

原著 (英文)

●平成25年度 (2013.4~2014.3) ●

●1.Sodium-coupled neutral amino acid transporter 4 functions as a regulator of protein synthesis during liver development.

Kondou H, Kawai M※, Tachikawa K※, Kimoto A※, Yamagata M※, Koinuma T※, Yamazaki M※, Nakayama M※, Mushiake S※, Ozono K, Michigami T※
Hepatol Res, Epub ahead of print, 2013.

●2.Weaver syndrome and EZH2 mutations : Clarifying the clinical phenotype.

Tatton-Brown K※, Murray A※, Hanks S※, Douglas J※, Armstrong R※, Banka S※, Bird LM※, Clericuzio CL※, Cormier-Daire V※, Cushing T※, Flinter F※, Jacquemont ML※, Joss S※, Kinning E※, Lynch SA※, Magee A※, McConnell V※, Medeira A※, Ozono K, Patton M※, Rankin J※, Shears D※, Simon M※, Splitt M※, Strenger V※, Stuurman K※, Taylor C※, Titheradge H※, Van Maldergem L※, Temple IK※, Cole T※, Seal S ; Childhood Overgrowth Consortium※, Rahman N※
Am J Med Genet A, 161A (12) : 2972-2980, 2013.

●3.Therapeutic Efficacy and Safety of GH in Japanese Children with Down Syndrome Short Stature Accompanied by GH Deficiency.

Meguri K※, Inoue M※, Narahara K※, Sato T※, Takata A※, Ohki N※, Ozono K
Clin Pediatr Endocrinol, 22 (4) : 65-72, 2013.

●4.Identification of AP2S1 Mutation and Effects of Low Calcium Formula in an Infant with Hypercalcemia and Hypercalciuria.

Fujisawa Y※, Yamaguchi R※, Satake E※, Ohtaka K※, Nakanishi T※, Ozono K, Ogata T※
J Clin Endocrinol Metab, 98 (12) : E2022-E2027, 2013.

●5.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 (3) : 1457-1466, 2014.

●6.Abdominal neuroblastoma with inferior vena cava right atrial extension.

Uehara S※, Takama Y※, Yoshida H, Hashii Y, Oue T※, Usui N※
J Ped Surg Case Reports, 1 : 119-121, 2013.

●7.Salvage allogeneic hematopoietic SCT for primary graft failure in children.

Kato M※, Matsumoto K※, Suzuki R※, Yabe H※, Inoue M※, Kigasawa H※, Inagaki J※, Koh K※, Hashii Y, Tauchi H※, Suminoe A※, Kikuta A※, Sakamaki H※, Kawa K※, Kato K※, Fukuda T※
Bone Marrow Transplant, 48 (9) : 1173-1178, 2013.

●8. Autosomal dominant anhidrotic ectodermal dysplasia with immunodeficiency caused by a novel NFKBIA mutation, p.Ser36Tyr, presents with mild ectodermal dysplasia and non-infectious systemic inflammation.

Yoshioka T✉, Nishikomori R✉, Hara J✉, Okada K✉, Hashii Y, Okafuji I✉, Nodomi S✉, Kawai T✉, Izawa K✉, Ohnishi H✉, Yasumi T✉, Nakahata T✉, Heike T✉

J Clin Immunol, 33 (7) : 1165-1174, 2013.

●9. IKZF1 deletion is associated with a poor outcome in pediatric B-cell precursor acute lymphoblastic leukemia in Japan.

Asai D✉, Imamura T✉, Suenobu S✉, Saito A✉, Hasegawa D✉, Deguchi T✉, Hashii Y, Matsumoto K✉, Kawasaki H✉, Hori H✉, Iguchi A✉, Kosaka Y✉, Kato K✉, Horibe K✉, Yumura-Yagi K✉, Hara J✉, Oda M✉ : Japan Association of Childhood Leukemia Study (JACLS) Cancer Med, 2 (3) : 412-419, 2013.

