別紙４

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
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

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
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, Mursotsuki 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