

原著 (英文)

●平成26年度 (2014.4~2015.3) ●

●1) Dysregulated gene expression in primary osteoblasts and osteocytes isolated from hypophosphatemic Hyp mice.

Miyagawa K※, Yamazaki M※, Kawai M※, Nishino J※, Koshimizu T※, Ohata Y, Tachikawa K※, Mikuni-Takagaki Y※, Kogo M※, Ozono K, Michigami T※
PLoS One, 9 (4) : e93840, 2014.

●2) Elevated fibroblast growth factor 23 exerts its effects on placenta and regulates vitamin D metabolism in pregnancy of Hyp mice.

Ohata Y, Yamazaki M※, Kawai M※, Tsugawa N※, Tachikawa K※, Koinuma T※, Miyagawa K※, Kimoto A※, Nakayama M※, Namba N, Yamamoto H※, Okano T※, Ozono K, Michigami T※
J Bone Miner Res, 29 (7) : 1627-1638, 2014.

●3) Sympathetic activation induces skeletal Fgf23 expression in a circadian rhythm-dependent manner.

Kawai M※, Kinoshita S※, Shimba S※, Ozono K, Michigami T※
J Biol Chem, 289 : 1457-1466, 2014.

●4) Overgrowth syndrome associated with a gain-of-function mutation of the natriuretic peptidoreceptor 2 (NPR2) gene.

Miura K※, Kim OH※, Lee HR※, Namba N, Michigami T※, Yoo WJ※, Choi H※, Ozono K, Cho TJ※
Am J Med Genet A, 164A : 156-163, 2014.

●5) Interleukin-1-induced acute bone resorption facilitates the secretion of fibroblast growth factor 23 into the circulation.

Yamazaki M※, Kawai M※, Miyagawa K※, Ohata Y, Tachikawa K※, Kinoshita S※, Nishino J※, Ozono K, Michigami T※
J Bone Miner Metab, 33 : 342-354, 2014.

●6) 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.

Takeyari S, Yamamoto T※, Kinoshita Y※, Fukumoto S※, GlorieuxFH※, Michigami T※, Hasegawa K※, Kitaoka T, Kubota T, Imanishi Y※, Shimotsuji T※, Ozono K
Bone, 67C : 56-62, 2014.

- 7) Guidelines on the management and treatment of glucocorticoid-induced osteoporosis of the Japanese Society for Bone and Mineral Research : 2014 update.
Suzuki Y✳, Nawata H✳, Soen S✳, Fujiwara S✳, Nakayama H, Tanaka I✳, Ozono K, Sagawa A✳, Takayanagi R✳, Tanaka H✳, Miki T✳, Masunari N✳, Tanaka Y✳
J Bone Miner Metab, 32 (4) : 337-350, 2014.

- 8) Prognostic factors for acute encephalopathy with bright tree appearance.
Azuma J✳, Nabatame S, Nakano S, Iwatani Y, Kitai Y✳, Tominaga K, Kagitani-Shimono K, Okinaga T✳, Yamamoto T✳, Nagai T, Ozono K
Brain Dev, 2014.

- 9) Efficacy of micafungin in pediatric immunocompromised patients with invasive fungal infections.
Hashii Y, Kusuki S✳, Takizawa S✳, Tokimasa S✳, Ohta H, Hara J✳, Ozono K
Pediatr Int, 56 (6) : 834-837, 2014.

- 10) De novo WDR45 mutation in a patient showing clinically Rett syndrome with childhood iron deposition in brain.
Ohba C✳, Nabatame S, Iijima Y✳, Nishiyama K✳, Tsurusaki Y✳, Nakashima M✳, Miyake N✳, Tanaka F✳, Ozono K, Saitsu H✳, Matsumoto N✳
J Hum Genet, 59 (5) : 292-295, 2014.

- 11) Therapeutic use of oral sodium phosphate (phosribbon® combination granules) in hereditary hypophosphatemic rickets.
Ozono K, Hasegawa Y✳, Minagawa M✳, Adachi M✳, Namba N, Kazukawa I✳, Kitaoka T, Asakura Y✳, Shimura A✳, Naito Y✳
Clin Pediatr Endocrinol, 23 (1) : 9-15, 2014.

