原著(邦文)

●令和２年度（2020.4～2021.3）●

●１）Long-term efficacy and safety of two doses of Norditropin®（somatropin） in Noonan syndrome : a 4-year randomized, double-blind, multicenter trial in Japanese patients.
Horikawa R, Ogata T, Matsubara Y, Yokoya S, Ogawa Y, Nishijima K, Endo T, Ozono K.
Endocr J, 67（8） : 803-818, 2020.

●２） Endocrinological Features of Hartsfield Syndrome in an Adult Patient With a Novel Mutation of FGFR1.
Kobayashi S, Tanigawa J, Kondo H, Nabatame S, Maruoka A, Sho H, Tanikawa K, Inui R, Otsuki M, Shimomura I, Ozono K, Hashimoto K.
J Endocr Soc, 2 ; 4（5）, 2020.

●３） A Nationwide Questionnaire Survey Targeting Japanese Pediatric Endocrinologists Regarding Transitional Care in Childhood, Adolescent, and Young Adult Cancer Survivors.
Miyoshi Y, Yorifuji T, Shimizu C, Nagasaki K, Kawai M, Ishiguro H, Okada S, Kanno J, Takubo N, Muroya K, Ito J, Horikawa R, Yokoya S, Ozono K.
Clin Pediatr Endocrinol, 29（2） : 55-62, 2020.

●４） Effect of Asfotase Alfa on Muscle Weakness in a Japanese Adult Patient of Hypophosphatasia
With Low ALP Levels.
Koyama H, Yasuda S, Kakoi S, Ohata Y, Shimizu Y, Hasegawa C, Hayakawa A, Akiyama T, Yagi T, Aotani D, Imaeda K, Ozono K, Kataoka H, Tanaka T.
Intern Med, 59（6） : 811-815, 2020.

●５） Burden of Illness in Adults with Hypophosphatasia : Data from the Global Hypophosphatasia
Patient Registry.
Seefried L, Dahir K, Petryk A, Högler W, Linglart A, Martos-Moreno GÁ, Ozono K, Fang S, Rockman-Greenberg C, Kishnani PS.
J Bone Miner Res, 35（11） : 2171-2178, 2020.

●６） A case of HDR syndrome coexisting with tetralogy of Fallot, with a novel GATA3 mutation, which manifested as a renal abscess.
Ikeuchi M, Kiyota K, Itonaga T, Kawano-Matsuda F, Ohata Y, Fujiwara M, Kubota T, Ozono K, Ihara K.
CEN Case Rep, 10（2） : 241-243, 2020.

●７） Highly sensitive detection of GATA1 mutations in patients with myeloid leukemia associated with Down syndrome by combining Sanger and targeted next generation sequencing.
Terui K, Toki T, Taga T, Iwamoto S, Miyamura T, Hasegawa D, Moritake H, Hama A, Nakashima K,

Kanezaki R, Kudo K, Saito AM, Horibe K, Adachi S, Tomizawa D, Ito E.
Genes Chromosomes Cancer, 59（3）：160-167, 2020.

●8） Prevalence of germline GATA2 and SAMD9 / 9L variants in paediatric haematological disorders with monosomy 7.
Yoshida M, Tanase-Nakao K, Shima H, Shirai R, Yoshida K, Osumi T, Deguchi T, Mori M, Arakawa Y,
Takagi M, Miyamura T, Sakaguchi K, Toyoda H, Ishida H, Sakata N, Imamura T, Kawahara Y, Morimoto A,
Koike T, Yagasaki H, Ito S, Tomizawa D, Kiyokawa N, Narumi S, Kato M.
Br J Haematol, 191（5）：835-843, 2020.

●9） Conditioning regimen for allogeneic bone marrow transplantation in children with acquired bone marrow failure : fludarabine / melphalan vs. fludarabine / cyclophosphamide.
Yoshida N, Takahashi Y, Yabe H, Kobayashi R, Watanabe K, Kudo K, Yabe M, Miyamura T, Koh K, Kawaguchi H, Goto H, Fujita N, Okada K, Okamoto Y, Kato K, Inoue M, Suzuki R, Atsuta Y, Kojima S,
Pediatric Aplastic Anemia Working Group of the Japan Society for Hematopoietic Cell Transplantation.
Bone Marrow Transplant, 55（7）：1272-1281, 2020.