●10. 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✉ : the Japan Association of Childhood Leukemia Study (JACLS) & Children's Cancer and Leukemia Study Group (CCLSG).

Cancer Med, 2014. [Epub ahead of print]

●11. Impact of cow's milk allergy on enterocolitis associated with Hirschsprung's disease.

Umeda S✉, Kawahara H✉, Yoneda A✉, Tazuke Y✉, Tani G✉, Ishii T✉, Goda T✉, Hirano K✉, Ikeda K, Ida S✉, Nakayama M✉, Kubota A✉, Fukuzawa M✉

Pediatr Surg Int, 29 (11) : 1159-1163, 2013.

●12. 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, 2013. [Epub ahead of print]

●13. 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 (2) : 235-243, 2014.

●14. Integrative genomics identifies candidate microRNAs for pathogenesis of experimental biliary atresia.

Bessho K, Shanmukhappa K✉, Sheridan R✉, Shivakumar P✉, Mourya R✉, Walters S✉, Kaimal V✉, Dilbone E✉, Jegga A✉G, Bezerra JA✉

BMC Syst Biol, 7 : 104, 2013.

●15. Gene expression signature for biliary atresia and a role for Interleukin-8 in pathogenesis of experimental disease.

Besho K, Mourya R✉, Shivakumar P✉, Walters S✉, Magee JC✉, Rao MP✉, Anil G Jegga AG✉, Bezerra JA✉

Hepatology 2014 Accepted for publication. [Epub ahead of print]

●16. 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, 2013. (DOI : 10.1055/s-0033-1352485)

【大阪小児先進医療研究会・平成25年度優秀学術論文賞】

●17. Drastic shift from positive to negative estrogen effect on bone morphogenetic protein signaling in pulmonary arterial endothelial cells under hypoxia.

Ichimori H, Kogaki S, Takahashi K, Ishida H, Narita J✉, Nawa N, Baden H, Uchikawa T✉, Okada Y, Ozono K.

Circulation Journal, 77 (8) : 2118-26, 2013.

●18. Effects of maternal hyperoxygenation in a case of severe congenital diaphragmatic hernia accompanied by hydrops fetalis.

Ishii R, Inamura N✉, Kubota A✉, Kayatani F✉, Shimada M✉, Ishii K✉, Hidaka N✉, Mitsuda N✉

J Ped Surg Case Reports, 2 (1) : 15-19, 2014.

●19. Clinical features of adult patients with Eisenmenger's syndrome in Japan and Korea.

Sakazaki H✉, Niwa K✉, Nakazawa M✉, Saji T✉, Nakanishi ✉T, Takamuro M✉, Ueno M✉, Kato H✉, Takatsuki S✉, Matsushima M✉, Kojima N✉, Ichida ✉F, Kogaki S, Kido S✉,

Arakaki Y✉, Waki K✉, Akagi T✉, Joo K✉, Muneuchi J✉, Suda K✉, Lee HJ✉, Shintaku H✉
Int J Cardiol, 167 (1) : 205-9, 2013.

●20. The use of cell-sheet technique eliminates arrhythmogenicity of skeletal myoblast-based therapy to the heart with enhanced therapeutic effects.

Narita T✉, Shintani Y✉, Ikebe C✉, Kaneko M✉, Harada N✉, Tshuma N✉, Takahashi K, Campbell NG✉, Copen SR✉, Yashiro K✉, Sawa Y✉, Suzuki K✉

Int J Cardiol, 168 (1) : 261-269, 2013.

●21. Detection and characterization of two novel mutations in the HNF4A gene in maturity-onset diabetes of the young type 1 in two Japanese families.

Fujiwara M, Namba N, Miura K, Kitaoka T, Hirai H✉, Kondou H, Shimotsuji T✉, Numakura C✉, Ozono K

Horm Res Paediatr, 79 (4) : 220-226, 2013.

●22. Insulin-like growth factor-1 receptor in mature osteoblasts is required for periosteal bone formation induced by reloading.