- 12) Late-onset Krabbe disease is predominant in Japan and its mutant precursor protein undergoes more effective processing than the infantile-onset form.
Hossain MA✳, Otomo T, Saito S✳, Ohno K✳, Sakuraba H✳, Hamada Y, Ozono K, Sakai N
Gene, 534 (2) : 144-154, 2014.

- 13) Weight control in schizophrenic patients through Sakata's Charting of Daily Weight Pattern and its associations with temperament and character.
Miyoshi R✳, Matsuo H✳, Naono-Nagatomo K✳, Ozono K, Araki R✳, Ishikawa M✳, Abe H✳, Taniguchi H, Ishida Y✳
Asian J Psychiatr, 7 (1) : 52-57, 2014.

- 14) Problems during the Long-Term Follow-Up after Surgery for Pediatric Solid Malignancies.
Oue T✳, Miyoshi Y, Hashii Y, Uehara S✳, Ueno T✳, Nara K✳, Usui N✳, Ozono K
Eur J Pediatr Surg, 25 (1) : 123-127, 2015.

- 15) Effect of growth hormone treatment on quality of life in Japanese children with growth hormone deficiency : an analysis from a prospective observational study.
Tanaka T✳, Hasegawa T✳, Ozono K, Tanaka H✳, Kanzaki S✳, Yokoya S✳, Chihara K✳, Iwamoto N✳, Seino Y✳
Clin Pediatr Endocrinol, 23 (3) : 83-92, 2014.

- 16) Intractable itch relieved by 4-phenylbutyrate therapy in patients with progressive familial intrahepatic cholestasis type 1.
Hasegawa Y, Hayashi H✳, Naoi S✳, Kondou H, Bessho K, Igarashi K✳, Hanada K✳, Nakao K, Kimura T, Konishi A, Nagasaka H✳, Miyoshi Y, Ozono K, Kusahara H✳
Orphanet J Rare Dis, 9 : 89, 2014.

- 17) Mycobacterium avium complex-associated peritonitis with CAPD after unrelated bone marrow transplantation.
Miyashita E, Yoshida H, Mori D✳, Nakagawa N, Miyamura T, Ohta H✳, Seki M✳, Tomono K✳, Hashii Y, Ozono K
Pediatr Int, 56 (6) : e96-98, 2014.

- 18) A fatal case of infantile malignant osteopetrosis complicated by pulmonary arterial hypertension after hematopoietic stem cell transplantation.
Kuroyanagi Y✳, Kawasaki H✳, Noda Y✳, Ohmachi T✳, Sekiya S✳, Yoshimura K✳, Ohe C✳, Michigami T✳, Ozono K, Kaneko K✳
Tohoku J Exp Med, 234 (4) : 309-312, 2014.

- 19) Negative feedback loop of cholesterol regulation is impaired in the livers of patients with Alagille syndrome.
Miyahara Y✳, Bessho K, Kondou H, Hasegawa Y, Yasuda K, Ida S✳, Ihara Y✳, Mizuta K✳, Miyoshi Y, Ozono K
Clin Chim Acta, 440 : 49-54, 2015.

- 20) Correlation between outbreaks of multidrug-resistant Pseudomonas aeruginosa infection and use of bronchoscopes suggested by epidemiological analysis.
Machida H✳, Seki M✳, Yoshioka N✳, Yabuno K✳, Miyawaki K✳, Yoshida H, Yamamoto N✳, Hamaguchi S✳, Tomono K✳
Biol Pharm Bull, 37 (1) : 26-30, 2014.

- 21) Long-term follow-up of children with refractory immune thrombocytopenia treated with rituximab.
Matsubara K✳, Takahashi Y✳, Hayakawa A✳, Tanaka F✳, Nakadate H✳, Sakai M✳, Maeda N✳, Oka T✳, Ishii E✳, Bessho F✳, Morimoto T✳, Goto H✳, Hashii Y, Hatakeyama N✳, Shirahata A✳, Imaizumi M✳
Int J Hematol, 99 (4) : 429-436, 2014.

- 22) ABL kinase mutation and relapse in 4 pediatric Philadelphia chromosome-positive acute lymphoblastic leukemia cases.
Aoe M✳, Shimada A✳, Muraoka M✳, Washio K✳, Nakamura Y✳, Takahashi T✳, Imada M✳, Watanabe T✳, Okada K✳, Nishiuchi R✳, Miyamura T, Chayama K✳, Shibakura M✳, Oda M✳, Morishima T✳
Int J Hematol, 99 (5) : 609-615, 2014.

