

. 研究成果の刊行に関する一覧表

書籍

無し

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
Yamashita A, Morioka M, Kishi H, Kimura T, Yahara Y, Okada M, Fujita K, Sawai H, Ikegawa S, Tsumaki N.	Statin treatment rescues FGFR3 skeletal dysplasia phenotypes.	Nature.	513(7 519):	507-11.	2014
Miyazaki O, Sawai H, Murotsuki J, Nishimura G, Horiuchi T.	Nationwide radiation dose survey of computed tomography for fetal skeletal dysplasias.	Pediatr Radiol.	44(8):	971-9.	2014
Okada M, Ikegawa S, Morioka M, Yamashita A, Saito A, Sawai H, Murotsuki J, Ohashi H, Okamoto T, Nishimura G, Imaizumi K, Tsumaki N.	Modeling type II collagenopathy skeletal dysplasia by directed conversion and induced pluripotent stem cells.	Hum Mol Genet.	24(2):	299-313.	2014
高田雅代, 渡邊淳, 澤井英明, 丸山秀彦, 塚原紗耶, 渋川昇平, 片山典子, 立石洋子, 熊澤一真, 中西美恵, 多田克彦, 森茂弘, 森田啓督, 山邊陽子, 中村信, 影山操 .	出生前に超音波検査により疑われ, 出生後の遺伝子解析で確定診断した周産期型低フォスファターゼ症の1例 .	日本周産期・新生児医学会雑誌	50:	362 -367.	2014

Kaga A, Murotsuki J, Kamimura M, Kimura M, Saito-Hakoda A, Kanno J, Hoshi K, Kure S, Fujiwara I.	Association of achondroplasia with Down syndrome: difficulty in prenatal diagnosis by sonographic and 3-D helical computed tomographic analyses.	Congenit Anom (Kyoto)	epub	epub	2014
Imai R, Miyazaki O, Horiuchi T, Kurosawa H, Nosaka S.	Local diagnostic reference level (DRL) based on size-specific dose estimates (SSDE): Assessment of pediatric abdominal/pelvic computed tomography (CT) at a Japanese national children's hospital	Pediatr Radiol	Epub	Epub	2014
Nagata E, Kano H, Kato F, Yamaguchi R, Nakashima S, Takayama S, Kosaki R, Tonoki H, Mizuno S, Watanabe S, Yoshiura K, Kosho T, Hasegawa T, Kimizuka M, Suzuki A, Shimizu K, Ohashi H, Haga N, Numabe H, Horii E, Nagai T, Yoshihashi H, Nishimura G, Toda T, Takada S, Yokoyama S, Asahara H, Sano S, Fukami M, Ikegawa S, Ogata T	Japanese founder duplications/triplications involving BHLHA9 are associated with split-hand/foot malformation with or without long bone deficiency and Gollop-Wolfgang complex.,	Orphanet J Rare Dis	9	125	2014
Mishima K, Kitoh H, Haga N, Nakashima Y, Kamizono J, Katagiri T, Susami T, Matsushita M, Ishiguro N:	Radiographic characteristics of the hand and cervical spine in fibrodysplasia ossificans progressiva.	Intractable Rare Dis Res	3	46-51	2014,

Matsushita M, Hasegawa S, Kitoh H, Mori K, Ohkawara B, Yasoda A, Masuda A, Ishiguro N, Ohno K.	Meclozine promotes longitudinal skeletal growth in transgenic mice with achondroplasia carrying a gain-of-function mutation in the FGFR3 gene.	Endocrinology	Epub	Epub	2014
Olney RC, Prickett TCR, Espiner EA, Mackenzie WG, Duker A, Ditro C, Zabel B, Hasegawa T, Kitoh H, Aylsworth AS, Bober MB.	C-type natriuretic peptide (CNP) plasma levels are elevated in subjects with achondroplasia, hypochondroplasia, and thanatophoric dysplasia.	J Clin Endocrinol Metab	Epub	Epub	2014
Matsushita M, Kitoh H, Subasioglu A, Colak FK, Dundar M, Mishima K, Nishida Y, Ishiguro N.	A glutamine repeat variant of the RUNX2 gene causes cleidocranial dysplasia.	Mol Syndromol	in press	in press	
Kitoh H, Kaneko H, Mishima K, Matsushita M, Kadono I, Nishida Y, Ishiguro N.	Early and late fracture following extensive limb lengthening in achondroplasia and hypochondroplasia	Bone Joint J	96-B: 273	1269-1	2014
Hoover-Fong J, Sobreira N, Jurgens J, Modaff P, Blout C, Moser A, Kim OH, Cho TJ, Cho SY, Kim SJ, Jin DK, Kitoh H, Park WY, Ling H, Hetrick KN, Doheny KF, Valle D, Pauli RM.	Mutations in PCYT1A, encoding a key regulator of phosphatidylcholine metabolism, cause spondylometaphyseal dysplasia with cone-rod dystrophy	Am J Hum Genet	94 2	105-11	2014