Kubota T, Elalieh HZ✉, Saless N✉, Fong C✉, Wang Y✉, Babey M✉, Cheng Z✉, Bikle DD✉
Acta Astronaut, 92 (1) : 73-78, 2013.

- 23.A human skeletal overgrowth mutation increases maximal velocity and blocks desensitization of guanylyl cyclase-B.
Robinson JW✳, Dickey DM✳, Miura K, Michigami T✳, Ozono K, Potter LR✳
Bone, 56 (2) : 375-382, 2013.

- 24.Treatment of hypophosphatemic rickets with phosphate and active vitamin D in japan : A questionnaire-based survey.
Fujiwara M, Namba N, Ozono K, Arisaka O✳, Yokoya S✳ : Committee on Drugs, Japanese Society for Pediatric Endocrinology✳
Clin Pediatr Endocrinol, 22 (1) : 9-14, 2013.

- 25.Serum fibroblast growth factor 23 is a useful marker to distinguish vitamin D-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.

- 26.Overgrowth syndrome associated with a gain-of-function mutation of the natriuretic peptide receptor 2 (NPR2) gene.
Miura K, Kim OH✳, Lee HR✳, Namba N, Michigami T✳, Yoo WJ✳, Choi IH✳, Ozono K, Cho TJ✳
Am J Med Genet A, 164A (1) : 156-163, 2014.

- 27.Therapeutic use of oral sodium phosphate (phosribbonR 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.

- 28.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, 2014. [Epub ahead of print]

- 29.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✳, Juppner H✳, Ozono K
Eur J Pediatr, 173 (6) : 799-804, 2014.

- 30.An adult patient with mucopolidosis III alpha/beta presenting with parkinsonism.
Hara M✳, Inokuchi T✳, Taniwaki T✳, Otomo T, Sakai N, Matsuishi T✳, Yoshino M✳
Brain Dev, 35 (5) : 462-465, 2013.

- 31. Isolated pyramidal tract impairment in the central nervous system of adult-onset Krabbe disease with novel mutations in the GALC gene.
Tokushige SI✉, Sonoo T✉, Maekawa R✉, Shirota Y✉, Hanajima R✉, Terao Y✉, Matsumoto H✉, Hossain MA, Sakai N, Shiio Y✉
Brain Dev. 35 (6) : 579-581, 2013.

- 32. 14-3-3 ε Gene variants in a Japanese patient with left ventricular noncompaction and hypoplasia of the corpus callosum.
Chang B✉, Gorbea C✉, Lezin G✉, Li L✉, Shan L✉, Sakai N, Kogaki S, Otomo T, Okinaga T✉, Hamaoka A✉, Yu X✉, Hata Y✉, Nishida N✉, Yost HJ✉, Bowles NE✉, Brunelli L✉, Ichida F✉
Gene, 515 (1) : 173-180, 2013.

- 33. A novel homozygous GALC mutation : Very early onset and rapidly progressive Krabbe disease.
Kardas F✉, Uzak AS✉, Hossain MA, Sakai N, Canpolat M✉, Y?k?lmaz A✉
Gene, 517 (1) : 125-127, 2013.

- 34. Microdeletions of 3p21.31 characterized by developmental delay, distinctive features, elevated serum creatine kinase levels, and white matter involvement.
Eto K✉, Sakai N, Shimada S✉, Shioda M✉, Ishigaki K✉, Hamada Y, Shinpo M, Azuma J✉, Tominaga K, Shimojima K✉, Ozono K, Osawa M✉, Yamamoto T✉
Am J Med Genet A, 161 (12) : 3049-3056, 2013.

- 35. 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.

