別紙４

研究成果の刊行に関する一覧表

書籍

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
Fukao T, Harding CO	Chapter 10 Ketone Synthesis and Utilization Defects	Sarafoglou K, Hoffmann GF, Roth KS	Pediatric Endocrinology and Inborn Errors of Metabolism	McGraw Hill Education	NewYork	2017	145-160
杉江秀夫 杉江陽子	(6)糖原病 (7)先天性糖質代謝異常症	矢﨑義雄 総編集	内科学11版	朝倉書店	東京	2017年	発行予定
小国弘量	薬物治療：小児期	日本てんかん学会	てんかん白書	南江堂	東京	2016	48-49
野口篤子	リジン尿性蛋白不耐症	日本小児科学会監修	小児慢性特定疾病―診断の手引き―	診断と治療社		2016	
深尾敏幸	ケトン性低血糖･アセトン血性嘔吐症.	五十嵐隆	小児科診療ガイドライン―最新の診療指針―	総合診療社	東京	2016	413-417

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
Yamada K, Kobayashi H, Bo R, Purevsuren J, Mushimoto Y, Takahashi T, Hasegawa Y, Taketani T, Fukuda S, Yamaguchi S	Efficacy of bezafibrate on fibroblasts of glutaric acidemia type II patients evaluated using an in vitro probe acylcarnitine assay	Brain & Development	39（1）	48-57	2017
Tanaka K, Nakamura K*, Matsumoto S, Kido J, Mitsubuchi H, Ohura T, Endo F	Citrulline administration for urea cycle disorders in Japan.	Pediatrics Int.	59	422-426	2017

Mori H, Momosaki K, Kido J, Tamura H, Tanaka K, Matsumoto S, Nakamura K, Mitsubuchi H, Endo F, Iwai M*	Amelioration of Brain Damage by Glycine in Neonatal Rat Brain Following Hypoxia-Ischemia.	Pediatrics Int	59	321–327	2017
Kido J, Kawasaki T, Mitsubuchi H, Kamohara H, Ohba T, Matsumoto S, Endo F, Nakamura K*	Hyperammonemia crisis following parturition in a female patient with ornithine transcarbamylase deficiency.	World J Hepatol	9	343-348	2017
Abdelkreem E, Akella R, Dave U, Sane S, Osuka H, Sasai H, Aoyama Y, Nakama M, Ohnishi H, Mahmoud S, Abd El Aal M, **Fukao T**	Clinical and mutational characterizations of 10 Indian patients with beta-ketothiolase deficiency.	JIMD reports,		DOI 10.1007/8904_2016_26	2017
Nguyen KN, Abdelkreem E, Colombo R, Hasegawa Y, Can NT, Bui TP, Le HT, Tran MT, Nguyen HT, Trinh HT, Aoyama Y, Sasai H, Yamaguchi S, Fukao T, Vu DC	Characterization and outcome of 41 patients with beta-ketothiolase deficiency: 10 years' experience of a medical center in northern Vietnam.	J Inherited Metab Dis		doi: 10.1007/s10545-017-0026-6	2017

Tanigawa J, Mimatsu H, Mizuno S, Okamoto N, Fukushi D, Tominaga K, Kidokoro H, Muramatsu Y, Nishi E, Nakamura S, Motooka D, Nomura N, Hayasaka K, Niihori T, Aoki Y, Nabatame S, Hayakawa M, Natsume J, Ozono K, Kinoshita T, Wakamatsu N, Murakami Y.	Phenotype-genotype correlations of PIGO deficiency with variable phenotypes from infantile lethality to mild learning difficulties	Hum Mut			2017
Kido J, Matsumoto S, Sakamoto R, Mitsubuchi H, Endo F and Nakamura K*	Liver transplantation may prevent neurodevelopmental deterioration in high risk patients with urea cycle disorders.	Pediatric Transplantation			(in press)
Kido J, Matsumoto S, Momosaki K, Sakamoto R, Mitsubuchi H, Inomata Y, Endo F, and Nakamura K*	Plasma exchange and chelator therapy rescues acute liver failure in Wilson disease without liver transplantation.	Hepatology Research			(in press)
Kido J, Yoshida T, Mitsubuchi H, Matsumoto S, Endo F and Nakamura K*	Clinical manifestations in two patients with pyruvate dehydrogenase deficiency and long-term survival.	Human Genome Variation			(in press)
Katata Y, Uematsu M, Sato H, Suzuki S, Nakayama T, Kubota Y, Kobayashi T, Hino-Fukuyo N, Saitsu H, Kure S.	Novel missense mutation in CLN8 in late infantile neuronal ceroid lipofuscinosis: The first report of a CLN8 mutation in Japan.	Brain Dev.	38	341–345	2016