Matsushita M, Kitoh H, Kaneko H, Mishima K, Itoh Y, Tokita Y, Ishiguro N.	A novel in-flame deletion of the RUNX2 gene causes a classic form of cleidocranial dysplasia	J Bone Miner Metab	32	96-99	2014
Matsushita M, Kitoh H, Michigami T, Tachikawa K, Kaneko H, Mishima K, Ishiguro N.	Benign prenatal hypophosphatasia: a treatable disease not to be missed.	Ped Radiol	44	340-343	2014
Watanabe A*, Satoh S, Fujita A, Naing BT, Orimo H, Shimada T.	Perinatal hypophosphatasia caused by uniparental isodisomy.	Bone.	60	93-97	2014
Miyagawa K, Yamazaki M, Kawai M, Nishino J, Koshimizu T, Ohata Y, Tachikawa K, Mikuni-Takagaki Y, Kogo M, Ozono K, Michigami T.	Dysregulated gene expression in primary osteoblasts and osteocytes isolated from hypophosphatemic Hyp mice.	PLoS ONE	9(4)	e93840	2014
Kawai M, Kinoshita S, Shimba S, Ozono K, Michigami T.	Sympathetic activation induces skeletal Fgf23 expression in a circadian rhythm-dependent manner	J Biol Chem,	289:	1457-1466	2014
Yamazaki M, Kawai M, Miyagawa K, Ohata Y, Tachikawa K, Kinoshita S, Nishino J, Ozono K, Michigami T.	Interleukin-1-induced acute bone resorption facilitates the secretion of fibroblast growth factor 23 into the circulation.	J Bone Miner Metab	Epub	Epub	2014
Takeyari S, Yamamoto T, Kinoshita Y, Fukumoto S, Glorieux FH, Michigami T, Hasegawa K, Kitaoka T, Kubota T, Imanishi Y, Shimotsuji T, Ozono K	.Hypophosphatemic osteomalacia and bone sclerosis caused by a novel homozygous mutation of the FAM20C gene in an elderly man with a mild variant of Raine syndrome., ,	Bone	67C:	56-62	2014

Kubota T, Kitaoka T, Miura K, Fujiwara M, Ohata Y, Miyoshi Y, Yamamoto K, Takeyari S, Yamamoto T, Namba N, Ozono K.	Serum fibroblast growth factor 23 is a useful marker to distinguish vitamin D-deficient rickets from hypophosphatemic rickets.	Horm Res Paediatr	81(4)	251-7	2014
Ozono K, Hasegawa Y, Minagawa M, Adachi M, Namba N, Kazukawa I, Kitaoka T, Asakura Y, Shimura A, Naito Y.	Therapeutic use of oral sodium phosphate (phosribbon®) combination granules) in hereditary hypophosphatemic rickets.	Clin Pediatr Endocrinol,	23(1)	9-15	2014
Kitaoka T, Miyoshi Y, Namba N, Miura K, Kubota T, Ohata Y, Fujiwara M, Takagi M, Hasegawa T, Jüppner H, Ozono K.	Two Japanese familial cases of Caffey disease with and without the common COL1A1 mutation and normal bone density, and review of the literature	Eur J Pediatr,	173(6)	799-804	2014
Matsushita M, Hasegawa S, Kitoh H, Mori K, Ohkawara B, Yasoda A, Masuda A, Ishiguro N, Ohno K.	Meclozine promotes longitudinal skeletal growth in transgenic mice with achondroplasia carrying a gain-of-function mutation in the FGFR3 gene.	Endocrinology.	156(2)	548-54	2015
Matsushita M, Kitoh H, Mishima K, Nishida Y, Ishiguro N.	A case of severe proximal focal femoral deficiency with overlapping phenotypes of Al-Awadi-Raas-Rothschild syndrome and Fuhrmann syndrome.	Pediatr Radiol	44(12)	1617-9	2014
Miura K, Kim OH, Lee HR, Namba N, Michigami T, Yoo WJ, Choi IH, Ozono K, Cho TJ	Overgrowth syndrome associated with a gain-of-function mutation of the natriuretic peptide receptor 2 (NPR2) gene	Am J Med Genet A.	164A(1)	156-63	2014

Kuroyanagi Y, Kawasaki H, Noda Y, Ohmachi T, Sekiya S, Yoshimura K, Ohe C, Michigami T, Ozono K, Kaneko K.	A fatal case of infantile malignant osteopetrosis complicated by pulmonary arterial hypertension after hematopoietic stem cell transplantation.	Tohoku J Exp Med.	234(4):	309-12.	2014
大薗恵一	骨格徵候を伴う、過成長症候群	PAS/ASPR Joint Meeting Report		8	2014
大薗恵一	軟骨無形成症	小児科診療	77 5	613-61 5	2014

・研究成果の刊行物・別冊