N. Hatabu, N. Katori, T. Sato, N. Maeda, E. Suzuki, O. Komiyama, H. Tsutsui, T. Nagao, H. Nakauchi-Takahashi, T. Matsunaga, T. Ishii, T. Hasegawa and K. Yamazawa	A Familial Case of a Whole Germline CDC73 Deletion Discordant for Primary Hyperparathyroidism	Horm Res Paediatr		1-8	2019
A. M. Oza, M. T. DiStefano, S. E. Hemphill, B. J. Cushman, A. R. Grant, R. K. Siegert, J. Shen, A. Chapin, N. J. Boczek, L. A. Schimmenti, J. B. Murry, L. Hasadsri, K. Nara, M. Kenna, K. T. Booth, H. Azaiez, A. Griffith, K. B. Avraham, H. Kremer, H. L. Rehm, S. S. Amr, A. N. Abou Tayoun and G. ClinGen Hearing Loss Clinical Domain Working	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss	Hum Mutat	39（11）	1593-1613	2018
M. Hosoya, S. B. Minami, C. Enomoto, T. Matsunaga and K. Kaga	Elongated EABR wave latencies observed in patients with auditory neuropathy caused by OTOF mutation	Laryngoscope Investig Otolaryngol	3（5）	388-393	2018
松永達雄, 加我君孝, 務台英樹, 奈良清光, 南修司郎, 山本修子, 藤岡正人 and 小川郁	臨床像起点の遺伝性難聴診療の確立	Otology Japan	28（2）	65-69	2018
増田圭奈子, 五島史行 and 松永達雄	小児めまいの問診票(日本語版 DHI-PC)の有用性の検討	Otology Japan	28（5）	708-714	2018
M. Hosoya, T. Saeki, C. Saegusa, T. Matsunaga, H. Okano, M. Fujioka and K. Ogawa	Estimating the concentration of therapeutic range using disease-specific iPS cells: Low-dose rapamycin therapy for Pendred syndrome	Regen Ther	10	54-63	2019
H. Mutai, F. Miya, H. Shibata, Y. Yasutomi, T. Tsunoda and T. Matsunaga	Gene expression dataset for whole cochlea of Macaca fascicularis	Sci Rep	8（1）	15554	2018

H. Suzuki, K. Kurosawa, K. Fukuda, K. Ijima, R. Sumazaki, S. Saito, R. Kosaki, A. Hirasawa, Y. Okazaki, K. Imai, T. Matsunaga, T. Iwata and K.Kosaki	Japanese pathogenic variant database: DPV	Translational Science of Rare Diseases	3（3-4）	133-137	2018
松永達雄	【聴覚障害の早期発見と支援体制】 先天性難聴児のゲノム診療の意義と動向	公衆衛生	82（6）	468-473	2018
松永達雄	【知っておきたい遺伝学的検査と遺伝外来 ABC】遺伝学的診療の進め方	耳鼻咽喉科・頭頸部外科	90（8）	598-604	2018
松永達雄	ゲノム医療(遺伝子医療)の今　希少疾患・難病の遺伝カウンセリング	保健の科学	60(10)	677-681	2018
R. Kosaki, H. Ono, H. Terashima and K.Kosaki	Timothy syndrome-like condition with syndactyly but without prolongation of the QT interval	Am J Med Genet A	176（7）	1657-1661	2018
T. Uehara, T. Takenouchi, R. Kosaki, K. Kurosawa, S. Mizuno and K.Kosaki	Redefining the phenotypic spectrum of de novo heterozygous CDK13 variants: Three patients without cardiac defects	Eur J Med Genet	61（5）	243-247	2018
C. Kusano, N. Hori, K. Izawa, R. Kosaki, G. Nishimura and T. Hasegawa	Trismus-pseudocamptodactyly syndrome with bilateral hypoplastic mandibular condyles and shallow mandibular fossa: A case report	Oral Science International	15（2）	90-92	2018
占部良介, 早川格, 上田菜穂子, 武井剛, 鈴木智, 神岡哲治, 寺嶋宙, 久保田雅也, 小崎里華 and 加藤光広	新生児期発症の難治性てんかん性脳症をきたした SCN2A 変異の男児例	てんかん研究	36（2）	501	2018
三浦真理子, 村本美香, 益田博司, 小崎里華, 伊藤裕司, 鏡雅代 and 和田友香	Prader-Willi 症候群との鑑別が困難であった Temple 症候群の新生児例	小児科臨床	71（6）	1121-1126	2018

早川格, 寺嶋宙, 小崎里華 and 久保田雅也	大頭と発達遅滞を呈しPTEN 遺伝子変異を認めた 2 例	脳と発達	50（5）	372	2018
A. Tamura, S. Uemura, K. Matsubara, E. Kozuki, T. Tanaka, N. Nino, T. Yokoi, A. Saito, T. Ishida, D. Hasegawa, I. Umeki, T. Niihori, Y. Nakazawa, K. Koike, Y. Aoki and Y. Kosaka	Co-occurrence of hypertrophic cardiomyopathy and juvenile myelomonocytic leukemia in a neonate with Noonan syndrome, leading to premature death	Clin Case Rep	6（7）	1202-1207	2018
S. Takahara, S. I. Inoue, S. Miyagawa-Tomita, K. Matsuura, Y. Nakashima, T. Niihori, Y. Matsubara, Y. Saiki and Y. Aoki	New Noonan syndrome model mice with RIT1 mutation exhibit cardiac hypertrophy and susceptibility to beta-adrenergic stimulation-induced cardiac fibrosis	EBioMedicine	42	43-53	2019
S. Suzuki-Muromoto, T. Miyabayashi, K. Nagai, S. Yamamura-Suzuki, M. Anzai, Y. Takezawa, R. Sato, Y. Okubo, W. Endo, T. Inui, N. Togashi, A. Kikuchi, T. Niihori, Y. Aoki, S. Kure and K. Haginoya	Leucine-485 deletion variant of BRAF may exhibit the severe end of the clinical spectrum of CFC syndrome	J Hum Genet	64（5）	499-504	2019
N. Ahiko, Y. Baba, M. Tsuji, R. Horikawa and K. Moriyama	Investigation of maxillofacial morphology and oral characteristics with Turner syndrome and early mixed dentition	Congenit Anom (Kyoto)	59（1）	43786	2019
K. Yamaji, J. Morita, T. Watanabe, K. Gunjigake, M. Nakatomi, M. Shiga, K. Ono, K. Moriyama and T. Kawamoto	Maldevelopment of the submandibular gland in a mouse model of apert syndrome	Dev Dyn	247（11）	1175-1185	2018
N. Higashihori, J. I. Takada, M. Katayanagi, Y. Takahashi and K. Moriyama	Frequency of missing teeth and reduction of mesiodistal tooth width in Japanese patients with tooth agenesis	Prog Orthod	19（1）	30	2018
Y. Takahashi, N. Higashihori, Y. Yasuda,	Examination of craniofacial morphology in Japanese patients with congenitally	Prog Orthod	19（1）	38	2018