●10） Long-term outcome in patients with Fanconi anemia who received hematopoietic stem cell
transplantation : a retrospective nationwide analysis.
Yabe M, Morio T, Tabuchi K, Tomizawa D, Hasegawa D, Ishida H, Yoshida N, Koike T, Takahashi Y, Koh K, Okamoto Y, Sano H, Kato K, Kanda Y, Goto H, Takita J, Miyamura T, Noguchi M, Kato K, Hashii Y, Astuta Y, Yabe H.
Int J Hematol, 113（1）：134-144, 2020.

●11） A risk-stratified therapy for infants with acute lymphoblastic leukemia : a report from the JPLSG MLL-10 trial.
Tomizawa D, Miyamura T, Imamura T, Watanabe T, Moriya Saito A, Ogawa A, Takahashi Y, Hirayama M,
Taki T, Deguchi T, Hori T, Sanada M, Ohmori S, Haba M, Iguchi A, Arakawa Y, Koga Y, Manabe A, Horibe K, Ishii E, Koh K.
Blood, 136（16）：1813-1823, 2020.

●12） Prognostic and therapeutic factors influencing the clinical outcome of metastatic Ewing sarcoma family of tumors : A retrospective report from the Japan Ewing Sarcoma Study Group.
Umeda K, Miyamura T, Yamada K, Sano H, Hosono A, Sumi M, Okita H, Kamio T, Maeda N, Fujisaki H,
Jyoko R, Watanabe A, Hosoya Y, Hasegawa D, Takenaka S, Nakagawa S, Chin M, Ozaki T ; Japan Ewing
Sarcoma Study Group. Prognostic and therapeutic factors influencing the clinical outcome of metastatic
Ewing sarcoma family of tumors : A retrospective report from the Japan Ewing Sarcoma Study Group.
Pediatr Blood Cancer, 136（16）：1813-1823, 2020.

●13） The outcomes of relapsed acute myeloid leukemia in children : Results from the Japanese
Pediatric Leukemia / Lymphoma Study Group AML-05R study.
Moritake H, Tanaka S, Miyamura T, Nakayama H, Shiba N, Shimada A, Terui K, Yuza Y, Koh K, Goto H,
Kakuda H, Saito A, Hasegawa D, Iwamoto S, Taga T, Adachi S, Tomizawa D.
The outcomes of relapsed acute myeloid leukemia in Children : Results from the Japanese Pediatric
Leukemia / Lymphoma Study Group AML-05R study.
Pediatr Blood Cancer, 68（1） : e28736, 2021.

●14） Clinical outcome of patients with recurrent or refractory localized Ewing's sarcoma family of
tumors : A retrospective report from the Japan Ewing Sarcoma Study Group.
Umeda K, Miyamura T, Yamada K, Sano H, Hosono A, Sumi M, Okita H, Kumamoto T, Kawai A, Hirayama J, Jyoko R, Sawada A, Nakayama H, Hosoya Y, Maeda N, Yamamoto N, Imai C, Hasegawa D,
Chin M, Ozaki T ; Japan Ewing Sarcoma Study Group.
Cancer Rep, Epub, 2021.

●15） Post-induction MRD by FCM and GATA1-PCR are significant prognostic factors for myeloid
leukemia of Down syndrome.
Taga T, Tanaka S, Hasegawa D, Terui K, Toki T, Iwamoto S, Hiramatsu H, Miyamura T, Hashii Y, Moritake H, Nakayama H, Takahashi H, Shimada A, Taki T, Ito E, Hama A, Ito M, Koh K, Hasegawa D,
Saito AM, Adachi S, Tomizawa D.
Leukemia, Epub, 2021.

●16） Effect of extramedullary disease on allogeneic hematopoietic cell transplantation for pediatric
acute myeloid leukemia : a nationwide retrospective study.
Sakaguchi H, Miyamura T, Tomizawa D, Taga T, Ishida H, Okamoto Y, Koh K, Yokosuka T, Yoshida N,
Sato M, Noguchi M, Okada K, Hori T, Takeuchi M, Kosaka Y, Inoue M, Hashii Y, Atsuta Y.
Bone Marrow Transplant, Epub, 2021.

