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
倉橋浩樹	網羅的着床前診断の基礎と実際	倉橋浩樹	網羅的着床前診断の基礎と実際	診断と治療社	東京	In press	
倉橋浩樹	小児科診療における遺伝カウンセリング		今日の小児治療指針第17版	医学書院	東京	In press	
山本俊至	症例でわかる小児神経疾患の遺伝学的アプローチ	山本俊至	症例でわかる小児神経疾患の遺伝学的アプローチ	診断と治療社	東京	In press	

雑誌
研究代表者・倉橋浩樹

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Kato T, Inagaki H, Miyai S, Suzuki F, Naru Y, Shinkai Y, Kato A, Kanyama K, Mizuno S, Muramatsu Y, Yamamoto T, Shinya M, Tazaki Y, Hiwatashi S, Ikeda T, Ozaki M, Kurahashi H.	The involvement of U-type dicentric chromosomes in the formation of terminal deletions with or without adjacent inverted duplications.	**Hum Genet**	In press.	In press.	2020
Ikeda M, Taniguchi-Ikeda M, Kato T, Shinkai Y, Hagiwara H, Sasaki N, Masaki T, Matsumura K, Sonoo M, Kurahashi H, Saito F.	Unexpected mutations by CRISPR/Cas9 CTG repeat excision in myotonic dystrophy and use of CRISPR interference as an alternative approach.	**Mol Ther**	In press.	In press.	2020
Kawamura R, Kato T, Miyai S, Suzuki F, Naru Y, Kato M, Tanaka K, Nagasaka M, Tsutsumi M, Inagaki H, Ioroi T, Yoshida M, Nao T, Conlin LK, Iijima K, Kurahashi H, Taniguchi-Ikeda M.	A case of a partheno genetic 46,XX/46,XY chimera presenting ambiguous genitalia.	**J Hum Genet**	In press.	In press.	2020
Kato T, Kawai M, Miyai S, Suzuki F, Tsutsumi M, Mizuno S, Ikeda T, Kurahashi H.	Analysis of the origin of double mosaic aneuploidy in two cases.	**Cytogenet Genome Res**	In press	In press.	2020

Kato M, Yagami A, Tsukamoto T, Shinkai Y, Kato T, Kurahashi H.	Novel mutation in the KITLG gene in familial progressive hyperpigmentation with or without hypopigmentation.	**J Dermatol**	In press.	In press.	2020
Miura H, Ohye T, Kozawa K, Hattori F, Kawamura Y, Ihira M, Kurahashi H, Yoshikawa T.	Coinfection with human herpesvirus (HHV)-6B in immunocompetent, healthy individuals with chromosomally integrated HHV-6A.	**J Pediatric Infect Dis Soc**	In press.	In press.	2020
Sato T, Sugiura-Ogasawara M, Ozawa F, Yamamoto T, Kato T, Kurahashi H, Kuroda T, Aoyama N, Kato K, Kobayashi R, Fukuda A, Utsunomiya T, Kuwahara A, Saito H, Takeshita T, Irahara M.	Preimplantation genetic testing for aneuploidy: a comparison of live birth rates in patients with recurrent pregnancy loss due to embryonic aneuploidy or recurrent implantation failure.	**Hum Reprod**	34(12)	2340-2348	2019
Terasawa S, Kato A, Nishizawa H, Kato T, Yoshizawa H, Noda Y, Miyazaki J, Ito M, Sekiya T, Fujii T, Kurahashi H.	Multiplex PCR in noninvasive prenatal diagnosis for FGFR3-related disorders.	**Congenit Anom (Kyoto)**	59(1)	4-10	2019
Noda Y, Kato T, Kato A, Nishizawa H, Miyazaki J, Ito M, Terasawa S, Sekiya T, Fujii T, Kurahashi H.	Potentially effective method for fetal gender determination by non-invasive prenatal testing for X-linked disease.	**Congenit Anom (Kyoto)**	59(3)	88-92	2019
Yokoi K, Nakajima Y, Ohye T, Inagaki H, Wada Y, Fukuda T, Sugie H, Yuasa I, Ito T, Kurahashi H.	Disruption of the responsible gene in a phosphoglucomutase 1 deficiency patient by homozygous chromosomal inversion.	**JIMD Rep**	43	85-90	2019
Tahara S, Tahara T, Horiguchi N, Kato T, Shinkai Y, Yamashita H, Yamada H, Kawamura T, Terada T, Okubo M, Nagasaka M, Nakagawa Y, Shibata T, Yamada S, Urano M, Tsukamoto T, Kurahashi H, Kuroda M, Ohmiya N.	DNA methylation accumulation in gastric mucosa adjacent to cancer after Helicobacter pylori eradication.	**Int J Cancer**	144(1)	80-88	2019
Kawai M, Tsutsumi M, Suzuki F, Sameshima K, Dowa Y, Kyoya T, Inagaki H, Kurahashi H.	Two siblings with 11qter deletion syndrome that had been rescued in their mother by uniparental disomy.	**Eur J Med Genet**	62(3)	224-228	2019

