, Kogo M, Ozono K	Genetic correction of induced pluripotent stem cells mediated by transcription activator-like effector nucleases targeting ALPL recovers enzyme activity and calcification in vitro	Mol Genet Metab	127(2)	158-165	2019
Kubota T, Adachi M, Kitaoka T, Hasegawa K, Ohata Y, Fujiwara M, Michigami T, Mochizuki H, Ozono K	Clinical practice guidelines for achondroplasia	Clin Pediatr Endocrinol	29(1)	25-42	2020
Yamamoto K, Kubota T, Takeyari S, Kitaoka T, Miyata K, Nakano Y, Nakayama H, Ohata Y, Yanagi K, Kaname T, Okada Y, Ozono K	Parental somatogonadal COL2A1 mosaicism contributes to intrafamilial recurrence in a family with type 2 collagenopathy	Am J Med Genet A	182(3)	454-460	2020
山田崇弘	質疑応答　プロからプロへ. 骨系統疾患の出生前診断の行方は？	日本医事新報	4995	53	2020
Kawabata K, Morikawa M, Ishikawa S, Nakagawa K, Chiba K, Yamada T, Saito Y, Akimoto T, Cho K, Minakami H	Fetal middle cerebral artery peak systolic velocity as a predictor of fetal anemia in unselected women giving birth at or near term	Taiwan J Obstet Gynecol	58	212-217	2019
Taguchi I, Yamada T*, Akaishi R, Imoto I, Kurosawa K, Nakatani K, Nomura F, Hamanoue H, Hyodo M, Murakami H, Yoshihashi H, Yotsumoto J, Kosugi S	Attitudes of clinical geneticists and certified genetic counselors to genome editing and its clinical applications: A nation-wide questionnaire survey in Japan	J Hum Genet	64	945-954	2019
Yamaguchi Y, Tayama C, Tomikawa J, Akaishi R, Kamura H, Matsuoka K, Wake N, Minakami H, Kato K, Yamada T, Nakabayashi K, Hata K	Placenta-specific epimutation at H19-DMR among common pregnancy complications: its frequency and effect on the expression patterns of H19 and IGF2	Clinical Epigenetics	11	113	2019

Yamamoto Y, Kanai M, Kou T, Sugiyama A, Nakamura E, Miyake H, Yamada T, Nishigaki M, Kondo T, Murakami H, Torishima M, Matsumoto S, Kosugi S, Muto M	Clinical significance of TP53 variants as possible secondary findings in tumor-only next-generation sequencing	J Hum Genet	65	125-132	2020 (IF=2.912)
Nagata M, Setoh K, Takahashi M, Higasa K, Kawaguchi T, Kawasaki H, Wada T, Watanabe A, Sawai H, Tabara Y, Yamada T*, Matsuda F, Kosugi S	Association of ALPL variants with serum alkaline phosphatase and bone traits in the general Japanese population: The Nagahama Study	J Hum Genet	65	337-343	2020
Sato T, Kojima T, Samura O, Kawaguchi S, Nakamura A, Nakajima M, Tanuma-Takahashi A, Nakabayashi K, Hata K, Ikegawa S, Nishimura G, Okamoto A, Yamada T*	Two Unrelated Pedigrees with Achondrogenesis Type 1b Carrying a Japan-specific Pathogenic Variant in SLC26A2	Am J Med Genet A	182	735-739	2020
Yotsumoto J, Sekizawa A, Inoue S, Suzumori N, Samura O, Yamada T, Miura K, Masuzaki H, Sawai H, Murotsuki J, Hamanoue H, Kamei Y, Endo T, Fukushima A, Katagiri Y, Takeshita N, Ogawa M, Nishizawa H, Okamoto Y, Tairaku S, Kaji T, Maeda K, Matsubara K, Ogawa M, Osada H, Ohba T, Kawano Y, Sasaki A, Sago H	Qualitative investigation of the factors that generate ambivalent feelings in women who give birth after receiving negative results from non-invasive prenatal testing	BMC Pregnancy and Childbirth	20	112	2020
Io S, Watanabe A, Yamada S, Mandai M, Yamada T	Perinatal benign hypophosphatasia antenatally diagnosed through measurements of parental serum alkaline phosphatase and ultrasonography	Congenit Anom (Kyoto)	in press	in press	2020
Kitano A, Nakaguro M, Tomotaki S, Hanaoka S, Kawai M, Saito A, Hayakawa M, Takahashi Y, Kawasaki H, Yamada T, Ikeda M, Onda T, Cho K, Haga H, Nakazawa A, Minamiguchi S	A familial case of alveolar capillary dysplasia with misalignment of the pulmonary veins: The clinicopathological features and unusual glomeruloid endothelial proliferation	Diagnostic Pathology	15	48	2020