●17） Pharmacokinetics, Safety, and Efficacy of Glecaprevir / Pibrentasvir in Adolescents With Chronic Hepatitis C Virus : Part 1 of the DORA Study.
Jonas MM, Squires RH, Rhee SM, Lin CW, Bessho K, Feiterna-Sperling C, Hierro L, Kelly D, Ling SC,
Strokova T, Del Valle-Segarra A, Lovell S, Liu W, Ng TI, Porcalla A, Gonzalez YS, Burroughs M, Sokal E.
Hepatology, 71（2） : 456-462, 2020.

●18） Proposal of a liver histology-based scoring system for bile salt export pump deficiency.
Zen Y, Kondou H, Nakazawa A, Tanikawa K, Hasegawa Y, Bessho K, Imagawa K, Ishige T, Inui A, Suzuki M, Kasahara M, Yamamoto K, Yoshioka T, Kage M, Hayashi H.

Hepatol Res, 50（6）：754-762, 2020.

●19） Assessment of Adenosine Triphosphatase Phospholipid Transporting 8B1（ATP8B1）Function in Patients With Cholestasis With ATP8B1 Deficiency by Using Peripheral Blood Monocyte-Derived Macrophages.
Mizutani A, Sabu Y, Naoi S, Ito S, Nakano S, Minowa K, Mizuochi T, Ito K, Abukawa D, Kaji S, Sasaki M,
Muroya K, Azuma Y, Watanabe S, Oya Y, Inomata Y, Fukuda A, Kasahara M, Inui A, Takikawa H, Kusuhara H, Bessho K, Suzuki M, Togawa T, Hayashi H.
Hepatol Commun, 26（5）：52-62, 2020.

●20） Effects of Prophylactic Antibiotics on Length of Stay and Total Costs for Pediatric Acute Pancreatitis : A Nationwide Database Study in Japan.
Ikeda Kurakawa K, Okada A, Jo T, Ono S, Bessho K, Michihata N, Matsui H, Yamaguchi S, Fushimi K,
Kadowaki T, Yasunaga H.
Pancreas, 49（10）：1321-1326, 2020.

●21） Refractory liver dysfunction was remarkably improved with chelating agents of Wilson's disease, in a patient with systemic lupus erythematosus-like syndrome after a parvovirus B19 infection.
Nameki S, Maeda Y, Shibahara T, Fukui J, Shimizu T, Bessho K, Fujiwara H.
Mod Rheumatol Case Rep, 5（1）：1-6, 2020.

●22） Safety and Efficacy of Everolimus Rescue Treatment After Pediatric Living Donor Liver Transplantation.
Ueno T, Kodama T, Noguchi Y, Deguchi K, Nomura M, Saka R, Watanabe M, Tazuke Y, Bessho K, Okuyama H.
Transplant Proc, 52（6）：1829-1832, 2020.

●23）Serum Trough Concentration and Effects of Mycophenolate Mofetil Based on Pathologic Findings in Infants After Liver Transplantation.
Ueno T, Kodama T, Noguchi Y, Deguchi K, Nomura M, Saka R, Watanabe M, Tazuke Y, Bessho K, Okuyama H.
Transplant Proc, 52（6）：1855-1857, 2020.

●24） Beta-D-Glucan Levels With Use of an Anti-adhesion Barrier Film in Pediatric Living Donor Liver Transplantation.
Ueno T, Kodama T, Noguchi Y, Deguchi K, Nomura M, Saka R, Watanabe M, Tazuke Y, Bessho K, Okuyama H.
Transplant Proc, 52（6）：1818-1820, 2020.

●25） One Year of Preemptive Valganciclovir Administration in Children After Liver Transplantation.
Ueno T, Kodama T, Noguchi Y, Deguchi K, Nomura M, Saka R, Watanabe M, Tazuke Y, Bessho K, Okuyama H.
Transplant Proc, 52（6）：1852-1854, 2020.

●26） Neonatal cholestasis can be the first symptom of McCune–Albright syndrome : A case report and review of literature.
Satomura Y, Bessho K, Kitaoka T, Takeyari S, Ohata Y, Kubota T, Ozono K.
World J Clin Pediatr, 10（2） : 7-14, 2021.