Hayano S, Okuno Y, Tsutsumi M, Inagaki H, Fukasawa Y, Kurahashi H, Kojima S, Takahashi Y, Kato T.	Frequent intragenic microdeletions of elastin in familial supravalvular aortic stenosis.	**Int J Cardiol**	274	290-295	2019
Tsuchiya H, Akiyama T, Kuhara T, Nakajima Y, Ohse M, Kurahashi H, Kato T, Maeda Y, Yoshinaga H, Kobayashi K.	A case of dihydropyrimidinase deficiency incidentally detected by urine metabolome analysis.	**Brain Dev**	41(3)	280-284	2019
Ishihara N, Inagaki H, Miyake M, Kawamura Y, Yoshikawa T, Kurahashi H.	A case of early onset life-threatening epilepsy associated with a novel ATP1A3 gene variant.	**Brain Dev**	41(3)	285-291	2019
Boda H, Miyata M, Inagaki H, Shinkai Y, Kato T, Yoshikawa T, Kurahashi H.	FOXA2 gene mutation in a patient with congenital complex pituitary hormone deficiency.	**Eur J Med Genet**	62(11)	103570	2019
Hitachi K, Nakatani M, Takasaki A, Ouchi Y, Uezumi A, Ageta H, Inagaki H, Kurahashi H, Tsuchida K.	Myogenin promoter-associated lncRNA Myoparr is essential for myogenic differentiation.	**EMBO Rep**.	20(3)	e47468	2019
Inagaki H, Ota S, Nishizawa H, Miyamura H, Nakahira K, Suzuki M, Nishiyama S, Kato T, Yanagihara I, Kurahashi H.	Obstetric complication-associated ANXA5 promoter polymorphisms may affect gene expression via DNA secondary structures.	**J Hum Genet**	64(5)	459-466	2019
Tahara T, Tahara S, Horiguchi N, Kato T, Shinkai Y, Okubo M, Terada T, Yoshida D, Funasaka K, Nagasaka M, Nakagawa Y, Kurahashi H, Shibata T, Tsukamoto T, Ohmiya N.Y, Yoshinaga H, Kobayashi K.	Prostate stem cell antigen gene polymorphism is associated with H. pylori-related promoter DNA methylation in nonneoplastic gastric epithelium.	**Cancer Prev Res (Phila)**	12(9)	579-584	2019
Hitachi K, Inagaki H, Kurahashi H, Okada H, Tsuchida K, Honda M.	Deficiency of Vgll2 gene alters the gene expression profiling of skeletal muscle subjected to mechanical overload.	**Front Sports Act Living**	1	41	2019
Yokoi K, Nakajima Y, Shinkai Y, Sano Y, Imamura M, Akiyama T, Yoshikawa T, Ito T, Kurahashi H.	Clinical and genetic aspects of mild hypophosphatasia in Japanese patients.	**Mol Genet Metab Rep**	21	100515	2019

Toshimitsu M, Nagaoka S, Kobori S, Ogawa M, Suzuki F, Kato T, Miyai S, Kawamura R, Inagaki H, Kurahashi H, Murotsuki J.	Exome-First Approach in Fetal Akinesia Reveals Chromosome 1p36 Deletion Syndrome.	**Case Rep Obstet Gynecol**.	2019	6753184	2019
Tsutsumi M, Hattori H, Akita N, Maeda N, Kubota T, Horibe K, Fujita N, Kawai M, Shinkai Y, Kato M, Kato T, Kawamura R, Suzuki F, Kurahashi H.	A female patient with retinoblastoma and severe intellectual disability carrying an X;13 balanced translocation without rearrangement in the RB1 gene: a case report.	**BMC Med Genomics**	12(1)	182	2019