J. I. Takada and K. Moriyama	missing teeth: a cross-sectional study				
森山啓司	顎顔面先天異常に対する歯科矯正学的アプローチ頭蓋縫合早期癒合症の臨床・研究を中心に	中・四国矯正歯科学会雑誌	30（1）	43471	2018
T. Yokoi, T. Saito, J. I. Nagai and K. Kurosawa	17q21.32-q22 Deletion in a girl with osteogenesis imperfecta, tricho-dento-osseous syndrome, and intellectual disability	Congenit Anom (Kyoto)	59（2）	51-52	2019
S. Hayashi, T. Yokoi, C. Hatano, Y. Enomoto, Y. Tsurusaki, T. Naruto, M. Kobayashi, H. Ida and K. Kurosawa.	Biallelic mutations of EGFR in a compound heterozygous state cause ectodermal dysplasia with severe skin defects and gastrointestinal dysfunction	Hum Genome Var	5	11	2018
T. Yokoi, Y. Enomoto, Y. Tsurusaki, T. Naruto and K. Kurosawa	Nonsyndromic intellectual disability with novel heterozygous SCN2A mutation and epilepsy	Hum Genome Var	5	20	2018
黒澤健司	指定難病最前線　ヤング・シンプソン症候群	新薬と臨牀	67（11）	1371-1374	2018
L. Guo, D. R. Bertola, A. Takanohashi, A. Saito, Y. Segawa, T. Yokota, S. Ishibashi, Y. Nishida, G. L. Yamamoto, J. Franco, R. S. Honjo, C. A. Kim, C. M. Musso, M. Timmons, A. Pizzino, R. J. Taft, B. Lajoie, M. A. Knight, K. H. Fischbeck, A. B. Singleton, C. R. Ferreira, Z. Wang, L. Yan, J. Y. Garbern, P. O. Simsek-Kiper, H. Ohashi, P. G. Robey, A. Boyde, N. Matsumoto, N. Miyake, J. Spranger, R. Schiffmann, A. Vanderver, G. Nishimura, M. Passos-Bueno, C. Simons, K. Ishikawa and S. Ikegawa	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation	Am J Hum Genet	104（5）	925-935	2019

T. Motojima, K. Fujii, H. Ohashi and H. Arakawa	Catathrenia in Pitt-Hopkins syndrome associated with 18q interstitial deletion	Pediatr Int	60（5）	479-481	2018
R. Matsuura, S. I. Hamano, T. Iwamoto, K. Shimizu and H. Ohashi	First Patient With Salla Disease Confirmed by Genomic Analysis in Japan	Pediatr Neurol	81（52-53	2018	
E. Nishi, M. Takasugi, R. Kawamura, S. Shibuya, S. Takamizawa, T. Hiroma, T. Nakamura and T.Kosho.	Clinical courses of children with trisomy 13 receiving intensive neonatal and pediatric treatment	Am J Med Genet A	176（9）	1941-1949	2018
S. Shibuya, Y. Miyake, S. Takamizawa, E. Nishi, K. Yoshizawa, T. Hatata, K. Yoshizawa, K. Fujita, M. Noguchi, J. Ohata, T. Hiroma, T. Nakamura and T.Kosho.	Safety and efficacy of noncardiac surgical procedures in the management of patients with trisomy 13: A single institution-based detailed clinical observation	Am J Med Genet A	176（5）	1137-1144	2018
M. Uehara, T.Kosho, N. Yamamoto, H. E. Takahashi, T. Shimakura, J. Nakayama, H. Kato and J. Takahashi.	Spinal manifestations in 12 patients with musculocontractural Ehlers-Danlos syndrome caused by CHST14/D4ST1 deficiency (mcEDS-CHST14)	Am J Med Genet A	176（11）	2331-2341	2018
T. Yamaguchi, K. Takano, Y. Inaba, M. Morikawa, M. Motobayashi, R. Kawamura, K. Wakui, E. Nishi, S. I. Hirabayashi, Y. Fukushima, H. Kato, J. Takahashi and T.Kosho.	PIEZO2 deficiency is a recognizable arthrogryposis syndrome: A new case and literature review	Am J Med Genet A	179（6）	948-957	2019
T. Hirose, N. Takahashi, P. Tangkawattana, J. Minaguchi, S. Mizumoto, S. Yamada, N. Miyake, S. Hayashi, A. Hatamochi, J. Nakayama, T. Yamaguchi, A. Hashimoto, Y. Nomura, K. Takehana, T.Kosho and T. Watanabe.	Structural alteration of glycosaminoglycan side chains and spatial disorganization of collagen networks in the skin of patients with mcEDS-CHST14	Biochim Biophys Acta Gen Subj	1863（3）	623-631	2019

P. J. Van Der Sluijs, S. Jansen, S. A. Vergano, M. Adachi-Fukuda, Y. Alanay, A. Alkindy, A. Baban, A. Bayat, S. Beck-Wodl, K. Berry, E. K. Bijlsma, L. A. Bok, A. F. J. Brouwer, I. Van Der Burgt, P. M. Campeau, N. Canham, K. Chrzanowska, Y. W. Y. Chu, B. H. Y. Chung, K. Dahan, M. De Rademaeker, A. Destree, T. Dudding-Byth, R. Earl, N. Elcioglu, E. R. Elias, C. Fagerberg, A. Gardham, B. Gener, E. H. Gerkes, U. Grasshoff, A. Van Haeringen, K. R. Heitink, J. C. Herkert, N. S. Den Hollander, D. Horn, D. Hunt, S. G. Kant, M. Kato, H. Kayserili, R. Kersseboom, E. Kilic, M. Krajewska-Walasek, K. Lammers, L. W. Laulund, D. Lederer, M. Lees, V. Lopez-Gonzalez, S. Maas, G. M. S. Mancini, C. Marcelis, F. Martinez, I. Maystadt, M. Mcguire, S. Mckee, S. Mehta, K. Metcalfe, J. Milunsky, S. Mizuno, J. B. Moeschler, C. Netzer, C. W. Ockeloen, B. Oehl-Jaschkowitz, N. Okamoto, S. N. M. Olminkhof, C. Orellana, L. Pasquier, C. Pottinger, V. Riehmer, S. P. Robertson, M. Roifman, C. Rooryck, F. G. Ropers, M. Rosello, C. a. L. Ruivenkamp, M. S. Sagiroglu, S. Sallevelt, A. S. Calvo, P. O. Simsek-Kiper, G. Soares, L. Solaeche, F. M. Sonmez, M. Splitt, D. Steenbeek, A. P. A. Stegmann, C. Stumpel, S. Tanabe, E. Uctepe, G.	Correction: The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome	Genet Med		2019	

