掲載した論文（発表題目）	発表者氏名	発表した場所（学会誌・雑誌等名）	発表した時期	国内・外の
TBX1 mutation identified by exome sequencing in a Japanese family with 22q11.2 deletion syndrome-like craniofacial features and hypocalcemia.	Ogata T, Niihori T, Tanaka N, Kawai M, Nagashima T, Funayama R, Nakayama K, Nakashima S, Kato F, Fukami M, Aoki Y, Matsubara Y	PLoS One.	2014	国外
Seven Novel Mutations in Bulgarian Patients with Acute Hepatic Porphyrias (AHP).	Dragneva S, Szyszka-Niagolov M, Ivanova A, Mateva L, Izumi R, Aoki Y, Matsubara Y.	JIMD Rep.	2014	国外
New BRAF knockin mice provide a pathogenetic mechanism of developmental defects and a therapeutic approach in cardio-facio-cutaneous syndrome.	Inoue SI, Moriya M, Watanabe Y, Miyagawa-Tomita S, Niihori T, Oba D, Ono M, Kure S, Ogura T, Matsubara Y, Aoki Y.	Hum Mol Genet.	2014	国外
GNE myopathy associated with congenital thrombocytopenia: A report of two siblings.	Izumi R, Niihori T, Suzuki N, Sasahara Y, Rikiishi T, Nishiyama A, Nishiyama S, Endo K, Kato M, Warita H, Konno H, Takahashi T, Tateyama M, Nagashima T, Funayama R, Nakayama K, Kure S, Matsubara Y, Aoki Y, Aoki M.	Neuromuscul Disord.	2014	国外
Targeted Next-Generation Sequencing Effectively Analyzed the Cystic Fibrosis Transmembrane Conductance Regulator Gene in Pancreatitis.	Nakano E, Masamune A, Niihori T, Kume K, Hamada S, Aoki Y, Matsubara Y, Shimosegawa T.	Dig Dis Sci.	2014	国外
A novel heterozygous MAP2K1 mutation in a patient with Noonan syndrome with multiple lentigines.	Nishi E, Mizuno S, Nanjo Y, Niihori T, Fukushima Y, Matsubara Y, Aoki Y, Kosho T.	Am J Med Genet A.	2014	国外
Molecular basis of non-syndromic hypospadias: systematic mutation screening and genome-wide copy-number analysis of 62 patients.	Kon M, Suzuki E, Dung VC, Hasegawa Y, Mitsui T, Muroya K, Ueoka K, Igarashi N, Nagasaki K, Oto Y, Hamajima T, Yoshino K, Igarashi M, Kato-Fukui Y, Nakabayashi K, Hayashi K, Hata K, Matsubara Y, Moriya K, Ogata T, Nonomura K,	Hum Reprod.	2015	国外

	Fukami M.			
Lack of genomic rearrangements involving the aromatase gene *CYP19A1* in breast cancer.	Fukami M*, Suzuki J, Nakabayashi K, Tsunashima R, **Ogata T**, Shozu M, Noguchi S:	*Breast Cancer*	2014	**国外**
Long term follow up study for a patient with Floating-Harbor syndrome due to a hotspot *SRCAP* mutation.	Nagasaki K*, Asami T, Sato H, Ogawa Y, Kikuchi T, Saitoh A, **Ogata T**, Fukami M:	*Am J Med Genet A*	2014	**国外**
Aromatase excess syndrome in a family with upstream deletion of *CYP19A1*.	Shihara D, Miyado M, Nakabayashi K, Shozu M, Nagasaki K, **Ogata T**, Fukami M*:	*Clin Endocrinol*	2014	**国外**
Compound heterozygous deletions in pseudoautosomal region 1 in an infant with mild manifestsations of Langer mesomelic dysplasia.	Tsuchiya T, Shibata M, Numabe H, Jinnno T, Nakabayashi K, Nishimura G, Nagai T, **Ogata T**, Fukami M*:	*Am J Med Genet A*	2014	**国外**
Disorder of sex development in an infant with molecularly confirmed 46,XY,+der(10)t(10;21)(q21.1;q21.3), –21	Yagasaki H*, Nakane T, Saito T, Koizumi K, Kobayashi K, **Ogata T**:	*Am J Med Genet A*	2014	**国外**
Prenatal Genetic testing for a microdeletion at chromosome 14q32.2 imprinted region leading to upd(14)pat-like phenotype.	Sasaki A, Sumie M, Eada S, Kosaki R, Kurosawa K, Fukami M, Sago H, **Ogata T**, Kagami M*:	*Am J Med Genet A*	2014	**国外**
Hypertrophic cells in hypophagic intrauterine growth retarded rats without catch-up growth.	Kitsuda K*, Yamaguchi R, Nagata E, Nakagawa Y, Ohzeki T, **Ogata T**, Ishii M, Nakanishi T:	Kitasato Med J	2014	**国外**