●27） Growth hormone treatment for extremely low birthweight children born small for gestational age.
Onuma S, Ida S, Maeyama T, Shoji Y, Etani Y, Kawai M.
Pediatr Int, 63（1） : 46-52, 2021.

●28） Successful neurosurgical separation of conjoined spinal cords in pygopagus twins : case report and literature review.
Yokota C, Kagawa N, Bamba Y, Tazuke Y, Kitabatake Y, Nakagawa T, Hirayama R, Okuyama H, Kishima H.
J. Nerurosurg, 1（9） : CASE218, 2021.

●29） Prognosis of conventional vs. high-frequency ventilation for congenital diaphragmatic hernia : a retrospective cohort study.
Fuyuki M, Usui N, Taguchi T, Hayakawa M, Masumoto K, Kanamori Y, Amari S, Yamoto M, Urushihara N,
Inamura N, Yokoi A, Okawada M, Okazaki T, Toyoshima K, Furukawa T, Terui K, Ohfuji S, Tazuke Y, Uchida K, Okuyama H, Esumi G, Oomura J, Sakai K, Kondo T, Matsuura T, Motokura K, Kawataki M,
Katsumata K, Inoue M, Nagata K, Ito M, Miura R, Ueda K, Sato Y, Saitou A, Muramatsu Y, Sekimoto S,
Ikuta Y, Takama Y, Saka R, Matsuura A, Kitabatake Y, Taniguchi H, Takeuchi M, Kawamura A, Mochizuki N,
Fukumoto K, Ueda Y, Takayasu H, Urita Y, Kimura S.
J. Perinatol, 41（4） : 814-823, 2021.

●30） 4-Phenylbutyrate ameliorates apoptotic neural cell death in Down syndrome by reducing protein aggregates.
Hirata K, Nambara T, Kawatani K, Nawa N, Yoshimatsu H, Kusakabe H, Banno K, Nishimura K, Ohtaka M,
Nakanishi M, Taniguchi H, Arahori H, Wada K, Ozono K, Kitabatake Y.
Sci Rep, 10（1） ,14047, 2020.

●31） Prenatal clinical manifestations in individuals with COL4A1 / 2 variants.
Itai T, Miyatake S, Taguri M, Nozaki F, Ohta M, Osaka H, Morimoto M, Tandou T, Nohara F, Takami Y,
Yoshioka F, Shimokawa S, Okuno-Yuguchi J, Motobayashi M, Takei Y, Fukuyama T, Kumada S, Miyata Y,
Ogawa C, Maki Y, Togashi N, Ishikura T, Kinoshita M, Mitani Y, Kanemura Y, Omi T, Ando N, Hattori A,
Saitoh S, Kitai Y, Hirai S, Arai H, Ishida F, Taniguchi H, Kitabatake Y, Ozono K, Nabatame S, Smigiel R,
Kato M, Tanda K, Saito Y, Ishiyama A, Noguchi Y, Miura M, Nakano T, Hirano K, Honda R, Kuki I, Takanashi JI, Takeuchi A, Fukasawa T, Seiwa C, Harada A, Yachi Y, Higashiyama H, Terashima H,

Kumagai T, Hada S, Abe Y, Miyagi E, Uchiyama Y, Fujita A, Imagawa E, Azuma Y, Hamanaka K, Koshimizu E, Mitsuhashi S, Mizuguchi T, Takata A, Miyake N, Tsurusaki Y, Doi H, Nakashima M, Saitsu H, Matsumoto N.
J. Med. Genet, Epub, 2020.

●32） Successful management of fetal hemolytic disease due to strong anti-Rh17 with plasma exchange and intrauterine transfusion in a woman with the D--phenotype.
Mimura M, Endo M, Takahashi A, Doi Y, Sakuragi M, Kiyokawa T, Taniguchi H, Kitabatake Y, Handa M,
Tomimatsu T, Tomiyama Y, Isaka Y, Kimura T.
Int. J. Hematol, 111（1） : 149-154, 2020.

●33） Bridge to recovery with Berlin Heart EXCOR in children <10 kg with Dilated cardiomyopathy : a
histological analysis.
Tominaga Y, Ueno T, Kido T, Kanaya T, Narita J, Ishida H, Toda K, Kuratani T, Sawa Y.
Eur J Cardiothorac Surg, 58（2） : 253-260, 2020.