E. Utine, H. E. Veenstra-Knol, S. Venkateswaran, C. Vilain, C. Vincent-Delorme, A. T. Vulto-Van Silfhout, P. Wheeler, G. N. Wilson, L. C. Wilson, B. Wollnik, T.Kosho, D. Wieczorek, E. Eichler, R. Pfundt, B. B. A. De Vries, J. Clayton-Smith and G. W. E. Santen.					
H. Morokawa, M. Kamiya, K. Wakui, M. Kobayashi, T. Kurata, K. Matsuda, R. Kawamura, H. Kanno, Y. Fukushima, Y. Nakazawa and T.Kosho.	Myelodysplastic syndrome in an infant with constitutional pure duplication 1q41-qter	Hum Genome Var	5（6	2018	
N. Koitabashi, T. Yamaguchi, D. Fukui, T. Nakano, A. Umeyama, K. Toda, R. Funada, M. Ishikawa, R. Kawamura, K. Okada, A. Hatamochi, T.Kosho and M. Kurabayashi.	Peripartum Iliac Arterial Aneurysm and Rupture in a Patient with Vascular Ehlers-Danlos Syndrome Diagnosed by Next-Generation Sequencing	Int Heart J	59（5）	1180-1185	2018
Y. Ogawa, K. Nakamura, N. Ezawa, T. Yamaguchi, T. Yoshinaga, D. Miyazaki, T.Kosho and Y. Sekijima.	A novel CACNA1A nonsense variant in a patient presenting with paroxysmal exertion-induced dyskinesia	J Neurol Sci	399（214-216	2019	
D. Kumaki, Y. Nakamura, N. Sakai, T.Kosho, A. Nakamura, S. Hirabayashi, T. Suzuki, M. Kamimura and H. Kato.	Efficacy of Denosumab for Glucocorticoid-Induced Osteoporosis in an Adolescent Patient with Duchenne Muscular Dystrophy: A Case Report	JBJS Case Connect	8（2）	e22	2018
T. Kawakami, H. Nakazawa, F. Kawakami, S. Matsuzawa, Y. Sudo, H. Sakai, S. Nishina, N. Senoo, Y. Senoo, M. Komatsu, T. Umemura, T. Yamaguchi, T.Kosho, T. Fujiwara, H. Harigae and F. Ishida.	[Successful treatment of X-linked sideroblastic anemia with ALAS2 R452H mutation using vitamin B6]	Rinsho Ketsueki	59（4）	401-406	2018

家里明日美, 黄瀬恵美子, 石川真澄, 山本佳那, 大場崇旦, 伊藤勅子, 金井敏晴, 前野一真, 山下浩美, 高野亨子, 鹿島大靖, 菊地範彦, 宮本強, 塩沢丹里, 福嶋義光, 伊藤研一 and 古庄知己	遺伝性乳がん卵巣がん症候群における遺伝カウンセリング受診者の臨床的特徴　信州大学医学部附属病院における 20 年間の取り組みから	日本遺伝カウンセリング学会誌	39（1）	53-59	2018
M. Igarashi, K. Mizuno, M. Kon, S. Narumi, Y. Kojima, Y. Hayashi, T. Ogata and M. Fukami	GATA4 mutations are uncommon in patients with 46,XY disorders of sex development without heart anomaly	Asian J Androl	20（6）	629-631	2018
H. Mano, S. Fujiwara, K. Takamura, H. Kitoh, S. Takayama, T. Ogata, S. Hashimoto and N. Haga	Congenital limb deficiency in Japan: a cross-sectional nationwide survey on its epidemiology	BMC Musculoskelet Disord	19（1）	262	2018
T. Hiraide, T. Ogata, S. Watanabe, M. Nakashima, T. Fukuda and H. Saitsu	Coexistence of a CAV3 mutation and a DMD deletion in a family with complex muscular diseases	Brain Dev	41（5）	474-479	2019
D. Shimizu, S. Iwashima, K. Sato, S. Hayano, M. Fukami, H. Saitsu and T. Ogata	GATA4 variant identified by whole-exome sequencing in a Japanese family with atrial septal defect: Implications for male sex development	Clin Case Rep	6（11）	2229-2233	2018
H. Hattori, H. Hiura, A. Kitamura, N. Miyauchi, N. Kobayashi, S. Takahashi, H. Okae, K. Kyono, M. Kagami, T. Ogata and T. Arima	Association of four imprinting disorders and ART	Clin Epigenetics	11（1）	21	2019
M. Kagami, A. Yanagisawa, M. Ota, K. Matsuoka, A. Nakamura, K. Matsubara, K. Nakabayashi, S. Takada, M. Fukami and T. Ogata	Temple syndrome in a patient with variably methylated CpGs at the primary MEG3/DLK1:IG-DMR and severely hypomethylated CpGs at the secondary MEG3:TSS-DMR	Clin Epigenetics	11（1）	42	2019
K. Matsubara, M. Itoh, K. Shimizu, S. Saito, K. Enomoto, K. Nakabayashi, K. Hata, K.	Exploring the unique function of imprinting control centers in the PWS/AS-responsible	Clin Epigenetics	11（1）	36	2019