- 23) Early use of allogeneic hematopoietic stem cell transplantation for infants with MLL gene-rearrangement-positive acute lymphoblastic leukemia.
Koh K✉, Tomizawa D✉, Moriya-Saito A✉, Watanabe T✉, Miyamura T, Hirayama M✉, Takahashi Y✉, Ogawa A✉, Kato K✉, Sugita K✉, Sato T✉, Deguchi T✉, Hayashi Y✉, Takita J✉, Takeshita Y✉, Tsurusawa M✉, Horibe K✉, Mizutani S✉, Ishii E✉
Leukemia, 29 : 290-296, 2015.

- 24) Prognostic impact of gained chromosomes in high-hyperdiploid childhood acute lymphoblastic leukaemia : a collaborative retrospective study of the Tokyo Children's Cancer Study Group and Japan Association of Childhood Leukaemia Study.
Kato M✉, Imamura T✉, Manabe A✉, Hashii Y, Koh K✉, Sato A✉, Takahashi H✉, Hori H✉, Taki T✉, Inoue M✉, Hayashi Y✉, Horibe K✉, Tsuchida M✉, Kojima S✉, Oda M✉, Ohara A✉
Br J Haematol, 166 (2) : 295-8, 2014.

- 25) Outcome of TCF3-PBX1 positive pediatric acute lymphoblastic leukemia patients in Japan : a collaborative study of Japan Association of Childhood Leukemia Study (JACLS) and Children's Cancer and Leukemia Study Group (CCLSG).
Asai D✉, Imamura T✉, Yamashita Y✉, Suenobu S✉, Moriya-Saito A✉, Hasegawa D✉, Deguchi T✉, Hashii Y, Endo M✉, Hatakeyama N✉, Kawasaki H✉, Hori H✉, Horibe K✉, Yumura-Yagi K✉, Hara J✉, Watanabe A✉, Kikuta A✉, Oda M✉, Sato A✉ ; Japan Association of Childhood Leukemia Study (JACLS) & Children's Cancer and Leukemia Study Group (CCLSG)
Cancer Med, 3 (3) : 623-631, 2014.

- 26) Outcome of children with relapsed acute myeloid leukemia following initial therapy under the AML99 protocol.
Nakayama H✉, Tabuchi K✉, Tawa A✉, Tsukimoto I✉, Tsuchida M✉, Morimoto A✉, Yabe H✉, Horibe K✉, Hanada R✉, Imaizumi M✉, Hayashi Y✉, Hamamoto K✉, Kobayashi R✉, Kudo K✉, Shimada A✉, Miyamura T, Moritake H✉, Tomizawa D✉, Taga T✉, Adachi S✉
Int J Hematol, 100 (2) : 171-179, 2014.

- 27) Inferior outcomes of stage III T lymphoblastic lymphoma relative to stage IV lymphoma and T-acute lymphoblastic leukemia : long-term comparison of outcomes in the JACLS NHL T-98 and ALL T-97 protocols.
Kobayashi R✉, Takimoto T✉, Nakazawa A✉, Fujita N✉, Akazai A✉, Yamato K✉, Yazaki M✉, Deguchi T✉, Hashii Y, Kato K✉, Hatakeyama N✉, Horibe K✉, Hori H✉, Oda M✉ ; Lymphoma Committee, Japanese Association of Childhood Leukemia Study
Int J Hematol, 99 (6) : 743-749, 2014.

- 28) An overall characterization of pediatric acute lymphoblastic leukemia with 2 overexpression.
Yano M✉, Imamura T✉, Asai D✉, Moriya-Saito A✉, Suenobu S✉, Hasegawa D✉, Deguchi T✉, Hashii Y, Kawasaki H✉, Hori H✉, Kosaka Y✉, Kato K✉, Horibe K✉, Yumura-Yagi K✉, Hara J✉, Matsumoto K✉, Kiyokawa N✉, Oda M✉, Sato A✉ ; Japan Association of Childhood Leukemia Study
Genes Chromosomes Cancer, 53 (10) : 815-823, 2014.