●34） Reversible Cerebral Vasoconstriction Syndrome after Heart Transplantation.
Tsukahara R, Ishida H, Narita J, Ishii R, Ozono K.
Pediatr Int, in press 2021.

●35） Cardiac Fibroblasts Play Pathogenic Roles in Idiopathic Restrictive Cardiomyopathy.
Tsuru H, Ishida H, Narita J, Ishii R, Suginobe H, Ishii Y, Wang R, Kogaki S, Taira M, Ueno T, Miyashita Y,
Kioka H, Asano Y, Sawa Y, Ozono K.
Circ J, 85（5） : 677-686, 2021.

●36） Heart Rate Reduction Improves Right Ventricular Function and Fibrosis in Pulmonary Hypertension.
Ishii R, Okumura K, Akazawa Y, Malhi M, Ebata R, Sun M, Fujioka T, Kato H, Honjo O, Kabir G, Kuebler WM, Connelly K, Maynes JT, Friedberg MK.
Am J Respir Cell Mol Biol, 63（6） : 843-855, 2020.

●37） Longitudinal prediction of transplant-free survival by echocardiography in pediatric dilated
cardiomyopathy.
Ishii R, Steve Fan CP, Mertens L, Manlhiot C, Friedberg MK.
Can J Cardiol, S0828-282X（20） : 31145-4, 2020.

●38） Asymmetric Regional Work Contributes to Right Ventricular Fibrosis, Inefficiency and Dysfunction in Pulmonary Hypertension versus Regurgitation.
Ebata R, Fujioka T, Diab SG, Pieles G, Ishii R, Ide H, Sun M, Slorach C, Liu K, Honjo O, Stortz G, Friedberg MK.
J Am Soc Echocardiog, S0894-7317（20） : 30805-1, 2020.

●39） Pulmonary artery banding is a relevant model to study the right ventricular remodeling and

dysfunction that occurs in pulmonary arterial hypertension.
Akazawa Y, Okumura K, Ishii R, Slorach C, Hui W, Ide H, Honjo O, Sun M, Kabir G, Connelly K, Friedberg MK.
J Appl Physio（l 1985）, 129（2） : 238-246, 2020.

●40） Idiopathic True Brachial Artery Aneurysm in a 21-Month-Old Infant : A Case Study.
Ishigaki S, Ishii R, Ishida H, Narita J, Kogaki S, Watanabe K, Shibuya T, Sawa Y, Ozono K.
J Pediatr Cardiol Card Surg, 4（2） : 84-89, 2020.

●41） Pediatric patient with restrictive cardiomyopathy on staged biventricular assist device support with Berlin Heart EXCOR® underwent heart transplantation successfully : the first case in Japan.
Araki K, Ueno T, Taira M, Kanaya T, Watanabe T, Tominaga Y, Ishii R, Ishida H, Narita J, Toda K, Kuratani T, Sawa Y.
J Artif Organs, Epub, 2020.

●42） A Randomized Study of Safety and Efficacy of Two Doses of Ambrisentan to Treat Pulmonary
Arterial Hypertension in Pediatric Patients Aged 8 Years up to 18 Years.
Ivy D, Beghetti M, Juaneda-Simian E, Miller D, Lukas M, Ioannou C, Okour M, Narita J, Berger R.
J Pediatr X. Volume 5, 100055, 2020.

●43） Innovative Therapeutic Strategy Using Prostaglandin I2 Agonist （ONO1301） Combined with Nano Drug Delivery System For Pulmonary Arterial Hypertension.
Kaynaya T, Miyagawa S, Kawamura T, Sakai Y, Masada K, Nawa N, Ishida H, Narita J, Toda K, Kuratani T,
Sawa Y.
Sci Rep, 31 ; 11（1） : 7292, 2021.

●44） Autologous skeletal myoblast patch implantation prevents the deterioration of myocardial ischemiaand right heart dysfunction in a pressure-overloaded right heart porcine model.
Araki K, Miyagawa S, Kawamura T, Ishii R, Watanabe T, Harada A, Taira M, Toda K, Kuratani T, Ueno T,
Sawa Y.
PLoS One, 16（2） : e0247381, 2021.