Kurosawa, T. Ogata, M. Fukami and M. Kagami	region: finding from array-based methylation analysis in cases with variously sized microdeletions				
T. Yoshida, T. Matsuzaki, M. Miyado, K. Saito, T. Iwasa, Y. Matsubara, T. Ogata, M. Irahara and M. Fukami	11-oxygenated C19 steroids as circulating androgens in women with polycystic ovary syndrome	Endocr J	65（10）	979-990	2018
J. R. Hernandez Mora, C. Tayama, M. Sanchez-Delgado, A. Monteagudo-Sanchez, K. Hata, T. Ogata, J. Medrano, M. E. Poo-Llanillo, C. Simon, S. Moran, M. Esteller, J. Tenorio, P. Lapunzina, M. Kagami, D. Monk and K. Nakabayashi	Characterization of parent-of-origin methylation using the Illumina Infinium MethylationEPIC array platform	Epigenomics	10（7）	941-954	2018
E. Suzuki, H. Shima, M. Kagami, S. Soneda, T. Tanaka, S. Yatsuga, J. Nishioka, Y. Oto, T. Kamiya, Y. Naiki, T. Ogata, Y. Fujisawa, A. Nakamura, S. Kawashima, S. Morikawa, R. Horikawa, S. Sano and M. Fukami	(Epi)genetic defects of MKRN3 are rare in Asian patients with central precocious puberty	Hum Genome Var	6	7	2019
M. Miyado, M. Fukami, S. Takada, M. Terao, K. Nakabayashi, K. Hata, Y. Matsubara, Y. Tanaka, G. Sasaki, K. Nagasaki, M. Shiina, K. Ogata, Y. Masunaga, H. Saitsu and T. Ogata	Germline-Derived Gain-of-Function Variants of Gsalpha-Coding GNAS Gene Identified in Nephrogenic Syndrome of Inappropriate Antidiuresis	J Am Soc Nephrol	30（5）	877-889	2019
M. Nakashima, J. Tohyama, E. Nakagawa, Y. Watanabe, C. G. Siew, C. S. Kwong, K. Yamoto, T. Hiraide, T. Fukuda, T. Kaname, K. Nakabayashi, K. Hata, T. Ogata, H. Saitsu and N. Matsumoto	Identification of de novo CSNK2A1 and CSNK2B variants in cases of global developmental delay with seizures	J Hum Genet	64（4）	313-322	2019

T. Inoue, H. Yagasaki, J. Nishioka, A. Nakamura, K. Matsubara, S. Narumi, K. Nakabayashi, K. Yamazawa, T. Fuke, A. Oka, T. Ogata, M. Fukami and M. Kagami	Molecular and clinical analyses of two patients with UPD(16)mat detected by screening 94 patients with Silver-Russell syndrome phenotype of unknown aetiology	J Med Genet	Sep 21		2018
A. Nakamura, K. Muroya, H. Ogata-Kawata, K. Nakabayashi, K. Matsubara, T. Ogata, K. Kurosawa, M. Fukami and M. Kagami	A case of paternal uniparental isodisomy for chromosome 7 associated with overgrowth	J Med Genet	55（8）	567-570	2018
R. Matsushita, K. Nagasaki, T. Ayabe, Y. Miyoshi, S. Kinjo, H. Haruna, K. Ihara, T. Hasegawa, S. Ida, K. Ozono, K. Minamitani and E. Thyroid Committee of the Japanese Society for Pediatric.	Present status of prophylactic thyroidectomy in pediatric multiple endocrine neoplasia 2: a nationwide survey in Japan 1997-2017.	J Pediatr Endocrinol Metab	32(6)	585-595	2019
K. Kurata, K. Hosono, A. Hikoya, A. Kato, H. Saitsu, S. Minoshima, T. Ogata and Y. Hotta	Clinical characteristics of a Japanese patient with Bardet-Biedl syndrome caused by BBS10 mutations	Jpn J Ophthalmol	62（4）	458-466	2018
A. Takata, N. Miyake, Y. Tsurusaki, R. Fukai, S. Miyatake, E. Koshimizu, I. Kushima, T. Okada, M. Morikawa, Y. Uno, K. Ishizuka, K. Nakamura, M. Tsujii, T. Yoshikawa, T. Toyota, N. Okamoto, Y. Hiraki, R. Hashimoto, Y. Yasuda, S. Saitoh, K. Ohashi, Y. Sakai, S. Ohga, T. Hara, M. Kato, K. Nakamura, A. Ito, C. Seiwa, E. Shirahata, H. Osaka, A. Matsumoto, S. Takeshita, J. Tohyama, T. Saikusa, T. Matsuishi, T. Nakamura, T. Tsuboi, T. Kato, T. Suzuki, H. Saitsu, M. Nakashima, T. Mizuguchi, F. Tanaka, N.	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder	Cell Rep	22（3）	734-747	2018

Mori, N. Ozaki and N. Matsumoto					
M. Nakashima, Y. Negishi, I. Hori, A. Hattori, S. Saitoh and H. Saitsu	A case of early-onset epileptic encephalopathy with a homozygous TBC1D24 variant caused by uniparental isodisomy	Am J Med Genet A	179（4）	645-649	2019
I. Hori, F. Miya, Y. Negishi, A. Hattori, N. Ando, K. A. Boroevich, N. Okamoto, M. Kato, T. Tsunoda, M. Yamasaki, Y. Kanemura, K.Kosaki and S. Saitoh	A novel homozygous missense mutation in the SH3-binding motif of STAMBP causing microcephaly-capillary malformation syndrome	J Hum Genet	63（9）	957-963	2018
M. Yamada, T. Uehara, H. Suzuki, T. Takenouchi, H. Yoshihashi, H. Suzumura, S. Mizuno and K.Kosaki	SATB2-associated syndrome in patients from Japan: Linguistic profiles	Am J Med Genet A	179（6）	896-899	2019
K. Kato, S. Mizuno, M. Inaba, S. Fukumura, N. Kurahashi, K. Maruyama, D. Ieda, K. Ohashi, I. Hori, Y. Negishi, A. Hattori and S. Saitoh	Distinctive facies, macrocephaly, and developmental delay are signs of a PTEN mutation in childhood	Brain Dev	40（8）	678-684	2018
D. Fukushi, K. Yamada, K. Suzuki, M. Inaba, N. Nomura, Y. Suzuki, K. Katoh, S. Mizuno and N. Wakamatsu	Clinical and genetic characterization of a patient with SOX5 haploinsufficiency caused by a de novo balanced reciprocal translocation	Gene	655	65-70	2018
H. Imura, S. Suzuki, S. Mizuno, C. Sakuma and N. Natsume	A case of Tetrasomy 15q with left cleft lip and alveolus	Journal of Oral and Maxillofacial Surgery Medicine and Pathology	29（5）	427-429	2017
Y. Suzuki, Y. Enokido, K. Yamada, M. Inaba, K. Kuwata, N. Hanada, T. Morishita, S. Mizuno and N. Wakamatsu	The effect of rapamycin, NVP-BEZ235, aspirin, and metformin on PI3K/AKT/mTOR signaling pathway of PIK3CA-related overgrowth spectrum (PROS)	Oncotarget	8（28）	45470-45483	2017