Yamada K, Kobayashi H, Bo R, Takahashi T, Purevsuren J, Hasegawa Y, Taketani T, Fukuda S, Ohkubo T, Yokota T, Watanabe M, Tsunemi T, Mizusawa H, Takuma H, Shioya A, Ishii A, Tamaoka A, Shigematsu Y, Sugie H, Yamaguchi S	Clinical, biochemical and molecular investigation of adult-onset glutaric acidemia type II: Characteristics in comparison with pediatric cases	Brain & Development	38 (3)	293-301	2016
Togawa T, Sugiura T, Ito K, Endo T, Aoyama K, Ohashi K, Negishi Y, Kudo T, Ito R, Kikuchi A, Arai-Ichinoi N, Kure S, Saitoh S.	Molecular genetic dissection and neonatal/infantile intrahepatic cholestasis using targeted next-generation sequencing.	J Pediatr.	171	171-177	2016
Otsuka H, Sasai H, Abdelkreem E, Kawamoto N, Kawamoto M, Kamiya T, Tanimoto Y, Kikuchi A, Kure S, Numakura C, Hayasaka K, **Fukao T**	Effectiveness of Medium-Chain Triglyceride Oil Therapy in Two Japanese Citrin-Deficient Siblings: Evaluation Using Oral Glucose Tolerance Tests	Tohoku J Exp Med	240	323-328	2016
Otsuka H, Sasai H, Nakama M, Aoyama Y, Abdelkreem E, Ohnishi H, Konstantopoulou V, Sass JO, **Fukao T**	Exon 10 skipping in *ACAT1* caused by a novel mutation (c.949G>A) located at an exonic splice enhancer site	Mol Med Rep	14	4906-4910	2016

Purevsuren J, Bolormaa B, Narantsetseg C, Batsolongo R, Enkhchimeg O, Bayalag M, Hasegawa Y, Shintaku H, Yamaguchi S	The first Mongolian cases of phenylketonuria in selective screening of inborn errors of metabolism.	MGMR	9	71-74	2016
Bo R, Hasegawa Y, Yamada K, Kobayashi H, Taketani T, Fukuda S, Yamaguchi S	A fetus with mitochondrial trifunctional protein deficiency: Elevation of 3-OH-acylcarnitines in amniotic fluid functionally assured the genetic diagnosis	Molecular Genetics and Metabolism Reports	6	1-4	2016
Mashima R, Sakai E, Kosuga M, Okuyama T.	Levels of enzyme activities in six lysosomal storage diseases in Japanese neonates determined by liquid chromatography-tandem mass spectrometry.	Mol Genet Metab Rep.	9	6-11	2016
Mashima R, Tanaka M, Sakai E, Nakajima H, Kumagai T, Kosuga M, Okuyama T.	A selective detection of lysophosphatidylcholine in dried blood spots for diagnosis of adrenoleukodystrophy by LC-MS/MS.	Mol Genet Metab Rep.	7	16-19.	2016
Mashima R, Sakai E, Tanaka M, Kosuga M, Okuyama T	The levels of urinary glycosaminoglycans of patients with attenuated and severe type of mucopolysaccharidosis II determined by liquid chromatography-tandem mass spectrometry.	Mol Genet Metab Rep.	7	87-91	2016