●45） Long-term outcomes for Asian patients with X-linked hypophosphataemia : rationale and design of the SUNFLOWER longitudinal, observational cohort study.
Kubota T, Fukumoto S, Hae Il Cheong, Michigami T, Namba N, Ito N, Tokunaga S, Gibbs Y, Ozono K.
BMJ Open, 10（6） : e036367, 2020.

●46） Potential pathological role of single nucleotide polymorphism（c.787T>C） in alkaline phosphatase （ALPL ） for the phenotypes of hypophosphatasia.
Matsuda N, Takasawa K, Ohata Y, Takishima S, Kubota T, Ishihara Y, Fujiwara M, Ogawa E, Morio T,
Kashimada K, Ozono K.
Endocr J, 67（12） : 1227-1232, 2020.

●47） Once-daily, subcutaneous vosoritide therapy in children with achondroplasia : a randomised,
double-blind, phase 3, placebo-controlled, multicentre trial.
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.
Lancet, 396（10252） : 684-692, 2020.

●48） 4-phenylbutyric acid enhances the mineralization of osteogenesis imperfecta iPSC-derived
osteoblasts.
Takeyari S, Kubota T, Ohata Y, Fujiwara M, Kitaoka T, Taga Y, Mizuno K, Ozono K.
J Biol Chem, 296 : 100027, 2020.

●49） Incidence rate of vitamin D deficiency and FGF23 levels in 12- to 13-year-old adolescents in Japan.
Koyama S, Kubota T, Naganuma J, Arisaka O, Ozono K, Yoshihara S.
J Bone Miner Metab, 39（3） : 456-462, 2020.

●50） Case Report : Efficacy of Reduced Doses of Asfotase Alfa Replacement Therapy in an Infant with Hypophosphatasia Who Lacked Severe Clinical Symptoms.
Fujisawa Y, Kitaoka T, Ono H, Nakashima S, Ozono K, Ogata T.
Front Endocrinol（ Lausanne）, 11 : 590455, 2020.

●51） Alkaline phosphatase in pediatric patients with genu varum caused by vitamin D-deficient rickets.
Mukai M, Yamamoto T, Takeyari S, Ohata Y, Kitaoka T, Kubota T, Yamamoto K, Kijima E, Hasegawa Y,
Michigami T, Ozono K.
Endocr J, 2021. Epub.

●52） Identification of children with chronic kidney disease through school urinary screening using
urinary protein / creatinine ratio measurement : an observational study.
Kajiwara N, Hayashi K, Fujiwara M, Nakayama H, Ozaki Y.
Clin Exp Nephrol, 24（5） : 450-457, 2020.

●53） Increased S1P expression in osteoclasts enhances bone formation in an animal model of Paget's disease.
Nagata Y, Miyagawa K, Ohata Y, Petrusca DN, Pagnotti GM, Mohammad KS, Guise TA, Windle JJ, David RG, Kurihara N.
J Cell Biochem, 122（3-4） : 335-348, 2021.

●54） Adult-onset leukoencephalopathy with homozygous LAMB1 missense mutation.
Yasuda R, Yoshida T, Mizuta I, Watanabe M, Nakano M, Sato R, Tokuda Y, Omi N, Sakai N, Nakagawa M,
Tashiro K, Mizuno T.
Neurol Genet, 6（4） : e442, 2020.

●55） Galactosialidosis Type IIb with Bilateral Macular Cherry-Red Spots but Mild Dysfunction.
Fukuyo H, Inoue Y, Takahashi H, Hatano Y, Shibuya T, Sakai N, Kawashima H.
Case Rep Ophthalmol, 11（2） : 306-314, 2020.