N. Okamoto	Okamoto syndrome has features overlapping with Au-Kline syndrome and is caused by HNRNPK mutation	Am J Med Genet A	179（5）	822-826	2019
A. Fassio, A. Esposito, M. Kato, H. Saitsu, D. Mei, C. Marini, V. Conti, M. Nakashima, N. Okamoto, A. Olmez Turker, B. Albuz, C. N. Semerci Gunduz, K. Yanagihara, E. Belmonte, L. Maragliano, K. Ramsey, C. Balak, A. Siniard, V. Narayanan, C. R. R. Group, C. Ohba, M. Shiina, K. Ogata, N. Matsumoto, F. Benfenati and R. Guerrini	De novo mutations of the ATP6V1A gene cause developmental encephalopathy with epilepsy	Brain	141（6）	1703-1718	2018
T. Saikusa, M. Hara, K. Iwama, K. Yuge, C. Ohba, J. I. Okada, T. Hisano, Y. Yamashita, N. Okamoto, H. Saitsu, N. Matsumoto and T. Matsuishi	De novo HDAC8 mutation causes Rett-related disorder with distinctive facial features and multiple congenital anomalies	Brain Dev	40（5）	406-409	2018
K. Ueda, K. Yanagi, T. Kaname and N. Okamoto	A novel mutation in the GATAD2B gene associated with severe intellectual disability	Brain Dev	41（3）	276-279	2019
K. Yamamoto-Shimojima, N. Okamoto, W. Matsumura, T. Okazaki and T. Yamamoto	Three Japanese patients with 3p13 microdeletions involving FOXP1	Brain Dev	41（3）	257-262	2019
K. Yamamoto-Shimojima, M. Kouwaki, Y. Kawashima, K. Itomi, K. Momosaki, S. Ozasa, N. Okamoto, K. Yokochi and T. Yamamoto	Natural histories of patients with Wolf-Hirschhorn syndrome derived from variable chromosomal abnormalities	Congenit Anom (Kyoto)			2018
M. Taniguchi-Ikeda, N. Morisada, H. Inagaki, Y. Ouchi, Y. Takami, M. Tachikawa, W. Satake,	Two patients with PNKP mutations presenting with	Clin Genet	93（4）	931-933	2018

K. Kobayashi, S. Tsuneishi, S. Takada, H. Yamaguchi, H. Nagase, K. Nozu, N. Okamoto, H. Nishio, T. Toda, I. Morioka, H. Wada, H. Kurahashi and K. Iijima	microcephaly, seizure, and oculomotor apraxia				
T. Imaizumi, Y. Mogami, N. Okamoto, K. Yamamoto-Shimojima and T. Yamamoto	De novo 1p35.2 microdeletion including PUM1 identified in a patient with sporadic west syndrome	Congenit Anom (Kyoto)		2019	
J. Kennedy, D. Goudie, E. Blair, K. Chandler, S. Joss, V. McKay, A. Green, R. Armstrong, M. Lees, B. Kamien, B. Hopper, T. Y. Tan, P. Yap, Z. Stark, N. Okamoto, N. Miyake, N. Matsumoto, E. Macnamara, J. L. Murphy, E. McCormick, H. Hakonarson, M. J. Falk, D. Li, P. Blackburn, E. Klee, D. Babovic-Vuksanovic, S. Schelley, L. Hudgins, S. Kant, B. Isidor, B. Cogne, K. Bradbury, M. Williams, C. Patel, H. Heussler, C. Duff-Farrier, P. Lakeman, I. Scurr, U. Kini, M. Elting, M. Reijnders, J. Schuurs-Hoeijmakers, M. Wafik, A. Blomhoff, C. A. L. Ruivenkamp, E. Nibbeling, A. J. M. Dingemans, E. D. Douine, S. F. Nelson, D. D. D. Study, V. A. Arboleda and R. Newbury-Ecob	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants	Genet Med	21（4）	850-860	2019
Y. Mogami, Y. Suzuki, Y. Murakami, T. Ikeda, S. Kimura, K. Yanagihara, N. Okamoto and T. Kinoshita	Early infancy-onset stimulation-induced myoclonic seizures in three siblings with inherited glycosylphosphatidylinositol (GPI) anchor deficiency	Epileptic Disord	20（1）	42-50	2018

N. Okamoto, T. Kohmoto, T. Naruto, K. Masuda and I. Imoto	Primary microcephaly caused by novel compound heterozygous mutations in ASPM	Hum Genome Var	5	18015	2018
K. Shimojima, N. Okamoto, K. Ohmura, H. Nagase and T. Yamamoto	Infantile spasms related to a 5q31.2-q31.3 microdeletion including PURA	Hum Genome Var	5	18007	2018
M. P. Adam, S. Banka, H. T. Bjornsson, O. Bodamer, A. E. Chudley, J. Harris, H. Kawame, B. C. Lanpher, A. W. Lindsley, G. Merla, N. Miyake, N. Okamoto, C. T. Stumpel, N. Niikawa and B. Kabuki Syndrome Medical Advisory	Kabuki syndrome: international consensus diagnostic criteria	J Med Genet	56（2）	89-95	2019
A. Inoko, T. Yano, T. Miyamoto, S. Matsuura, T. Kiyono, N. Goshima, M. Inagaki and Y. Hayashi	Albatross/FBF1 contributes to both centriole duplication and centrosome separation	Genes Cells	23（12）	1023-1042	2018
T. Miyamoto, S. N. Akutsu, H. Tauchi, Y. Kudo, S. Tashiro, T. Yamamoto and S. Matsuura	Exploration of genetic basis underlying individual differences in radiosensitivity within human populations using genome editing technology	J Radiat Res	59（suppl_2）	ii75-ii82	2018
H. Hidaka, K. Higashimoto, S. Aoki, H. Mishima, C. Hayashida, T. Maeda, Y. Koga, H. Yatsuki, K. Joh, H. Noshiro, R. Iwakiri, A. Kawaguchi, K. I. Yoshiura, K. Fujimoto and H. Soejima	Comprehensive methylation analysis of imprinting-associated differentially methylated regions in colorectal cancer	Clin Epigenetics	10（1）	150	2018
K. Joh, F. Matsuhisa, S. Kitajima, K. Nishioka, K. Higashimoto, H. Yatsuki, T. Kono, H. Koseki and H. Soejima	Growing oocyte-specific transcription-dependent de novo DNA methylation at the imprinted Zrsr1-DMR	Epigenetics Chromatin	11（1）	28	2018