研究分担者・大橋博文

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Daida A, Hamano SI, Ikemoto S, Hirata Y, Matsuura R, Koichihara R, Oba D, Ohashi H.	Use of Perampanel and a Ketogenic Diet in Nonketotic Hyperglycinemia: A Case Report.	**Neuropediatrics**	In press.	In press.	2020
Shimizu K, Oba D, Nambu R, Tanaka M, Oguma E, Murayama K, Ohtake A, Yoshiura KI, Ohashi H.	Possible mitochondrial dysfunction in a patient with deafness, dystonia, and cerebral hypomyelination (DDCH) due to BCAP31 Mutation.	**Mol Genet Genomic Med**	8(3)	e1129	2020
Sekiguchi F, Tsurusaki Y, Okamoto N, Teik KW, Mizuno S, Suzumura H, Isidor B, Ong WP, Haniffa M, White SM, Matsuo M, Saito K, Phadke S, Kosho T, Yap P, Goyal M, Clarke LA, Sachdev R, McGillivray G, Leventer RJ, Patel C, Yamagata T, Osaka H, Hisaeda Y, Ohashi H, Shimizu K, Nagasaki K, Hamada J, Dateki S, Sato T, Chinen Y, Awaya T, Kato T, Iwanaga K, Kawai M, Matsuoka T, Shimoji Y, Tan TY, Kapoor S, Gregersen N, Rossi M, Marie-Laure M, McGregor L, Oishi K, Mehta L, Gillies G, Lockhart PJ, Pope K, Shukla A, Girisha KM, Abdel-Salam GMH, Mowat D, Coman D, Kim OH, Cordier MP, Gibson K, Milunsky J, Liebelt J, Cox H, El Chehadeh S, Toutain A, Saida K, Aoi H, Minase G, Tsuchida N, Iwama K, Uchiyama Y, Suzuki T, Hamanaka K, Azuma Y, Fujita A, Imagawa E, Koshimizu E, Takata A, Mitsuhashi S, Miyatake S, Mizuguchi T, Miyake N, Matsumoto N.	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients.	**J Hum Genet**	64(12)	1173-1186	2019

Niihori T, Nagai K, Fujita A, Ohashi H, Okamoto N, Okada S, Harada A, Kihara H, Arboga st T, Funayama R, Shirota M, Nakayama K, Abe T, Inoue SI, Tsai IC, Matsumoto N, Davis EE, Katsanis N, Aoki Y.	Germline-Activating RRAS2 Mutations Cause Noonan Synd rome.	**Am J Hum G enet**	104(6)	1233-1240	2019

研究分担者・黒澤健司

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Tominaga M, Saito T, Masuno M, Umeda Y, Kurosawa K.	Developmental delay and dysmorphic features in a girl with a de novo 5.4 Mb deletion of 13q12.11-q12.13.	**Congenit Ano m(Kyoto).**		PMID:3120 6199	2019
Nishimura N, Murakami H, S aito T, Masuno M, Kurosawa K.	Tumor predisposition in an individual with chromosomal rearran gements of 1q31.2-q4 1 encompassing cell division cycle protein	**Congenit Ano m (Kyoto).**		PMID:31595 586	2019

研究分担者・山本俊至

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Imaizumi T, Yamamoto-Shi mojima K, Yanagishita T, Ondo Y, Yamamoto T	Analyses of breakpoint-j unctions of complex gen omic rearrangements co mprising multiple consec utive microdeletions by nanopore sequencing.	**J Hum Genet**	In pres s.	In press.	2020
Suzuki T, Togawa T, Kann o H, Ogura H, Yamamoto T, Sugiura T, Kouwaki M, Saitoh S.	A novel α-spectrin patho genic variant in trans to α-spectrin LELY causin g neonatal jaundice with	**J Pediatr He mat/Onc**	In pres s.	In press.	2020
Kanda S, Ohmuraya M, A kagawa H, Horita S, Yoshi da Y, Kaneko N, Sugawara N, Ishizuka K, Miura K, Harita Y, Yamamoto T, O ka A, Araki K, Furukawa T, Hattori M.	Deletion in the cobalami n synthetase W domain-containing protein 1 gen e is associated with con genital anomalies of the kidney and urinary Tra ct.	**J Am Soc Ne phrol.**	31	139-147	2020
Yamamoto-Shimojima K, I maizumi T, Akagawa H, K anno H, Yamamoto T.	Primrose syndrome assoc iated with unclassified i mmunodeficiency and a novel ZBTB20 mutation.	**Am J Med G enet A.**	182	521-526	2020
Imaizumi T, Yamamoto-Shi mojima K, Yamamoto H, Yamamoto T..	Establishment of a simpl e and rapid method to d etect MECP2 duplication s using digital polymeras e chain reaction.	**Congenit Ano m (Kyoto)**	60	10-14	2020

