

III. 研究成果の刊行に関する一覧表

書籍

著者氏名	書籍全体の編集者名	書籍名	出版社名	出版年
ムコ多糖症(MPS)型診療ガイドライン作成委員会	ライソゾーム(ファブリ病含む)に関する調査研究班	ムコ多糖症(MPS)型診療ガイドライン2017	株式会社診断と治療社	2017年3月
副腎白質ジストロフィー(ALD)診療ガイドライン作成委員会	ライソゾーム(ファブリ病含む)に関する調査研究班	副腎白質ジストロフィー(ALD)診療ガイドライン2017	株式会社診断と治療社	2017年3月
ボンペ病診療ガイドライン作成委員会	ライソゾーム(ファブリ病含む)に関する調査研究班	ボンペ病診療ガイドライン2017	株式会社診断と治療社	2017年3月
酒井規夫		ライソゾーム病・ペルオキシソーム病診断の手引き	診断と治療社	2015年3月
酒井規夫	大園恵一, 金子一成編	これでOK小児救急ケーススタディ	診断と治療社	2015年3月
酒井規夫	井田博幸編	急性呼吸困難で救急外来受診氏診断されたゴーシェ病I型の1症例, ゴーシェ病症例集	Medical Tribune	2015年5月
酒井規夫	大野耕策編	肝脾腫の特徴, カタプレキシー, ニーマンピック病C型の診断と治療	医薬ジャーナル	2015年6月
酒井規夫	衛藤義勝編	組織障害の軽症な時期からERTを開始することの重要性を示唆する1症例, ファブリー病症例集	Medical Tribune	2015年12月

雑誌

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Kuranobu N, Murakami J, Okamoto K, Nishimura R, Murayama K, Takamura A, Umeda T, Eto Y, Kanzaki S.	Cholesterol ester storage disease with a novel LIPA mutation (L264P) that presented massive hepatomegaly: A case report.	Hepatol Res	2016 Mar; 46(5)	477-82	2016

Yokoi T, Yokoi K, Akiyama K, Higuchi T, Shimada Y, Kobayashi H, Sato T, Ohteki T, Ohtsu M, Nakauchi H, Ida H, Ohashi T.	Non-myeloablative preconditioning with ACK2 (anti-c-kit antibody) is efficient in bone marrow transplantation for murine models of mucopolysaccharidosis type II.	Mol Genet Metab	2016 Nov; 119(3)	232-238	2016
Pastores GM, Turkia HB, Gonzalez DE, Ida H, Tantony AA, G, Qin Y, Dirh Q, Zimran A.	Development of anti-velaglutamase alfa antibodies in clinical trial-treated patients with Gaucher disease.	Blood Cell Dis	Mol and Dis.59	37-43	2016
小林博司	ガングリオシド蓄積症～GM1・GM2ガングリオシドーシス	小児内科2016	vol.48増刊 小児疾患 診療のための 病態生理	p158-164	2016
小林博司	筋型糖原病～Pompe病を中心に	小児内科2016	vol.48 No.12 特集 小児の筋疾患	p1972-1977	2016
Sato Y, Kobayashi H, Higuchi T, Shimada Y, Era T, Kimura S, Eto Y, Ida H, Ohashi T.	Disease modeling and lentiviral gene transfer in patient-specific induced pluripotent stem cells from late-onset Pompe disease patient.	Mol Ther Methods Clin Dev.	2015 Jul 8	2	2015
Saito O, Kusano E, Akimoto T, Asano Y, Kitagawa T, Suzuki K, Ishige N, Akiba T, Saito A, Ishimura E, Hattori M, Hisada A, Guili C, Maruyama H, Kobayashi M, Ohashi T, Matsuda I, Eto Y.	Prevalence of Fabry disease in dialysis patients: Japan Fabry disease screening study (J-FAST).	Clin Exp Nephrol.	2016 Apr	20(2) 284-93	2016
Shimada Y, Wakabayashi T, Akiyama K, Hoshina H, Higuchi T, Kobayashi H, Eto Y, Ida H, Ohashi T.	A method for measuring disease-specific iduronic acid from the non-reducing end of glycosaminoglycan in mucopolysaccharidosis type II mice.	Mol Genet Metab.	2016 Feb;	140-3117(2)	2016
Wakabayashi T, Shimada Y, Akiyama K, Higuchi T, Fukuda T, Kobayashi H, Eto Y, Ida H, Ohashi T.	Hematopoietic Stem Cell Gene Therapy Corrects Neuroprotective Phenotype in Murine Model of Mucopolysaccharidosis Type II.	Hum Gene Ther.	26(6)	357-66	2015

Hossain MA, Otomo T, Saito S, Ohno K, Sakuraba H, Hamada Y, Ozono K, <u>Sakai N.</u>	Late-onset Krabbe disease is predominant in Japan and its mutant precursor protein undergoes more effective processing than the infantile-onset form.	<i>Gene</i>	534(2)	144-54	2014
Hossain MA, Higaki K, Saito S, Ohno K, Sakuraba H, Nanba E, Suzuki Y, Ozono K, <u>Sakai N.</u>	Chaperone therapy for Krabbe disease: potential for late-onset GALC mutations.	2015 Sep <i>J Hum Genet</i>	60(9)	539-45	2015
Hossain MA, Higaki K, Shinpo M, Nanba E, Suzuki Y, Ozono K, <u>Sakai N.</u>	Chemical chaperone treatment for galactosialidosis: Effect of N-ethylmaleimide on $\beta$ -galactosidase activities in fibroblasts.	<i>Brain Dev</i>	38(2)	175-80	2016
Shibazaki T, Hirabayashi K, Saito S, Shigemura T, Nakazawa Y, Sakashita K, Takagi M, Shiohara M, Adachi K, Nanba E, <u>Sakai N</u> , Koike K.	Clinical and laboratory outcomes after umbilical cord blood transplantation in a patient with mucopolipidosis II alpha/beta.	<i>Am J Med Genet A</i>	170A(5)	1278-82	2016
Kato S, Yabe H, Takakura H, Mugiishi H, Ishige M, Tanaka A, Kato K, Yoshida N, Adachi S, <u>Sakai N</u> , Hashii Y, Ohashi T, Sasahara Y, Suzuki Y, Tabuchi K.	Hematopoietic stem cell transplantation for inborn errors of metabolism: A report from the Research Committee on Transplantation for Inborn Errors of Metabolism of the Japanese Ministry of Health, Labour and Welfare and the Working Group of the Japanese Society for Hematopoietic Cell Transplantation.	<i>Pediatr Transplant</i>	20(2)	203-14	2016
Mamada N, Nakamagoe K, Shioyama A, Furuta J, <u>Sakai N</u> , Ishii A, Tamakawa A.	Adult-onset Krabbe disease presenting as acute hemiparesis and progressive demyelination detected by diffusion-weighted imaging.	<i>J Neurol Sci</i>	367	326-8	2016
<u>Sakai N</u> , Otomo T.	Challenge of phenotype estimation for optimal treatment of Krabbe disease.		94(11)	1025-30	2016
Yoshimura A, Kibe T, Irahara K, <u>Sakai N</u> , Yokochi K.	Predominant Corticospinal Tract Involvement in a Late Infant with Krabbe Disease.	<i>Jpn Clin Med</i>	7	23-6	2016

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酒井規夫	ファブリー病 小児科		57(3)	235-240	2016
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Matsukawa T, Koshi KM, Mitsui J, et al.	Slowly progressive $\alpha$ -mannosidase B deficiency with survival to adulthood diagnosed by whole-exome sequencing.	J Neurol Sci	2017. 372	6-10	2017
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