T. Yamada, G. Sugiyama, K. Higashimoto, A. Nakashima, H. Nakano, T. Sumida, H. Soejima and Y. Mori	Beckwith-Wiedemann syndrome with asymmetric mosaic of paternal disomy causing hemihyperplasia	Oral Surg Oral Med Oral Pathol Oral Radiol	127（3）	e84-e88	2019
K. Nishioka, H. Miyazaki and H. Soejima	Unbiased shRNA screening, using a combination of FACS and high-throughput sequencing, enables identification of novel modifiers of Polycomb silencing	Sci Rep	8（1）	12128	2018
副島英伸	【婦人科医が注意すべき悪性腫瘍関連疾患の新知識-他科エキスパートに聞く】 Beckwith-Wiedemann 症候群	産科と婦人科	85（6）	667-672	2018
T. Kikuiri, H. Mishima, H. Imura, S. Suzuki, Y. Matsuzawa, T. Nakamura, S. Fukumoto, Y. Yoshimura, S. Watanabe, A. Kinoshita, T. Yamada, M. Shindoh, Y. Sugita, H. Maeda, Y. Yawaka, T. Mikoya, N. Natsume and K. I. Yoshiura	Patients with SATB2-associated syndrome exhibiting multiple odontomas	Am J Med Genet A	176（12）	2614-2622	2018
T. Shirakawa, Y. Nakashima, S. Watanabe, S. Harada, M. Kinoshita, T. Kihara, Y. Hamasaki, S. Shishido, K. I. Yoshiura, H. Moriuchi and S. Dateki	A novel heterozygous GLI2 mutation in a patient with congenital urethral stricture and renal hypoplasia/dysplasia leading to end-stage renal failure	CEN Case Rep	7（1）	94-97	2018
K. Kiyota, K. I. Yoshiura, R. Houbara, H. Miyahara, S. Korematsu and K. Ihara	Auto-immune disorders in a child with PIK3CD variant and 22q13 deletion	Eur J Med Genet	61（10）	631-633	2018
Y. S. Hori, A. Yamada, N. Matsuda, Y. Ono, D. Starenki, N. Sosonkina, K. I. Yoshiura, N. Niikawa and T. Ohta	A Novel Association between the 27-bp Deletion and 538G>A Mutation in the ABCC11 Gene	Hum Biol	89（4）	305-307	2017

S. Sato, H. Itonaga, M. Taguchi, Y. Sawayama, D. Imanishi, H. Tsushima, T. Hata, Y. Moriuchi, H. Mishima, A. Kinoshita, K. I. Yoshiura and Y. Miyazaki	Clonal dynamics in a case of acute monoblastic leukemia that later developed myeloproliferative neoplasm	Int J Hematol	108（2）	213-217	2018
Y. Murakami, K. Miura, S. Sato, A. Higashijima, Y. Hasegawa, S. Miura, K. I. Yoshiura and H. Masuzaki	Reference values for circulating pregnancy-associated microRNAs in maternal plasma and their clinical usefulness in uncomplicated pregnancy and hypertensive disorder of pregnancy	J Obstet Gynaecol Res	44（5）	840-851	2018
T. Koga, K. Migita, T. Sato, S. Sato, M. Umeda, F. Nonaka, S. Fukui, S. Y. Kawashiri, N. Iwamoto, K. Ichinose, M. Tamai, H. Nakamura, T. Origuchi, Y. Ueki, J. Masumoto, K. Agematsu, A. Yachie, K. I. Yoshiura, K. Eguchi and A. Kawakami	MicroRNA-204-3p inhibits lipopolysaccharide-induced cytokines in familial Mediterranean fever via the phosphoinositide 3-kinase gamma pathway	Rheumatology (Oxford)	57（4）	718-726	2018
吉浦孝一郎	全ゲノム関連解析研究(GWAS)の原理と考え方	遺伝子医学	9（1）	114-121	2019
Y. Ishijima, T. Iizuka, K. Kagami, S. Masumoto, K. Nakade, Y. Mitani, Y. Niida, A. Watanabe, R. Yamazaki, M. Ono and H. Fujiwara.	Prenatal diagnosis facilitated prompt enzyme replacement therapy for prenatal benign hypophosphatasia.	J Obstet Gynaecol	40(1)	132-134	2020
T. Mushiroda, Y. Takahashi, T. Onuma, Y. Yamamoto, T. Kamei, T. Hoshida, K. Takeuchi, K. Otsuka, M. Okazaki, M. Watanabe, K. Kanemoto, T. Oshima, A. Watanabe, S. Minami, K. Saito, H. Tanii, Y. Shimo, M. Hara, S. Saitoh, T. Kinoshita, M. Kato, N. Yamada, N. Akamatsu, T. Fukuchi, S. Ishida, S.	Association of HLA-A*31:01 Screening With the Incidence of Carbamazepine-Induced Cutaneous Adverse Reactions in a Japanese Population	JAMA Neurol	75（7）	842-849	2018

Yasumoto, A. Takahashi, T. Ozeki, T. Furuta, Y. Saito, N. Izumida, Y. Kano, T. Shiohara, M. Kubo and G. S. Group					
R. Ikeue, A. Nakamura-Takahashi, Y. Nitahara-Kasahara, A. Watanabe, T. Muramatsu, T. Sato and T. Okada	Bone-Targeted Alkaline Phosphatase Treatment of Mandibular Bone and Teeth in Lethal Hypophosphatasia via an scAAV8 Vector	Mol Ther Methods Clin Dev	10	360-370	2018
渡邉淳	【最新遺伝医学研究と遺伝カウンセリング(シリーズ 3)　最新 多因子遺伝性疾患研究と遺伝カウンセリング】 (第 5 章)多因子疾患の遺伝情報と社会 社会における遺伝リテラシー向上	遺伝子医学 MOOK	別冊（最新多因子遺伝性疾患研究と遺伝カウンセリング）	283-288	2018
T. Sato, M. Kato, K. Moriyama, K. Haraguchi, H. Saitsu, N. Matsumoto and H. Moriuchi	A case of tubulinopathy presenting with porencephaly caused by a novel missense mutation in the TUBA1A gene	Brain Dev	40（9）	819-823	2018
加藤光広	新生児科医が知っておきたい脳の発生と脳形成異常	日本周産期・新生児医学会雑誌	53（2）	411	2017
M. Kamada, M. Nakatsui, R. Kojima, S. Nohara, E. Uchino, S. Tanishima, M. Sugiyama, K. Kosaki, K. Tokunaga, M. Mizokami and Y. Okuno.	MGeND: an integrated database for Japanese clinical and genomic information.	Hum Genome Var.	6	53	2019
H. Suzuki, T. Yoshida, N. Morisada, T. Uehara, K. Kosaki, K. Sato, K. Matsubara, T. Takano-Shimizu and T. Takenouchi.	De novo NSF mutations cause early infantile epileptic encephalopathy.	Ann Clin Transl Neurol.	6(11)	2334-2339.	2019
T. Takeda, K. Banno, Y. Kobayashi, M. Adachi, M. Yanokura, E. Tominaga, K. Kosaki and D. Aoki.	Mutations of RAS genes in endometrial polyps.	Oncol Rep.	42(6)	2303-2308.	2019