- 36. Leigh syndrome with Fukuyama congenital muscular dystrophy : A case report.
Kondo H, Tanda K✉, Tabata C✉, Hayashi K✉, Kihara M✉, Kizaki Z✉,
Taniguchi-Ikeda M✉, Mori M✉, Murayama K✉, Ohtake A✉
Brain Dev, 2013. [Epub ahead of print]

- 37. 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, Saito H✉, Matsumoto N✉
J HumGenet, 2014. [Epub ahead of print]

- 38. A case of severe progressive early-onset epileptic encephalopathy : unique GABAergic interneuron distribution and imaging.
Inoue T✉, Kawawaki H✉, Kuki I✉, Nabatame S, Tomonoh Y✉, Sukigara S✉, Horino A✉, Nukui M✉, Okazaki S✉, Tomiwa K✉, Kimura-Ohba S, Inoue T✉, Hirose S✉, Shiomi M✉, Itoh M✉
J Neurol Sci, 327 (1-2) : 65-72, 2013.

- 39. Altered microstructural connectivity of the superior cerebellar peduncle is related to motor dysfunction in children with autistic spectrum disorders.
Hanaie R✳, Mohri I, Kagitani-Shimono K, Tachibana M, Azuma J✳, Matsuzaki J✳, Watanabe Y✳, Fujita N✳, Taniike M.
Cerebellum, 12 (5) : 645-56, 2013.

- 40. Effect of adrenocorticotrophic hormone therapy for epileptic spasms developing after the age of 1 year.
Fukui M✳, Shimakawa S✳, Kuki I✳, Kawawaki H✳, Mogami Y✳, Ysuzuki Y✳, Nakano S, Okinaga T
Seizure, 2013 (in press)

- 41. Which is the Most Appropriate Disconnection Surgery for Refractory Epilepsy in Childhood?
Kishima H✳, Oshino S✳, Tani N✳, Maruo Y✳, Morris S✳, KhooMing H✳, Yanagisawa T✳, Shimono K, Okinaga T✳, Hirata M✳, Kato A✳, Yoshimine T✳
Neurol Med Chir (Tokyo), 53 (11) : 814-820, 2013.

- 42. Oral mexiletine for lidocaine-responsive neonatal epilepsy.
Nakazawa M✳, Okumura A✳, Nijima S✳, Yamashita S✳, Shimono K, Hirose S✳, Shimizu T✳
Brain Dev, 35 (7) : 667-669, 2013.

- 43. 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, et al.
J Autism Dev Disord (Epub ahead of print).

- 44. Development of Preference for Conspecific Faces in Human Infants.
Sanefuji W✳, Wada K, Yamamoto T✳, Mohri I, Taniike M
Dev Psychol, 2013 (Epub ahead of print).

- 45. Psychometric properties and population-based score distributions of the Japanese Sleep Questionnaire for Preschoolers.
Shimizu S✳, Kato-Nishimura K✳, Mohri I, Shimono K, Tachibana M, Ohno Y✳, Taniike M
Sleep Med (Epub ahead of print).

- 46. 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✳, Shimono K, Mohri I, Taniike M
Japan Society for Research on Emotions (Epub ahead of print).

- 47. Directing and maintaining infants' attention in mother-infant interaction on infants with and without autism spectrum disorder.
Shizawa M✳, Sanefuji W✳, Mohri I
Japanese Journal of Special Education , 50 (1) : 3-10, 2013.

●48. Long-term administration of intranasal oxytocin is a safe and promising therapy for early adolescent boys with autism spectrum disorders.

Tachibana M, Kagitani-Shimono K, Mohri I, Yamamoto T✳, Sanefuji W✳, Nakamura A✳, Oishi M✳, Kimura T✳, Onaka T✳, Ozono K, Taniike M
J Child Adolesc Psychopharmacol, 23 (2) : 123-127, 2013.

●49. 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 (Epub ahead of print).

●50. Body movement analysis during sleep for children with ADHD using video image processing.

Nakatani M✳, Okada S✳, Shimizu S✳, Mohri I, Ohno Y✳, Taniike M, Makikawa M✳
Conf Proc IEEE Eng Med Biol Soc, 2013 : 6389-6392, 2013.