Matsuoka T, Miwa Y, Tajika M, Sawada M, Fujimaki K, Soga T, Tomita H, Uemura S, Nishino I, Fukuda T, Sugie H, Kosuga M, Okuyama T, Umeda Y.	Divergent clinical outcomes of alpha-glucosidase enzyme replacement therapy in two siblings with infantile-onset Pompe disease treated in the symptomatic or pre-symptomatic state.	Mol Genet Metab Rep.	18(9)	98-105.	2016
Erdol S, Türe M, Yakut T, Saglam H, Sasai H, Abdelkreem E, Ohtsuka H, **Fukao T**	A Turkish patient with Succinyl-CoA:3-oxoacid CoA transferase deficiency mimicking diabetic ketoacidosis.	Journal of Inborn Errors of Metabolism and Screening		DOI: 10.1177/2326409816651281	2016
Akagawa S, **Fukao T**, Akagawa Y, Sasai H, Kohdera U, Kino M, Shigematsu Y, Aoyama Y, Kaneko K	Japanese male siblings with 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (HSD10 disease) without neurological regression.	JIMD reports		DOI: 10.1007/8904_2016_570	2016
Takano H, Ishihara T, Kosuga M, Okuyama T.	A Senile Case of Late-onset Pompe's Disease.	Intern Med.	55(18)	2723-5	2016
Kosuga M, Mashima R, Hirakiyama A, Fuji N, Kumagai T, Seo J-H, Nikaido M, Saito S, Ohno K, Sakuraba H, Okuyama T.	Molecular diagnosis of 65 families with mucopolysaccharidosis type II (Hunter syndrome) characterized 16 novel mutations in the *IDS* gene: genetic, pathological and structural studies of iduronate-2-sulfatase.	Mol Genet Metab	118	190-197	2016

Noguchi A, Nakamura K, Murayama K, Yamamoto S, Komatsu H, Kizu R, Takayanagi M, Okuyama T, Endo F, Takasago Y, Shoji Y, Takahashi T.	Clinical and genetic features of Japanese patients with lysinuric protein intolerance	Pediatr Int.	58(10)	979-983	2016
Onouchi et al.	Variations in ORAI1 gene associated with Kawasaki disease	PLoS One	11	e0145486	2016
Fujita M et al.	Income Related Inequality of Health Care Access in Japan: A Retrospective Cohort Study	PLoS One	11	e0151690	2016
Yamamoto M	Survey of motivation to participate in a birth cohort	J Hum Genet	61	787-791	2016
Mori-Yoshimura M, Segawa K, Minami N, Oya Y, Komaki H, Nonaka I, Nishino I, Murata M	Cardiopulmonary dysfunction in patients with limb-girdle muscular dystrophy 2A.	Muscle Nerve		doi: 10.1002/mus.25369. PMID: 27500519	[Epub ahead of print]

Miyake N, Fukai R, Ohba C, Chihara T, Miura M, Shimizu H, Kakita A, Imagawa E, Shiina M, Ogata K, Okuno-Yuguchi J, Fueki N, Ogiso Y, Suzumura H, Watabe Y, Imataka G, Leong HY, Fattal-Valevski A, Kramer U, Miyatake S, Kato M, Okamoto N, Sato Y, Mitsuhashi S, Nishino I, Kaneko N, Nishiyama A, Tamura T, Mizuguchi T, Nakashima M, Tanaka F, Saitsu H, Matsumoto N	Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy.	Am J Hum Genet.	99(4)	950-961 doi: 10.1016/j.ajhg.2016.08.005. PMID: 27666374	Oct, 2016
Preethish-Kumar V, Pogoryelova O, Polavarapu K, Gayathri N, Seena V, Hudson J, Nishino I, Prasad C, Lochmuller H, Nalini A	Beevor's sign: a potential clinical marker for GNE myopathy.	Eur J Neurol.	23(8)	e46-8 doi: 10.1111/ene.13041. PMID: 27431025	Aug, 2016
Junji Azuma, Shin Nabatame, Toshiya Katsura, Kyoko Yamamoto, Hiroshi Kaneno, Eri Kijima, Yoshimi Mizoguchi, Tunesuke Shimotsuji , Takehisa Yamamoto, Keiichi Ozono.	Marked elevation of urinary β2-microglobulin in patients with reversible splenial lesions: A small case series.	J Neurol Sci	368	109-12	2016