●56） Genetic and environmental variation in educational attainment : an individual-based analysis of 28 twin cohorts.
Silventoinen K, Jelenkovic A, Sund R, Latvala A, Honda C, Inui F, Tomizawa R, Watanabe M, Sakai N,
Rebato E, Busjahn A, Tyler J, Hopper JL, Ordoñana JR, Sánchez-Romera JF, Colodro-Conde L, Calais-Ferreira L, Oliveira VC, Ferreira PH, Medda E, Nisticò L, Toccaceli V, Derom CA, Vlietinck RF,
Loos RJF, Siribaddana SH, Hotopf M, Sumathipala A, Rijsdijk F, Duncan GE, Buchwald D, Tynelius P,
Rasmussen F, Tan Q, Zhang D, Pang Z, Magnusson PKE, Pedersen NL, Dahl Aslan AK, Hwang AE, Mack TM, Krueger RF, McGue M, Pahlen S, Brandt I, Nilsen TS, Harris JR, Martin NG, Medland SE, Montgomery GW, Willemsen G, Bartels M, van Beijsterveldt CEM, Franz CE, Kremen WS, Lyons MJ,
Silberg JL, Maes HH, Kandler C, Nelson TL, Whitfield KE, Corley RP, Huibregtse BM, Gatz M, Butler DA, Tarnoki AD, Tarnoki DL, Park HA, Lee J, Lee SJ, Sung J, Yokoyama Y, Sørensen TIA, Boomsma DI, Kaprio J.
Sci Rep, 10（1） : 12681, 2020.

●57） A Phase 2/3 Trial of Pabinafusp Alfa, IDS Fused with Anti-Human Transferrin Receptor Antibody, Targeting Neurodegeneration in MPS-II.
Okuyama T, Eto Y, Sakai N, Nakamura K, Yamamoto T, Yamaoka M, Ikeda T, So S, Tanizawa K, Sonoda H,
Sato Y.
Mol Ther, 30 : S1525-0016（20）30496-2, 2020.

●58） Early detection of Niemann - pick disease type C with cataplexy and orexin levels : continuous observation with and without Miglustat.
Imanishi A, Kawazoe T, Hamada Y, Kumagai T, Tsutsui K, Sakai N, Eto K, Noguchi A, Shimizu T, Takahashi T, Han G, Mishima K, Kanbayashi T, Kondo H.
Orphanet J Rare, 15（1） : 269, 2020.

●59） A Case of Adult Krabbe Disease that was Successfully Treated with Intravenous Immunoglobulin.
Fukazawa R, Takeuchi H, Oka N, Shibuya T, Sakai N, Fujii A.
Intern Med, 60（8） : 1283-1286, 2020.

●60） Leigh Syndrome Due to NDUFV1 Mutations Initially Presenting as LBSL.
Borna NN, Kishita Y, Sakai N, Hamada Y, Kamagata K, Kohda M, Ohtake A, Murayama K, Okazaki Y.
Genes（Basel）, 11（11） : 1325, 2020.

●61） Experiences of patients with lysosomal storage disorders treated with enzyme replacement
therapy : a qualitative systematic review protocol.
Koto Y, Ueki S, Yamakawa M, Sakai N.
JBI Evid Synth, 19（3） : 702-708, 2020.

●62） Heritability and Environmental Correlation of Phase Angle with Anthropometric Measurements : A Twin Study.
Matsumoto D, Inui F, Honda C, Tomizawa R, Watanabe M, Silventoinen K, Sakai N.
Int J Environ Res Public Health, 17（21） : 7810, 2020.

●63） Safety of intrathecal delivery of recombinant human arylsulfatase A in children with metachromatic leukodystrophy : Results from a phase 1 / 2 clinical trial.
Í Dali C, Sevin C, Krägeloh-Mann I, Giugliani R, Sakai N, Wu J, Wasilewski M.
Mol Genet Metab, 131（1-2） : 235-244, 2020.

●64） Urinary mulberry bodies as a potential biomarker for early diagnosis and efficacy assessment of enzyme replacement therapy in Fabry nephropathy.
Yonishi H, Namba-Hamano T, Hamano T, Hotta M, Nakamura J, Sakai S, Minami S, Yamamoto T, Takahashi A, Kobayashi W, Maeda I, Hidaka Y, Takabatake Y, Sakai N, Isaka Y.
Nephrol Dial Transplant, 24 ; gfaa298, 2020.

●65） A case of infantile Tay-Sachs disease with late onset spasms.
Yamamoto N, Kuki I, Nagase S, Inoue T, Nukui M, Okazaki S, Furuichi Y, Adachi K, Nanba E, Sakai N,
Kawawaki H.
Brain Dev, 43（5） : 661-665, 2020.

●66） Lenticular nuclei to thalamic ratio on PET is useful for diagnosis of GLUT1 deficiency syndrome.
Natsume J, Ishihara N, Azuma Y, Nakata T, Takeuchi T, Tanaka M, Sakaguchi Y, Okai Y, Ito Y, Yamamoto H,
Ohno A, Kidokoro H, Hattori A, Nabatame S, Kato K.
Brain Dev, 43（1） : 69-77, 2020.