T. Takenouchi, H. Okuno and K. Kosaki.	Kosaki overgrowth syndrome: A newly identified entity caused by pathogenic variants in platelet-derived growth factor receptor-beta.	Am J Med Genet C Semin Med Genet.	181(4)	650-657.	2019
T. Uehara, T. Tsuchihashi, M. Yamada, H. Suzuki, T. Takenouchi and K. Kosaki.	CNOT2 haploinsufficiency causes a neurodevelopmental disorder with characteristic facial features.	Am J Med Genet A.	179(12)	2506-2509.	2019
M. Yamada, H. Suzuki, Y. Shiraishi and K. Kosaki.	Effectiveness of integrated interpretation of exome and corresponding transcriptome data for detecting splicing variants of genes associated with autosomal recessive disorders.	Mol Genet Metab Rep.	21	100531	2019
M. Yamada, T. Uehara, H. Suzuki, T. Takenouchi, H. Fukushima, N. Morisada, K. Tominaga, M. Onoda and K. Kosaki.	IFT172 as the 19th gene causative of oral-facial-digital syndrome.	Am J Med Genet A.	179(12)	2510-2513.	2019
S. Umetsu, A. Inui, S. Kobayashi, M. Shimura, T. Uehara, H. Uchida, R. Irie, T. Sogo, H. Komatsu, T. Yoshioka, K. Murayama, K. Kosaki, M. Kasahara and T. Fujisawa.	First cases of MPV17 related mitochondrial DNA depletion syndrome with compound heterozygous mutations in p.R50Q/p.R50W: a case report.	Hepatoma Research.			2020
G. S. Baynam, S. Groft, F. H. Van Der Westhuizen, S. D. Gassman, K. Du Plessis, E. P. Coles, E. Selebatso, M. Selebatso, B. Gaobinelwe, T. Selebatso, D. Joel, V. A. Llera, B. C. Vorster, B. Wuebbels, B. Djoudalbaye, C. P. Austin, J. Kumuthini, J. Forman, P. Kaufmann, J. Chipeta, D. Gavhed, A. Larsson, M. Stojiljkovic, A. Nordgren, E. J. A.	A call for global action for rare diseases in Africa.	Nat Genet.	52(1)	21-26.	2020

Roldan, D. Taruscio, D. Wong-Rieger, K. Nowak, G. A. Bilkey, S. Easteal, S. Bowdin, J. K. V. Reichardt, S. Beltran, K. Kosaki, C. D. M. Van Karnebeek, M. Gong, Z. Shuyang, R. Mehrian-Shai, D. R. Adams, R. D. Puri, F. Zhang, N. Pachter, M. Muenke, C. Nellaker, W. A. Gahl, H. Cederroth, S. Broley, M. Schoonen, K. M. Boycott and M. Posada.					
T. Fujisawa, Y. Aizawa, Y. Katsumata, K. Kimura, K. Hashimoto, T. Yamashita, H. Miyama, T. Kimura, K. Kosaki, S. Takatsuki, W. Shimizu and K. Fukuda.	Mexiletine shortens the QT interval in a pedigree of KCNH2 related long QT syndrome.	J Arrhythm.	36(1)	193-196.	2020
T. Fukaishi, I. Minami, S. Masuda, Y. Miyachi, K. Tsujimoto, H. Izumiyama, K. Hashimoto, M. Yoshida, S. Takahashi, K. Kashimada, T. Morio, K. Kosaki, Y. Maezawa, K. Yokote, T. Yoshimoto and T. Yamada.	A case of generalized lipodystrophy-associated progeroid syndrome treated by leptin replacement with short and long-term monitoring of the metabolic and endocrine profiles.	Endocr J.	67(2)	211-218.	2020
T. Hiraide, M. Kataoka, H. Suzuki, Y. Aimi, T. Chiba, S. Isobe, Y. Katsumata, S. Goto, K. Kanekura, Y. Yamada, H. Moriyama, H. Kitakata, J. Endo, S. Yuasa, Y. Arai, N. Hirose, T. Satoh, Y. Hakamata, M. Sano, S. Gamou, K. Kosaki and K. Fukuda.	Poor outcomes in carriers of the RNF213 variant (p.Arg4810Lys) with pulmonary arterial hypertension.	J Heart Lung Transplant.	39(2)	103-112.	2020
L. Li, C. Y. Fong, C. G. Tay, S. K. Tae, H. Suzuki, K. Kosaki and M. K. Thong.	Infantile neuroaxonal dystrophy in a pair of Malaysian siblings with progressive cerebellar atrophy: Description of an expanded phenotype with novel PLA2G6 variants.	J Clin Neurosci.	71	289-292.	2020

H. Murakami, T. Uehara, Y. Tsurusaki, Y. Enomoto, Y. Kuroda, N. Aida, K. Kosaki and K. Kurosawa.	Blended phenotype of AP4E1 deficiency and Angelman syndrome caused by paternal isodisomy of chromosome 15.	Brain Dev.	42(3)	289-292.	2020
N. Oiso, A. Kubo, A. Shimizu, H. Suzuki, K. Kosaki, T. Chikugo, K. Nakabayashi, K. Hata, S. Yanagihara, O. Ishikawa, Y. Matsubara, M. Amagai and A. Kawada.	Epidermodysplasia verruciformis without progression to squamous cell carcinomas in an elderly man: alpha-human papillomavirus infection in the evolving verruca.	Int J Dermatol.			2020
Y. Sakaguchi, T. Uehara, M. Sasaki, K. Fujimura, K. Kishi, K. Kosaki and T. Takenouchi.	Hereditary spastic paraplegia masqueraded by congenital melanocytic nevus syndrome: Dual pathogenesis of germline non-mosaicism and somatic mosaicism.	Eur J Med Genet.	63(4)	103803	2020
Y. Takeshita, T. Ohto, T. Enokizono, M. Tanaka, H. Suzuki, H. Fukushima, T. Uehara, T. Takenouchi, K. Kosaki and H. Takada.	Novel ARX mutation identified in infantile spasm syndrome patient.	Hum Genome Var.	7	9	2020
D. Taruscio, G. Baynam, H. Cederroth, S. C. Groft, E. W. Klee, K. Kosaki, P. Lasko, B. Melegh, O. Riess, M. Salvatore and W. A. Gahl.	The Undiagnosed Diseases Network International: Five years and more!	Mol Genet Metab.	129(4)	243-254.	2020
T. Uehara, M. Yamada, S. Umetsu, H. Nittono, H. Suzuki, T. Fujisawa, T. Takenouchi, A. Inui and K. Kosaki.	Biallelic Mutations in the LSR Gene Cause a Novel Type of Infantile Intrahepatic Cholestasis.	J Pediatr.	221	251-254.	2020
R. A. Vos, T. Katayama, H. Mishima, S. Kawano, S. Kawashima, J. D. Kim, Y. Moriya, T. Tokimatsu, A. Yamaguchi, Y. Yamamoto, H. Wu, P. Amstutz, E. Antezana, N. P. Aoki, K. Arakawa, J. T. Bolleman, E. Bolton,	BioHackathon 2015: Semantics of data for life sciences and reproducible research.	F1000Res.	9	136	2020