Nakamura K*, Kido J, Matsumoto S, Mitsubuchi H and Endo F	Clinical manifestations and growth of patients with urea cycle disorders in Japan	J. Hum. Genet	61	613-616	2016
Kido J, Mitsubuchi H, Ito F, Yoshida T, Matsumoto S, Sakamoto R, Endo F and Nakamura K*	Advanced endometrial cancer in phenylketonuria.	Medical Science Case Reports	3	108-111	2016
Sakamoto R, Nakamura K*, Kido J, Matsumoto S, Mitsubuchi H, Inomata Y, Endo F	Improvement in the prognosis and development of patients with methylmalonic acidemia after living donor liver transplant.	Pediatric Transplantation	20	1081-1086	2016
Kido J, Matsumoto S, Sakamoto R, Mitsubuchi H, Endo F and Nakamura K*	Pulmonary artery hypertension in methylmalonic academia.	Hemodialysis International		doi: 10.1111/hdi. 12506. [Epub ahead of print]	2016
杉江秀夫、杉江陽子	**精神医学症候群(第2 版) Ⅰ糖質代謝異常症**	**別冊日本臨牀新領域別症候群シリーズ** No.37 (	No37	161-166	2017
坂本修、市野井那津子、呉繁夫	**新生児マススクリーニングC3, C3/C2陽性を契機に発見されたプロピオン酸血症 - PCCA変異例とPCCB p.Y435C変異例との比較**	**日本マススクリーニング学会誌**	26	59-64	2016
吉田美智子、三上仁、池田秀之、梅木郁美、西野美奈子、星能元、島岡理、市野井那津子、菊池敦生、呉繁夫	**新生児マススクリーニングでのシトルリン/セリン比が早期診断の契機となったシトリン欠損症**	**岩手県立病院医学会雑誌**	56	55-59	2016
小国弘量	**ケトン食によるてんかんの治療（総説）**	**小児科**	57:	1033-10388 5	2016
小国弘量	**小児の薬物治療**	**クリニシアン.**	63	605-610	2016

小国弘量	小児難治性てんかんにおける成人期移行の問題	小児科臨床	69	729-733	2016
吉永 治美, 小国弘量	小児神経疾患における活性型ビタミンB6の意義	脳と発達	48	114-116	2016
大浦敏博、坂本修、岡野善行	シトリン欠損症	小児科診療	79	805-811	2016
高柳 正樹.	【小児慢性疾患の成人期移行の現状と問題点】先天性代謝異常　糖原病.	小児科臨床	69	p684-688	2016
杉江秀夫、杉江陽子	【筋ジストロフィー・筋疾患-最近の進歩】 代謝性ミオパチーの治　療現状と未来　筋型糖原病の治療戦略　病態からみた治療の進歩	医学の歩み	259	133-139	2016
杉江 秀夫	【肝胆膵の指定難病を整理する】 平成27年7月1日施行の指定難病　肝型糖原病(肝型グリコーゲン代謝異常症)	肝・胆・膵	72	699-705	2016
青天目信，水島昇	シンポジウム 5：オートファジーと小児神経疾患　序論	脳と発達	48(3)	174-6	2016
青天目信	てんかん食の調整と副作用	Epilepsy	10(2)	111-112	2016
野口 篤子, 高橋勉	【肝胆膵の指定難病を整理する】 平成27年7月1日施行の指定難病リジン尿性蛋白不耐症	肝・胆・膵	72巻4号	677-683	2016