IMAGe syndrome: clinical and genetic implications based on Iinvestigations in three Japanese patients.	Kato F, Hamajima T, Hasegawa T, Amano N, Horikawa R, Nishimura G, Nakashima S, Fuke T, Sano S, Fukami M, **Ogata T***:	*Clin Endocrinol*	2014	**国外**
Genome-wide parent-of-origin DNA methylation analysis reveals the intricacies of the human imprintome and suggests a germline methylation independent establishment of imprinting.	Court F, Tayama C, Romanelli V, Martin-Trujillo A, Iglesias-Platas I, Okamura K, Sugahara N, Simon C, Moore H, Harness J, Keirstead H, Vicente Sanchez-Mut J, Kaneki E, Lapunzina P, Soejima H, Wake N, Esteller M, **Ogata T**, Hata K, Nakabayashi K, Monk D*:	*Genome Res*	2014	**国外**
Identification and functional characterization of two novel *NPR2* mutations in Japanese patients with short stature.	Amano N, Mukai T, Ito Y, Narumi S, Tanaka T, Yokoya S, **Ogata T**, Hasegawa T*:	*J Clin Endocrinol Metab*	2014	**国外**
TBX1 mutation identified by exome sequencing in a Japanese family with 22q11.2 deletion syndrome-like craniofacial features and hypocalcemia.	**Ogata T***, Niihori T, Tanaka N, Kawai M, Nagashima T, Funayama R, Nakayama K, Nakashim S, Kato F, Fukami M, Aoki Y, Matsubara Y:	*PLoS One*	2014	**国外**

A missense single-nucleotide polymorphism in the sialic acid acetyl esterase gene is associated with anti–PIT-1 antibody syndrome.	Yamamoto M, Iguchi G, Bando H, Fukuoka H, Suda K, Takahashi M, Nishizawa H, Matsumoto R, Tojo K, Mokubo A, **Ogata T**, Takahashi Y*:	*Endocr J*	2014	**国外**
Mutation Spectrum and Phenotypic Variation in Nine Patients with SOX2 abnormalities.	Suzuki J, Azuma N, Dateki S, Soneda S, Muroya K, Yamamoto Y, Saito R, Sano S, Nagai T, Wada H, Endo A, Urakami T, **Ogata T**, Fukami M*:	*J Hum Genet* 59	2014	**国外**
Uniparental disomy of chromosome 8 leading to homozygosity of a *CYP11B1* mutation in a patient with congenital adrenal hyperplasia: Implication for a rare etiology of an autosomal recessive disorder.	Matsubara K, Kataoka N, Ogita S, Sano S, **Ogata T**, Fukami M*, Katsumata N:	*Endocr J*	2014	**国外**
Early vitamin K deficiency bleeding in a neonate associated with maternal Crohn's disease.	Ohishi A, Nakashima S, **Ogata T**, Iijima S:	*J Perinatol*	2014	**国外**
De novo frameshift mutation in fibroblast growth factor 8 in a male patient with gonadotropin deficiency.	Suzuki E, Yatsuga S, Igarashi M, Miyado M, Nakabayashi K, Hayashi K, Hata K, Umezawa A, Yamada G, **Ogata T**, Fukami M*:	*Horm Res Paediatr*	2014	**国外**