令和2年度

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, Kitoh H, 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, Kitoh H	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, Kitoh H, 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, Kitoh H	Severe achondroplasia due to *de novo* variants in the transmembrane domain of FGFR3 on the same allele: A case report	Mol Genet Genomic Med	8(3)	e1148	2020
Kitoh H, 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, Kitoh H	Effect of periosteal reaction on longitudinal bone growth in a mouse model of achondroplasia	Bone Reports	13	100708	2020
Kitoh H	Clinical aspects and current therapeutic approaches for FOP	Biomedicines	8(9)	325	2020
Mishima K, Mizuno S, Matsushita M, Nagata T, Kamiya Y, Kitoh H	Legg-Calve-Perthes disease in a patient with Bardet-Biedl syndrome -A case report of a novel *MKKS/BBS6* mutation	Clin Case Rep	00	1-6	2020

Kaneko H, Kitoh H, 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, Kitoh H	Neglected unstable slipped capital femoral epiphysis: A case report	Acta Scientfic 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, Ozono K, 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, Fisheleva 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, Yamada T	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, Yamada T*, 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, Yamada T*, 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, Yamada T*, 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, Yamada T*, 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, Yamada T, 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, Yamada T, 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, Yamada T, 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 2020	in press	in press	2020
佐々木佑菜, 山田崇弘, 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/s41439-020-00133-7	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	*J Bone Miner Metab,*	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
宮嵜　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

令和3年度

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Ushioda M, Sawai H, Numabe H, Nishimura G, Shibahara H.	Development of individuals with thanatophoric dysplasia surviving beyond infancy.	Pediatr Int.	Oct 1;64(1):	e15007.	2021

Mano H, Fujiwara S, Takamura K, Kitoh H, Takayama S, Ogata T, Haga N:	Treatment approaches for congenital transverse limb deficiency: Data analysis from an epidemiological nation survey in Japan.	J Orthop Sci	26:,	650-654	2021
Mano H, Fujiwara S, Haga N:	How children with congenital limb deficiencies visually attend to their limbs and prostheses: eye tracking of displayed still images and visuospatial body knowledge.	Dev Neurorehab	24(8);	547-554,	2021
芳賀信彦	: 小児整形の臨床-総論（連載:小児運動器難病に対する新しい治療）.	整形外科	72:	466-469,	2021
芳賀信彦	: 成人骨形成不全症患者運動器障害と診療.	日本遺伝カウンセリング学会誌	42(1)	: 57-61,	2021
Matsushita M, Mishima K, Nagata T, Kamiya Y, Imagama S, Kitoh H.	Asfotase alfa has a limited effect in improving the bowed limbs in perinatal benign hypophosphatasia: A case report.	Clin Pediatr Endocrinol	30(1)	:53-56,	2021
Matsushita M, Mishima K, Kamiya Y, Haga N, Fujiwara S, Ozono K, Kubota T, Kitaoka T, Imagama S, Kitoh H.	Health-related quality of life in adult patients with multiple epiphyseal dysplasia and spondyloepiphyseal dysplasia.	Prog Rehabil Med	6:	20210048,	2021

Kitoh H, Matsushita M, Mishima K, Kamiya Y, Sawamura K.	Disease-specific complications and multidisciplinary interventions in achondroplasia.	J Bone Miner Metab	40(2)	:189-195,	2022
Sawamura K, Mishima K, Matsushita M, Kamiya Y, Kitoh H.	A cross-sectional nationwide survey of osteosclerotic skeletal dysplasia in Japan.	J Orthop Sci	(Online ahead of print)		2021
Ajimi A, Matsushita M, Mishima K, Haga N, Fujiwara S,	Inconvenience and adaptation in Japanese adult achondroplas	Clin Pediatr Endocrinol	31(1)	:18-24,	2022
Ozono K, Kubota T, Kitaoka T, Imagama S, Kitoh H.	ia and hypochondroplasia: A cross-sectional study				
Savarirayan R, Ireland P, Ozono K, et al.	International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia.	Nature Reviews Endocrinology,	18(3) :	173-189,	2021.
R. Savarirayan, L. Tofts, Ozono K,	Safe and persistent growth-promoting effects of vosoritide in children with achondroplasia: 2-year results from an open-label, phase 3 extension study.	Genetics in Medicine	47, 2021	23 : 2443- 24	2021

Hoover-Fong J, Cheung MS, Fano V, Hagenas L, Hecht JT , Ireland P, Irving M, Mohnike K, Offiah AC, Okenfuss E, Ozono K, Raggio C, Tofts L, Kelly D, Shediac R, Pan W, Savarirayan R.	Lifetime impact of achondroplasia: Current evidence and perspectives on the natural history.	Bone,	146,	115872,	2021.
Ajimi A, Matsushita M, Mishima K, Haga N, Fujiwara S, Ozono K, Kubota T, Kitaoka T, Imagama S, Kitoh H.	Inconvenience and adaptation in Japanese adult achondroplasia and hypochondroplasia: A cross-sectional study.	Clinical Pediatric Endocrinology,	31(1)	: 18-24,	2022.
宮嵜　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.	日本小児放射治線学会誌、	37,	25-33,	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.	J Bone Miner Metab,	39:	804- 809,	2021
佐々木佑菜, 山田崇弘, 小杉眞司. 1072,2021	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.	51:	1067-1072	2021