- 29) Vaccination strategies to improve outcome of hematopoietic stem cell transplant in leukemia patients : early evidence and future prospects.
Hosen N✉, Maeda T✉, Hashii Y, Tsuboi A✉, Nishida S✉, Nakata J✉, Nakae Y✉, Takashima S✉, Oji Y✉, Oka Y✉, Kumanogoh A✉, Sugiyama H✉
Expert Rev Hematol, 7 (5) : 671-681, 2014.

- 30) Hematopoietic stem cell transplantation for patients with acute lymphoblastic leukemia and Down syndrome.
Goto H✉, Kaneko T✉, Shioda Y✉, Kajiwara M✉, Sakashita K✉, Kitoh T✉, Hayakawa A✉, Miki M✉, Kato K✉, Ogawa A✉, Hashii Y, Inukai T✉, Kato C✉, Sakamaki H✉, Yabe H✉, Suzuki R✉, Kato K✉
Pediatr Blood Cancer, 62 (1) : 148-152, 2015.

- 31) Perioperative management after high-dose chemotherapy with autologous or allogeneic hematopoietic stem cell transplantation for pediatric solid tumors.
Uehara S✉, Oue T✉, Nakahata K✉, Nara K✉, Ueno T✉, Owari M✉, Usui N✉, Miyamura T, Hashii Y
Eur J Pediatr Surg, 25 (1) : 118-122, 2015.

- 32) Invasive pulmonary aspergillosis due to *Aspergillus lentulus* : Successful treatment of a liver transplant patient.
Yoshida H, Seki M✉, Umeyama T✉, Urai M✉, Kinjo Y✉, Nishi I✉, Toyokawa M✉, Kaneko Y✉, Ohno H✉, Miyazaki Y✉, Tomono K✉
J Infect Chemother, 21 (6) : 479-481, 2015.

- 33) Gene expression signature for biliary atresia and a role for Interleukin-8 in pathogenesis of experimental disease.
Bessho K, Mourya R✉, Shivakumar P✉, Walters S✉, Magee JC✉, Rao M✉, Jegga AG✉, Bezerra JA✉
Hepatology 60, 211-223, 2014.

- 34) Reply : to PMID 24493287.
Mourya R✉, Bessho K, Shivakumar P✉, Bezerra JA✉
Hepatology, 61 (1) : 732-733, 2015.

- 35) Biliary repair and carcinogenesis are mediated by IL-33-dependent cholangiocyte proliferation.
Li J✉, Razumilava N✉, Gores GJ✉, Walters S✉, Mizuochi T✉, Mourya R✉, Bessho K, Wang YH✉, Glaser SS✉, Shivakumar P✉, Bezerra JA✉
J Clin Invest 124, 3241-3251, 2014.

- 36) Association of IL28B polymorphisms with virological response to peginterferon and ribavirin therapy in children and adolescents with chronic hepatitis C.
Tajiri H✉, Tanaka Y✉, Takano T✉, Suzuki M✉, Abukawa D✉, Miyoshi Y, Shimizu T✉, Brooks S✉
Hepatol Res, 44 : E38-E44, 2014.