Rationale and study design of the Japan environment and children's study (JECS).	Kawamoto T, Nitta H, Murata K, Toda E, Tsukamoto N, Hasegawa M, Yamagata Z, Kayama F, Kshi R, Ohya Y, Saito H, Sago H, Okuyama M, **Ogata T**, Yokoya S, Koresawa Y, Shibata Y, Nakayama S, Michikawa T, Takeuchi A, Saitoh H:	*BMC Public Health* 2014 Jan 10; 14:25.	2014	**国外**
A novel *de novo* point mutation of OCT-binding site in the *IGF2*/*H19*-imprinting control region in a patient with Beckwith-Wiedemann syndrome patient.	Higashimoto K, Jozaki K, Kosho T, Matsubara K, Sato T, Yamada D, Yatsuki H, Maeda T, Ohtsuka Y, Nishioka K, Joh K, Koseki H, **Ogata T**, SoejimaH*:	*Clin Genet*	2014	**国外**

Comprehensive and quantitative multilocus methylation analysis reveals the susceptibility of specific imprinted differentially methylated regions (DMRs) to aberrant methylation in Beckwith-Wiedemann syndrome with epimutations.	Maeda T, Higashimoto K, Jozaki K, Hitomi H, Nakabayashi K, Makita Y, Tonoki H, Okamoto N, Takada F, Ohashi H, Migita M, Kosaki R, Matsubara K, **Ogata T**, Matsuo M, Hamasaki Y, Ohtsuka Y, Nishioka K, Joh K, Mukai T, Hata K, Soejima H*:	*Genet Med*	2014	**国外**
Genome-wide copy number analysis and systematic mutation screening in 58 patients with hypogonadotropic hypogonadism.	Izumi Y, Suzuki E, Kanzaki S, Yatsuga S, Kinjo S, Igarashi M, Maruyama T, Sano S, Horikawa R, Sato N, Nakabayashi K, Hata K, Umezawa A, Ogata T, Yoshimura Y, Fukami M*:	*Fertil Steril*	2014	**国外**
Hemodynamic assessment in a child with renovascular hypertension using time-resolved three-dimensional cine phase-contrast MRI.	Ishikawa T*, Takehara Y, Yamashita S, Iwashima S, Sugiyama M, Wakayama T, Johnson K, Wieben O, Sakahara H, **Ogata T**:	*J Magn Reson Imaging*	2015	**国外**

Clinical and molecular studies in four patients with *SRY*-positive 46,XX testicular disorders of sex development: implications for variable sex development and genomic rearrangements.	Nakashima S, Oishi A, Takada F, Kawamura H, Igarashi M, Fukami M, **Ogata T***:	*J Hum Genet*	2014	**国外**
Japanese founder duplications/triplications involving *BHLHA9* are associated with split-hand/foot malformation with or without long bone deficiency and Gallop-Wolfgang complex.	Nagata E, Kano H, Kato F, Yamaguchi R, Nakashima S, Takayams S, Kosaki R, Tonoki H, Mizuno S, Watanabe S, Yoshiura K, Kosho T, Hasegawa T, Kimizuka M, Suzuki A, Shimizu K, Ohashi H, Haga N, Numabe H, Horii E, Nagai T, Yoshihashi H, Nishimura G, Toda T, Takada S, Yokoyama S, Asahara H, Sano S, Fukami M, Ikegawa S, **Ogata T***:	*Orphanet J Rare Dis*	2014	
Epimutations of the IG-DMR and the *MEG3*-DMR at the 14q32.2 imprinted region in two patients with Silver-Russell syndrome-compatible phenotype.	Kagami M, Mizuno S, Matsubars K, Nakabayashi K, Sano S, Fuke T, Fukami M, **Ogata T***:	*Eur J Hum Genet* (in press)		国外
Hypogonadotropic hypogonadism in a female patient previously diagnosed as having Waardenburg syndrome due to a *SOX10* mutation.	Izumi Y, Musha I, Suzuki E, Iso M, Jinno T, Horikawa R, Amemiya S, **Ogata T**, Fukami M, Ohtake A:	*Endocrine* (in press)		国外

