

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
新宅治夫	カテコールアミン・セロトニン代謝異常、	水澤英洋	別冊日本臨床新領域別症候群シリーズNo.28 神経症候群(第2版)その他の神経疾患を含めて	日本臨床	大阪	2014	615-621
伊藤康、小国弘量	グルコーストランスポーター1欠損症症後群		神経症候群(第2版)-その他の神経疾患を含めて-先天代謝異常症 別冊 日本臨床 新領域別症候群シリーズ No.28	日本臨床社	大阪	2014	823-826
伊藤康、小国弘量	グルコーストランスポーター1(GLUT-1)欠損症症候群.		神経症候群 VI(第2版)-その他の神経疾患を含めて-てんかん症候群 別冊 新領域別症候群シリーズ No.31	日本臨床社	大阪	2014	464-467
小国弘量	ミオクロニー(失立)脱力発作てんかん		神経症候群 VI(第2版)-その他の神経疾患を含めて-てんかん症候群 別冊 新領域別症候群シリーズ No.31	日本臨床社	大阪	2014	122-125
大浦敏博	特殊ミルク	位田忍他	先天代謝異常症 栄養食事指導ケースブック	診断と治療社	東京	2014年	128-132
大浦敏博	先天代謝異常症の食事療法に併用される経口製剤	位田忍他	先天代謝異常症 栄養食事指導ケースブック	診断と治療社	東京	2014年	133-134

高柳正樹	有機酸代謝異常症	日本先天代謝異常学会	引いて調べる先天代謝異常症	診断と治療	東京	2014	14-17
杉江秀夫	筋型糖原病	杉江秀夫	代謝性ミオパチー	診断と治療社	東京	2014年	1-9 31-41 66-68 75-76 79-82 242-251
青天目信	錐体外路症候(錐体外路障害)	遠藤文夫,井田博幸,山口清次,高柳正樹,深尾敏幸	引いて調べる先天代謝異常症	診断と治療社	東京	2014	74-75
青天目信	錐体路症候(錐体路障害)	同上	同上	同上	同上	同上	75-76
青天目信	統合失調様症状	同上	同上	同上	同上	同上	80-81
青天目信	発達退行	同上	同上	同上	同上	同上	85
杉江秀夫	筋型糖原病	永井良三	神経内科研修ノート	診断と治療社	東京	2015年	402-404

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Naiki M, Ochi N, Kato YS, Purevsuren J, Yamada K, Kimura R, Fukushi D, Hara S, Yamada Y, Kumagai T, Yamaguchi S, Wakamatsu N	Mutations in HADHB, which encodes the β -subunit of mitochondrial trifunctional protein, cause infantile onset hypoparathyroidism and peripheral polyneuropathy	American Journal of Medical Genetics A	164(5)	1180-1187	2014
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Ohtake A, Murayama, K, Mori M, Harashima H, Yamazaki T, Tamaru S, Yamashita Y, Kishita Y, Nakachi Y, Kohda M, Tokuzawa Y, Mizuno Y, Moriyama Y, Kato H, Okazaki Y	Diagnosis and molecular basis of mitochondrial respiratory chain disorders: exome sequencing for disease gene identification.	Biochim Biophys Acta	1840(4)	1355-1359	2014
Niizeki H, Shiohama A, Sasaki T, Seki A, Kabashima K, Otsuka A, Takeshita M, Hirakiyama A, Okuyama T, Tanese K, Ishiko A, Amagai M, Kudoh J.	The novel SLCO2A1 heterozygous missense mutation p.E427K and nonsense mutation p.R603* in a female patient with pachydermoperiostosis with an atypical phenotype.	Br J Dermatol.	170	1187-1189	2014
Vatanavicharn N, Yamada K, Aoyama Y, Fukao T, Densupsoontorn N, Jirapinyoe P, Sathienkijkanchai A, Yamaguchi S, Wasant P	Carnitine-acylcarnitine translocase deficiency: two neonatal cases with common splicing mutation and in vitro bezafibrate response	Brain Dev	inpress		2014
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Vatanavicharn N, Yamada K, Aoyama Y, Fukao T, Densupsoontorn N, Jirapinyo P, Sathienkijkanchai A, Yamaguchi S, Wasant P	Carnitine-acylcarnitine translocase deficiency: two neonatal cases with common splicing mutation and in vitro bezafibrate response.	Brain Dev	inpress		2014
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