●67） Mono-allelic and Bi-allelic Variants in NCDN Cause Neurodevelopmental Delay, Intellectual Disability and Epilepsy.
Fatima A, Hoeber J, Schuster J, Koshimizu E, Maya-Gonzalez C, Keren B, Mignot C, Akram T, Ali Z,
Miyatake S, Tanigawa J, Koike T, Kato M, Murakami Y, Abdullah U, Ali MA, Fadoul R, Laan L, Castillejo-López C, Liik M, Jin Z, Birnir B, Matsumoto N, Baig SM, Klar J, and Dahl N.
Am J Hum Genet, 108（4） : 739-748, 2021.

●68） High-dose pyridoxine treatment for inherited glycosylphosphatidylinositol deficiency.
Tanigawa J, Nabatame S, Tominaga K, Nishimura Y, Maegaki Y, Kinoshita T, Murakami Y, Ozono K.
Brain Dev, 43（6） : 680-687, 2021.

●69） Effectiveness of a Teacher Training Program for Students with Symptoms of Developmental
Disorders : Data from a Correspondence High School in Japan.
Ishii A, Okuno H, Nakaoka T, Iwasaka H, Taniike M.
Int J Environ Res Public Health, 17（9） : 3100, 2020.

●70） First night effect on polysomnographic sleep bruxism diagnosis varies among young subjects with different degrees of rhythmic masticatory muscle activity.
Haraki S, Tsujisaka A, Toyota R, Shiraishi Y, Adachi H, Ishigaki S, Yatani H,Taniike M, Kato T.
Sleep Med, 75 : 395-400, 2020.

●71） Polysomnographic analysis of respiratory events during sleep in young nonobese Japanese adults without clinical complaints of sleep apnea.
Okura M, Nonoue S, Tsujisaka A, Haraki S, Yokoe C, Taniike M, Kato T.
J Clin Sleep Med, 16（8） : 1303-1310, 2020.

●72） Examining the Treatment Efficacy of PEERS in Japan : Improving Social Skills Among Adolescents with Autism Spectrum Disorder.
Yamada T, Miura Y, Oi M, Akatsuka N, Tanaka K, Tsukidate N, Yamamoto T, Okuno H, Nakanishi M,
Taniike M, Mohri I, Laugeson EA.
J Autism Dev Disord, 50（3） : 976-997, 2020.

●73） APOE2 is associated with longevity independent of Alzheimer's disease.
Shinohara M, Kanekiyo T, Tachibana M, Kurti A, Shinohara M, Fu Y, Zhao J, Han X, Sullivan PM, Rebeck WG, Fryer JD, Heckman MG, Bu G.
elife, 19 ; 9 : e62199, 2020.

●74） Promising Effect of a New Ketogenic Diet Regimen in Patients with Advanced Cancer.
Hagihara K, Kajimoto K, Osaga S, Nagai N, Shimosegawa E, Nakata H, Saito H, Nakano M, Takeuchi M,
Kanki H, Kagitani-Shimono K, Kijima T.
Nutrients, 12（5） : 1473, 2020.

●75） An Interactive Smartphone App, Nenne Navi, for Improving Children's Sleep : Pilot Usability Study.
Yoshizaki A, Mohri I, Yamamoto T, Shirota A, Okada S, Murata E, Hoshino K, Kato-Nishimura K, Matsuzawa S, Kato T, Taniike M.
JMIR Pediatr Parent, 3（2） : e22102, 2020.

●76） Clinical evaluation of neuroinflammation in child-onset focal epilepsy : a translocator protein PET study.
Kagitani-Shimono K, Kato H, Hayashi R, Tominaga K, Nabatame S, Kishima H, Hatazawa J, Taniike M.

J Neuroinflammation, 18（1）: 8, 2021.

●77） Sleep and the General Behavior of Infants and Parents during the Closure of Schools as a Result of the COVID-19 Pandemic : Comparison with 2019 Data.
Shinomiya Y, Yoshizaki A, Murata E, Fujisawa TX, Taniike M, Mohri I.
Children, 8（2）: 168, 2021.