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M. Yamada, T. Uehara, H. Suzuki, T. Takenouchi, A. Inui, M. Ikemiyagi, I. Kamimaki and K. Kosaki.	Shortfall of exome analysis for diagnosis of Shwachman-Diamond syndrome: Mismapping due to the pseudogene SBDSP1.	Am J Med Genet A.			2020
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M. Miyado, M. Fukami, S. Takada, M. Terao, K. Nakabayashi, K. Hata, Y. Matsubara, Y. Tanaka, G. Sasaki, K. Nagasaki, M. Shiina, K. Ogata, Y.	Germline-Derived Gain-of-Function Variants of Gsalpha-Coding GNAS Gene Identified in Nephrogenic Syndrome of Inappropriate Antidiuresis.	J Am Soc Nephrol.	30(5)	877-889.	2019

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A. Hattori, K. Okamura, Y. Terada, R. Tanaka, Y. Katoh-Fukui, Y. Matsubara, K.	Transient multifocal genomic crisis creating chromothriptic and non-chromothriptic	BMC Med Genomics.	12(1)	77	2019

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A. Masamune, H. Kotani, F. L. Sorgel, J. M. Chen, S. Hamada, R. Sakaguchi, E. Masson, E. Nakano, Y. Kakuta, T. Niihori, R. Funayama, M. Shirota, T. Hirano, T. Kawamoto, A. Hosokoshi, K. Kume, L. Unger, M. Ewers, H. Laumen, P. Bugert, M. X. Mori, V. Tsvilovskyy, P. Weissgerber, U. Kriebs, C. Fecher-Trost, M. Freichel, K. N. Diakopoulos, A. Berninger, M. Lesina, K. Ishii, T. Itoi, T. Ikeura, K. Okazaki, T. Kaune, J. Rosendahl, M. Nagasaki, Y. Uezono, H. Algul, K. Nakayama, Y. Matsubara, Y. Aoki, C. Ferec, Y. Mori, H. Witt and T. Shimosegawa.	Variants That Affect Function of Calcium Channel TRPV6 Are Associated With Early-Onset Chronic Pancreatitis.	Gastroenterology.	158(6)	1626-1641 e8.	2020
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増井徹	ゲノム創薬におけるバイオバンクの役割、	Precision medicine	2（6）,	508-511	2019
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飯森宏仁、佐藤美保、鈴木寛子、彦谷明子、堀田喜裕、吉田朋世、仁科幸子、東範行	亜）急性後天共同性内斜視に関する全国調査―デジタルデバイスとの関連について―	眼臨紀	13 (1)	42-47,	2020

中尾志郎、仁科幸子、八木瞳、田中慎、吉田朋世、横井匡、東範行	外直筋鼻側移動術を施行した動眼神経麻痺の一例.	眼臨紀	13 (2)	105-110	2020
石井杏奈、仁科幸子、松岡真未、三井田千春、赤池祥子、新保由紀子、越後貫滋子、吉田朋世、横井匡、東範行	眼器質疾患をもつ低年齢児に対する Spot Vision Screener	日視会誌	48	73-80	2019
林思音、仁科幸子、森隆史、清水ふき、南雲幹、臼井千惠、杉山能子、八子恵子	三歳児眼科健診における屈折検査の有用性：システマテイックレビュー.	眼臨紀	12 (5)	373-377,	2019
田中慎、仁科幸子、中尾志郎、吉田朋世、横井匡、東範行	斜位近視を契機に発見された小脳腫瘍の小児例.	眼臨紀	12 (4)	323-327,	2019
重安千花，山田昌和，大家義則，川崎諭，東範行，仁科幸子，木下茂，外園千恵，大橋裕一，白石敦，坪田一男，榛村重人，村上晶，島﨑潤，宮田和典，前田直之，山上聡，臼井智彦，西田幸二	厚生労働科学研究費難治性疾患政策研究事業希少難治性角膜疾患の疫学調査研究班，角膜難病の標準的診断法および治療法の確立を目指した調査研究班：前眼部形成異常の診断基準および重症度分類.	日眼会誌	124 巻 2 号	89-95	2020
仁科 幸子	フォトスクリーナーによる弱視の早期発見	保育と保健	26 (1)	102-104	2020
仁科 幸子	デジタルデバイスと急性内斜視.	日本の眼科	91 (3)	338-339	2020
仁科 幸子	乳幼児の視覚スクリーニング	日本の眼科	90 (10)	1291-1292,	2019
仁科 幸子	乳幼児の視覚スクリーニング	東京小児科医会報	38 (1)	63-69	2019
仁科 幸子・佐藤美保	序説 弱視と斜視のカレントトピックス	あたらしい眼科	36 (8)	971-972	2019
吉田朋世・仁科 幸子	急性後天性共同性内斜視. 特集 弱視と斜視のカレントトピックス	あたらしい眼科	36 (8)	995-1001	2019

吉田朋世・仁科 幸子	デジタルデバイスと急性内斜視. 特集 デジタルデバイス時代の視機能管理.	あたらしい眼科	36 (7)	877-882	2019
仁科 幸子	レーバー先天盲. ～知っておきたい稀な網膜・硝子体ジストロフィー	オクリスタ	75 (6)	31-37	2019
仁科 幸子	手持ちフォトスクリーナー装置、乳幼児期の眼鏡・コンタクトレンズ	チャイルドヘルス	22 (6)	21-23, 47-49	2019
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鹿島大靖，岡賢二，品川真奈花，山中桜，小野元紀，竹内穂高，井田耕一，樋口正太郎，山田靖，小原久典，菊地範彦，宮本強，橘涼太，福嶋義光，古庄知己，家里明日美，伊藤研一，浅香志穂，上原剛，塩沢丹里.	腹腔鏡下リスク低減卵管卵巣摘出術により両側卵管采に漿液性卵管上皮内病変が発見された遺伝性乳癌卵巣癌症候群の 1 例.	信州医学雑誌.	67(3)	209-215.	2019
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