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研究成果の刊行に関する一覧表

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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
小崎健次郎	先天異常症候群　成長に応じた診療のポイントと政策的支援の手引き	小崎健次郎他	先天異常症候群 成長に応じた診療のポイントと政策的支援の手引き	鳥影社	東京	2017	1-339
森崎裕子	血管型エーラス・ダンロス症候群	桜井章洋	遺伝カウンセリングマニュアル	南江堂	東京	2016	167-168
森崎裕子	遺伝性出血性毛細血管拡張症	桜井章洋	遺伝カウンセリングマニュアル	南江堂	東京	2016	168-169
森崎裕子	遺伝性肺動脈性肺高血圧症	桜井章洋	遺伝カウンセリングマニュアル	南江堂	東京	2016	170
森崎裕子	マルファン症候群、およびその類縁疾患	桜井章洋	遺伝カウンセリングマニュアル	南江堂	東京	2016	222-224
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仁科幸子	斜視、眼瞼の異常	松原洋一、呉繁夫、左合治彦	こどもの病気. 遺伝について聞かれたら	診断と治療社	東京	2015	162-164
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小崎里華	新技術と臨床医の経験の融合が重要		MMJ　2015年10月号	毎日新聞出版	東京都	2015	282-3
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松浦伸也	B-1 Fanconi貧血、B-2 Roberts症候群、B-3 Bloom症候群、B-4 PCS症候群/MVA症候群、B-5 Rothmund-Thomson症候群、B-6 毛細血管拡張性失調症、C-1 Hutchinson-Gilford症候群、C-3 Cockayne症候群、C-4 Werner症候群、G-1 Neu-Laxova症候群、J-4 COFS症候群、IV-1 正常者の身長・体重・成長曲線、IV-2 正常者の頭囲・眼間距離、IV-3 中手骨・指節骨の長さ	梶井正、黒木良和、新川詔夫監修	新先天奇形症候群アトラス　改訂第二版	南江堂	東京	2015	
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渡邊みお、仁科幸子	小児の診察、視反応、未熟児網膜症の診察	江口秀一郎	眼科外来処置・小手術クローズアップ	メジカルビュー	東京	2014	4-7
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Yaguchi Y, Katagiri S, Fukushima Y, Yokoi T, Nishina S, Kondo M, Azuma N.	Electroretinographic effects of retinal dragging and retinal folds in eyes with familial exudative vitreoretinopathy.	Sci Rep. 2016 Jul 26;6:30523. doi: 10.1038/srep30523.			2016
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吉田 朋世、仁科 幸子、萬束 恭子、赤池 祥子、越後貫 滋子、横井 匡、東 範行	乳児内斜視早期手術後の両眼視機能.	眼臨紀	10 (1)	58-63	2016
伊藤里美・仁科幸子	小児ロービジョンケア	眼科	58 (12)	1487-1492	2016
仁科 幸子	内斜視に対する手術治療.	眼科	58 (3)	251-257	2016
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Takano K*, Ogasawara N, Matsunaga T, Mutai H, Sakurai A, Ishikawa A, Himi T	A novel nonsense mutation in the NOG gene causes familial NOG-related symphalangism spectrum disorder	Hum Genome Variation	3	16023	2016
永井遼斗、松永達雄*	図説シリーズ「目で見る遺伝医学」－難聴の遺伝医学	国立医療学会誌「医療」	88(3)	240-247	2016
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Sato M, Baba Y, Haruyama N, Higashihori N, Tsuji M, Suzuki S, Moriyama K.	Clinicostatistical analysis of congenitally missing permanent teeth in Japanese patients with cleft lip and/or palate	Orthodontic Waves.	75(2)	41-45	2016
Mochida K, Amano M (corresponding author), Miyake N, Matsumoto N, Hatamochi A, Kosho T (corresponding author)	Dermatan 4-O-sulfotransferase 1-deficient Ehlers-Danlos syndrome complicated by a large subcutaneous hematoma on the back.	J Dermatol	43(7)	832-3	2016
Kono M (corresponding author), Hasegawa-Murakami Y, Sugiura K, Ono M, Toriyama K, Miyake N, Hatamochi A, Kamei Y, Kosho T (corresponding author), Akiyama M	A 45-year-old Woman with Ehlers-Danlos Syndrome Caused by Dermatan 4-O-sulfotransferase-1 Deficiency: Implications for Early Ageing.	Acta Derm Venereol	96(6)	830-1	2016

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古庄知己	Ehlers-Danlos症候群Kosho type .	小児科診療「日本人が貢献した先天異常症候群」	79(12)	1761-1769	2016
Kon M, Saio K, Mitsui T, Miyado M, Igarashi M, Moriya K, Nonomura K, Shinohara M, Ogata T, Fukami M	Copy-number variations of the azoospermia factor region or SRY are not associated with the risk of hypospadias.	Sex Dev	10(1)	43084	2016
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Saito K, Matsuzaki T, Iwasa T, Miyado M, Saito H, Hasegawa T, Homma K, Inoue E, Kubota T, Irahara M, Ogata T, Fukami M	Steroidogenic pathways involved in androgen biosynthesis in eumenorrheic women and patients with polycystic ovary syndrome.	J Steroid Biochem Mol Biol	158	31-37	2016
Fujisawa Y, Sakaguchi K, Ono H, Yamaguchi R, Kato F, Kagami M, Fukami M, Ogata T	Combined steroidogenic characters of fetal adrenal and Leydig cells in childhood adrenocortical carcinoma.	J Steroid Biochem Mol Biol	159	86-93	2016
Asahina M, Endoh Y, Matsubayashi T, Fukuda T, Ogata T	Novel RAB3GAP1 compound heterozygous mutations in Japanese siblings with Warburg Micro syndrome.	Brain Dev	38（3）	337-340	2016