- 37) Statin treatment rescues FGFR3 skeletal dysplasia phenotypes.
Yamashita A✳, Morioka M✳, Kishi H✳, Kimura T, Yahara Y✳, Okada M✳, Fujita K✳, Sawai H✳, Ikegawa S✳, Tsumaki N✳
Nature, 513 (7519) : 507-511, 2014.
- 38) Replication of a GWAS signal in a Caucasian population implicates ADD3 in susceptibility to biliary atresia.
Tsai EA✳, Grochowski CM✳, Loomes KM✳, Bessho K, Hakonarson H✳, Bezerra JA✳, Russo PA✳, Haber BA✳, Spinner NB✳, Devoto M✳
Hum Genet, 133 : 235-243, 2014.
- 39) The ectoenzyme E-NPP3 negatively regulates ATP-dependent chronic allergic responses by basophils and mast cells.
Tsai SH✳, Kinoshita M✳, Kusu T✳, Kayama H✳, Okumura R✳, Ikeda K, Shimada Y✳, Takeda A✳, Yoshikawa S✳, Obata-Ninomiya K✳, Kurashima Y✳, Sato S✳, Umemoto E✳, Kiyono H✳, Karasuyama H✳, Takeda K✳
Immunity (in Press)
- 40) Natural history of chronic hepatitis B virus infection in childhood and efficacy of interferon therapy.
Takano T✳, Tajiri H✳, Etani Y✳, Miyoshi Y, Tanaka Y✳, Brooks S✳
Scand J Gastroenterol, 50 : 892-899, 2015.
- 41) Protection by vascular prostaglandin E2 signaling in hypoxic-ischemic encephalopathy.
Taniguchi H, Anacker C✳, Wang Q, Andreasson K✳
Exp Neurol, 255 : 30-37, 2014.
- 42) Application of transcutaneous carbon dioxide tension monitoring with low electrode temperatures in premature infants in the early postnatal period.
Hirata K, Nishihara M✳, Oshima Y✳, Hirano S✳, Kitajima H✳
Am J Perinatol, 31 (5) : 435-440, 2014.
- 43) Mesenteric hernia causing bowel obstruction in very low-birthweight infants.
Hirata K, Kawahara H✳, Shiono N✳, Nishihara M✳, Kubota A✳, Nakayama M✳, Kitajima H✳
Pediatr Int, 57 (1) : 161-163, 2015.
- 44) Perinatal Factors associated with Long-term Respiratory Sequelae in Extremely-low-birth-weight Infants.
Hirata K, Nishihara M✳, Shiraishi J✳, Hirano S✳, Matsunami K✳, Sumi K✳, Wada N✳, Kawamoto Y✳, Nishikawa M✳, Nakayama M✳, Kanazawa T✳, Kitajima H✳, Fujimura M✳
Arch Dis Child Fetal Neonatal Ed 2015. [accepted for publication]
- 45) Two Japanese familial cases of Caffey disease with and without the common COL1A1 mutation and normal bone density, and review of the literature.
Kitaoka T, Miyoshi Y, Namba N, Miura K✳, Kubota T, Ohata Y, Fujiwara M, Takagi M✳, Hasegawa T✳, Jüppner H✳, Ozono K
Eur J Pediatr, 173 (6) : 799-804, 2014.

- 46) Serum fibroblast growth factor 23 is a useful marker to distinguish vitaminD-deficient rickets from hypophosphatemic rickets.
Kubota T, Kitaoka T, Miura K✉, Fujiwara M, Ohata Y, Miyoshi Y, Yamamoto K, Takeyari S, Yamamoto T✉, Namba N, Ozono K
Horm Res Paediatr, 81 (4) : 251-257, 2014.

- 47) Abnormal pupillary light reflex with chromatic pupillometry in Gaucher disease, *Annals of Clinical and Translational Neurology*.
Narita A✉, Shirai K✉, Kubota N✉, Takayama N✉, Takahashi Y✉, Onuki T✉, Numakura C✉, Kato M✉, Hamada Y, Sakai N, Ohno A✉, Asami M✉, Matsushita S✉, Hayashi A✉, Kumada T✉, Fujii T✉, Horino A✉, Inoue T✉, Kuki I✉, Asakawa K✉, Ishikawa H✉, Ohno K✉, Nishimura Y✉, Tamasaki A✉, Maegaki Y✉, Ohno K✉
ACTN, 1 (2) : 135-140, 2014.

- 48) Mannose 6 phosphorylation of lysosomal enzymes controls B cell functions.
Otomo T, Schweizer M✉, Kollmann K✉, Schumacher V✉, Muschol N✉, Tolosa E✉, Mittrücker HW✉, Bräulke T✉
J Cell Biol, 19 ; 208 (2) : 171-180, 2015.

- 49) Efficacy of deep brain stimulation in an adolescent patient with DYT11 myoclonus-dystonia, *Neurology and Clinical Neuroscience*.
Kimura Y✉, Mihara M✉, Kawarai T✉, Kishima H✉, Sakai N, Takahashi M✉, Mochizuki H✉
Neurology and clinical Neuroscience, 2 : 57-59, 2014.

- 50) Expression of astrocyte-related receptors in cortical dysplasia with intractable epilepsy.
Sukigara S✉, Dai H✉, Nabatame S, Otsuki T✉, Hanai S✉, Honda R✉, Saito T✉, Nakagawa E✉, Kaido T✉, Sato N✉, Kaneko Y✉, Takahashi A✉, Sugai K✉, Saito Y✉, Sasaki M✉, Goto Y✉, Koizumi S✉, Itoh M✉
J Neuropathol Exp Neurol, 73 (8) : 798-806, 2014.