Imaizumi T, Yamamoto-Shimojima K, Yamamoto H, Yamamoto T..	Establishment of a simple and rapid method to detect MECP2 duplications using digital polymerase chain reaction.	**Congenit Anom (Kyoto)**	60	10-14	2020
Yamamoto T, Imaizumi T, Yamamoto-Shimojima K, Lu Y, Yanagishita T, Shimada S, Chong PF, Kira R, Ueda R, Ishiyama A, Takeshita E, Momosaki K, Ozasa S, Akiyama T, Kobayashi K, Oomatsu H, Kitahara H, Yamaguchi T, Imai K, Kurahashi H, Okumura A, Oguni H, Seto T, Okamoto N	Genomic backgrounds of Japanese patients with undiagnosed neurodevelopmental disorders.	**Brain Dev**	41	776-782	2019
Sato T, Sugiura-Ogasawara M, Ozawa F, Yamamoto T, Kato T, Kurahashi H, Kuroda T, Aoyama N, Kato K, Kobayashi R, Fukuda A, Utsunomiya T, Kuwahara A, Saito H, Takeshita T, Irahara M.	Preimplantation genetic testing for aneuploidy: a comparison of live birth rates in patients with recurrent pregnancy loss due to embryonic aneuploidy or recurrent implantation failure.	**Hum Reprod.**	34	2340-2348	2019
Okumura A, Shimojima K, Kurahashi H, Numoto S, Shimada S, Ishii A, Ohmori I, Takahashi S, Awaya T, Kubota T, Sakakibara T, Ishihara N, Hattori A, Torisu H, Tohyama J, Inoue T, Haibara A, Nishida T, Yuhara Y, Miya K, Tanaka R, Hirose S, Yamamoto T.	PRRT2 mutations in Japanese patients with benign infantile epilepsy and paroxysmal kinesigenic dyskinesia.	**Seizure**	71	1-5	2019
Yanagishita T, Yamamoto-Shimojima K, Koike T, Nasu H, Takahashi Y, Akiyama T, Nagata S, Yamamoto T.	Compound heterozygous ALDH7A1 mutation causes the hemi-allelic expression in a patient with pyridoxine-dependent epilepsy.	**Tokyo Women's Medical University Journal**	3	73-77	2019
Hoshina T, Seto T, Shimono T, Sakamoto H, Okuyama T, Hamazaki T, Yamamoto T	Narrowing down the region responsible for 1q23.3q24.1 microdeletion by identifying the smallest deletion.	**Hum Genome Var**	6	47	2019
Yamamoto-Shimojima K, Kouwaki M, Kawashima Y, Itomi K, Momosaki K, Ozasa S, Okamoto N, Yokochi K, Yamamoto T.	Natural histories of patients with Wolf-Hirschhorn syndrome derived from variable chromosomal abnormalities.	**Congenit Anom (Kyoto)**	59	169-173	2019
Tomita Y, Chong P-F, Yamamoto T, Akaminea S, Imaizumi T, Kira R.	Sequential radiologic findings in osteopathia striata with cranial sclerosis.	**Diagn Interv Imaging**	100	529-531	2019

Imaizumi T, Yamamoto-Shimojima K, Yamamoro T.	Advantages of ddPCR in detection of PLP1 duplications.	**Intractable Rare Dis Res.**	8	198-202	2019
Yamamoto-Shimojima K, Imaizumi T, Aoki Y, Inoue K, Kaname T, Okuno Y, Muramatsu H, Kato K, Yamamoto T.	Elucidation of the pathogenic mechanism and potential treatment strategy for a female patient with spastic paraplegia derived from a single-nucleotide deletion in PLP1.	**J Hum Genet.**	64	665-671	2019
Miyamoto S, Nakashima M, Ohashi T, Hiraide T, Kurosawa K, Yamamoto T, Takanashi J, Osaka H, Inoue K, Miyazaki T, Wada Y, Okamoto N, Saitsu H.	A case of de novo splice site variant in SLC35A2 showing developmental delays, spastic paraplegia, and delayed myelination.	**Mol Genet Genomic Med**	7	e814	2019
Yanagishita T, Yamamoto-Shimojima K, Nakano S, Sasaki T, Shigematsu H, Imai K, Yamamoto T.	Phenotypic features of 1q41q42 microdeletion including WDR26 and FBXO28 are clinically recognizable: The first case from Japan.	**Brain Dev**	41	452-455	2019
Imaizumi T, Mogami Y, Okamoto N, Yamamoto-Shimojima K, Yamamoto T.	De novo 1p35.2 microdeletion including PUM1 identified in a patient with sporadic West syndrome.	**Congenit Anom (Kyoto).**	59	193-194	2019

研究分担者・涌井敬子

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Oda Y, Uchiyama Y, Motomura A, Fujita A, Azuma Y, Harita Y, Mizuguchi T, Yanagi K, Ogata H, Hata K, Kaname T, Matsubara Y, Wakui K, Matsumoto N.	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome.	**J Hum Genet**	64	1005-1014	2019
Yokota Y, Moteki H, Nishio SY, Yamaguchi T, Wakui K, Kobayashi Y, Ohyama K, Miyazaki H, Matsuoka R, Abe S, Kumakawa K, Takahashi M, Sakaguchi H, Uehara N, Ishino T, Kosho T, Fukushima Y, Usami SI.	Frequency and clinical features of hearing loss caused by STRC deletions.	**Sci Rep**	9	4408	2019