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Okuno M, Yorifuji T, Kagami M, Ayabe T, Urakami T, Kawamura T, Kikuchi N, Yokota I, Toru Kikuchi, Amemiya S, Suzuki J, Ogata T, Sugihara S, Fukami M	The Japanese Study Group of Insulin Therapy for Childhood and Adolescent Diabetes (JSGIT) Chromosome 6q24 methylation defects are uncommon in childhood-onset non-autoimmune diabetes mellitus patients born appropriate-for-gestational age.	Clin Pediatr Endocrinol	25（3）	99-102	2016
Saito K, Matsuzaki T, Iwasa T, Miyado M, Saito H, Kubota T, Irahara M, Ogata T, Fukami M	Blood allopregnanolone levels in women with polycystic ovary syndrome.	Clin Endocrinol	85	151-152	2016
Luk H-M, Lo F-M I, Sano S, Matsbara K, Nakamura A, Ogata T, Kagami M	Silver-Russell syndrome in a patient with somatic mosaicism for upd(11)mat identified by buccal cell analysis.	Am J Med Genet A	170（7）	1938-1941	2016
Sano S, Nagasaki K, Kikuchi T, Nakabayashi K, Hata K, Fukami M, Kagami M, Ogata T	Beckwith-Wiedemann syndrome and pseudohypoparathyroidism type Ib in a patient with multilocus methylation defects: a female-dominant phenomenon?	J Hum Genet	61（8）	765-769	2016
Miyamichi D, Asahina M, Nakajima J, Sato M, Hosono K, Nomura T, Negishi T, Miyake N, Hotta Y, Ogata T, Matsumoto N	Novel HPS6 mutations identified by whole-exome sequencing in two Japanese sisters with suspected ocular albinism.	J Hum Genet	61（9）	839-842	2016

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Koyama Y, Homma K, Fukami M, Miwa M,Ikeda K, Ogata T, Murata M, Hasegawa T	Classic and non-classic 21-hydroxylase deficiency can be discriminated from P450 oxidoreductase deficiency in Japanese infants by urinary steroid metabolites.	Clin Pediatr Endocrinol	25（2）	37-44	2016
Miyoshi Y, Yorifuji T, Horikawa R, Takahashi I, Nagasaki K, Ishiguro H, Fujiwara I, Ito J, Oba M, Kawamoto H, Fujisaki H, Kato M, Shimizu C, Kato T, Matsumoto K, Sago H, Takimoto T, Okada H, Suzuki N, Yokoya S, Ogata T, Ozono K	Gonadal function, fertility, and reproductive medicine in childhood and adolescent cancer patients: a national survey of Japanese pediatric endocrinologists.	Clin Pediatr Endocrinol	25（2）	45-57	2016
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Fujisawa Y, Fukami M, Hasegawa T, Uematsu A, Muroya M, Ogata T	Long-term clinical course in three patients with MAMLD1 mutations.	Endocr J	63（9）	835-839	2016
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Montalbano A Juergensen A, Roeth R, Weiss B, Fukami M, Fricke-Otto S, Binder G, Ogata T, Decker E, Nuernberg G, Hassel 2, Rappold GA	Retinoic acid catabolizing enzyme CYP26C1 is a genetic modifier in SHOX deficiency.	EMBO Mol Med	8（12）	1455-1469	2016
Shima H, Yatsuga S, Nakamura A, Sano S, Sasaki T, Katsumata N, Suzuki E, Hata K, Nakabayashi K, Momozawa Y, Kubo M, Okamura K, Kure S, Matsubara Y, Ogata T, Narumi S, Fukami M	NR0B1 frameshift mutation in a boy with idiopathic central precocious puberty.	Sex Dev	10（4）	205-209	2016

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Miyado M, Inui M, Igarashi M, Katoh-Fukui Y, Takasawa K, Hakoda A, Kanno J, Kashimada K, Miyado K, Tamano M, Ogata T, Takada S, Fukami M	The p.R92W variant of NR5A1/Nr5a1 induces testicular development of 46,XX gonads in humans, but not in mice: Phenotypic comparison of human patients and mutation-induced mice.	Biol Sex Differ	56（7）	eCollection 10.1186/s13293-016-0114-6	2016
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Asahina M, Endoh Y, Matsubayashi T, Hirano K, Fukuda T, Ogata T	Genomewide array comparative genomic hybridization in 55 Japanese normokaryotypic patients with non-syndromic intellectual disability.	J Pediatr Neurol Disord	2(1)	108	2016

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Takenouchi T, Miura K, Uehara T, Mizuno S, Kosaki K.	Establishing SON in 21q22.11 as a cause a new syndromic form of intellectual disability: Possible contribution to Braddock-Carey syndrome phenotype.	American Journal of Medical Genetics	170	2587	2016
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岡本伸彦	日本が貢献した先天異常症候群　Coffin-Siris症候群	小児科診療	79	1807-1812	2016
岡本伸彦	結節性硬化症	小児内科	43	1520-1523	2016
岡本伸彦	Sener症候群	小児科診療	79	5-7	2016
岡本伸彦	染色体異常・先天奇形症候群 Smith-Lemli-Opitz症候群	小児科診療	79	6	2016
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Shimojima K, Ondo Y, Nishi E, Mizuno S, Ito M, Ioi A, Shimizu M, Sato M, Inoue M, Okamoto N, Yamamoto T	Loss-of-function mutations and global rearrangements in GPC3 in patients with Simpson-Golabi-Behmel syndrome	Hum Genome Var	3	16033	2016
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	950-961	2016

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