- 51) Progressively increased M50 responses to repeated sounds in autism spectrum disorder with auditory hypersensitivity : a magnetoencephalographic study.
Matsuzaki J, Kagitani-Shimono K, Sugata H✉, Hirata M✉, Hanaie R, Nagatani F, Tachibana M, Tominaga K, Mohri I, Taniike M
PLOS ONE, 9 (7) : e102599, 2014.

- 52) Abnormal corpus callosum connectivity, socio-communicative and motor deficit in children with autism spectrum disorders : A diffusion tensor imaging study.
Hanaie R✉, Mohri I, Kagitani-Shimono K, Tachibana M, Azuma J✉, Matsuzaki J✉, Watanabe Y✉, Fujita N✉, Taniike T
J Autism and Develop Dis, 44 : 2209-2220, 2014.

- 53) Psychometric properties and population-based score distributions of the Japanese Sleep Questionnaire for Preschoolers.
Shimizu S✉, Kato-Nishimura K✉, Mohri I, Kagitani-Shimono K, Tachibana M, Ohno Y✉, Taniike M
Sleep Med, 15 (4) : 451-458, 2014.

- 54) Emotional recognition of children with autism spectrum disorder.
Eto M✳, Sakai S, Kagitani-Shimono K, Mohri I, Taniike M
Journal of Japan Society for Research on Emotions, 22 (1) : 28-39, 2014.

- 55) SPTAN1 encephalopathy : distinct phenotypes and genotypes.
Tohyama J✳, Nakashima M✳, Nabatame S, Gaik-Siew C✳, Miyata R✳, Rener-Primec Z✳,
Kato M✳, Matsumoto N✳, Saitsu H✳
J Hum Genet, 2015. (in Ppress)

- 56) Altered microstructural connectivity of the arcuate fasciculus is related to language disability in children with autism spectrum disorder.
Kimura M✳, Hanaie R, Mohri I, Kagitani-Shimono K, Tachibana M, Matsuzaki J, Fujita N,
Watanabe Y✳, Taniike M
J Brain Sci. (in Press)
- 57) Nationwide survey of glucose transporter-1 deficiency syndrome (GLUT-1DS) in Japan.
Ito Y✳, Takahashi S✳, Kagitani-Shimono K, Natsume J✳, Yanagihara K✳, Fujii T✳, Oguni H✳
Brain Dev, doi : 10. 1066/j-braindeu. 2014.

- 58) Development and verification of child observation sheet for 5-year-old children.
Fujimoto K✳, Nagai T, Okazaki S✳, Kawajiri M✳, Tomiwa K✳
Brain Dev, 36 (2) : 107-115, 2014.

- 59) Daytime physical activity and sleep in preschoolers with developmental disorders.
Tatsumi Y✳, Mohri I, Shimizu S✳, Ohno Y✳, Tachibana M, Taniike M
J Paediatr Child Health, doi : 10. 1111/jpc. 12725, 2014.

- 60) Correlations between the Broad Autism Phenotype and Social Cognition among Mothers of Children with Autism Spectrum Disorder.
Hasegawa K✳, Sakai S, Okuno H, Eto M✳, Kagitani-Shimono K, Mohri I, Taniike M
Japanese Journal of Research on Emotions, 21 (3) : 143-155, 2014.

- 61) Duplication of the NPHP1 gene in patients with autism spectrum disorder and normal intellectual ability : a case series.
Yasuda Y✳, Hashimoto R✳, Fukai R✳, Okamoto N✳, Hiraki Y✳, Yamamori H✳, Fujimoto M✳, Ohi K✳, Taniike M, Mohri I, Nakashima M✳, Tsurusaki Y✳, Saitsu H✳, Matsumoto N✳, Miyake N✳, Takeda M✳
Ann Gen Psychiatry, 6 : 13 : 22, 2014.

- 62) Development of preference for conspecific faces in human infants.
Sanefuji W✳, Wada K, Yamamoto T, Mohri I, Taniike M
Dev Psychol, 50 (4) : 979-985, 2014.