Silver-Russell syndrome without body asymmetry in three patients with duplications of maternally derived chromosome 11p15 involving *CDKN1C*.	Nakashima S, Kato F, Kosho T, Nagasaki K, Kikuchi T, Kagami M, Fukami M, **Ogata T***:	*J Hum Genet* (in press)		国外
Detecting copy number variations in whole exome sequencing data using exome hidden markov model - an expectation of "exome-first" approach.	Miyatake S, Koshimizu E, Fujita A, Fukai R, Imagawa E, Ohba C, Kuki I, Makita Y, **Ogata T**, Nakashima M, Tsurusaki Y, Miyake N, Saitsu H, Matsumoto N*:	*J Hum Genet* (in press)		国外
Copy-number variations in Y chromosomal azoospermia factor regions identified by multiplex ligation-dependent probe amplification.	Saito K, Miyado M, Kobori Y, Tanaka Y, Ishikawa H, Yoshida A, Katsumi M, Saito H, Kubota T, Okada H, Ogata T, Fukami M*:	*J Hum Genet* (in press)		国外
Comprehensive clinical studies in 34 patients with molecularly defined UPD(14)pat and related conditions (Kagami-Ogata syndrome).	Kagami M, Kurosawa K, Miyazaki O, Ishino F, Matsuoka K, **Ogata T***:	*Eur J Hum Genet* (in press)		国外
Novel splice site mutation in *MAMLD1* in a patient with hypospadias.	Igarashi M, Wada Y, Kojima Y, Miyado M, Nakamura M, Muroya K, Mizuno K, Hayashi Y, Nonomura K, Jofri K, **Ogata T**, Fukami M*:	*Sex Dev* (in press)		国外

Molecular basis of non-syndromic hypospadias: Systematic mutation screening and genome-wide copy-number analysis of 62 patients.	Kon M, Suzu ki E, Dung VC, Hasegawa Y, Mitsui T, Muroya K, Ueoka K, Igarashi N, Nagasaki K, Oto Y, Hamajima T, Yoshino K, Igarashi M, Kato-Fukui Y, Nakabayashi K, Hayashi K, Hata K, Matsubara Y, Moriya K, **Ogata T**, Nonomura K, Fukami M*:	*Hum Reprod* (in press)		国外
Impact of a novel homozygous mutation in nicotinamide nucleotide transhydrogenase on mitochondrial DNA integrity in a case of familial glucocorticoid deficiency.	Fujisawa Y, Napoli E, Wong S, Song G, Yamaguchi R, Matsui T, Nagasaki K, **Ogata T**, Giulivi C	*BBA Clinical* (in press)		国外
SOX3 overdosage permits normal sex development in females with random X inactivation.	Igarashi M, Mikami H, Katsumi M, Miyado M, Izumi Y, **Ogata T**, Fukami M*:	*Sex Dev* (in press)		国外

Microhomology-Mediated Microduplication in the Y Chromosomal Azoospermia Factor a (AZFa) Region in a Male with Mild Asthenozoospermia.	Momori Katsumi[a], Hiromichi Ishikawa[b], Yoko Tanaka[c], Kazuki Saito[a, d], Yoshitomo Kobori[e], Hiroshi Okada[e], Hidekazu Saito[d], Kazuhiko Nakabayashi[f], Yoichi Matsubara[g], Tsutomu Ogata[a, h], Maki Fukami[a], Mami Miyado[a]	Cytogenetic and Genome Research (in press)		国外
Aromatase excess syndrome: a rare autosomal dominant disorder leading to pre- or peri-pubertal onset gynecomastia.	Fukami M*, Miyado M, Nagasaki K, Shozu M, **Ogata T**:	*Pediatr Endocr Rev*	2014	国外
Understanding the pathological manifestations of aromatase excess syndrome: lessons for the clinic.	Shozu M*, Fukami M, **Ogata T**:	Expert Rev Endocrinol Metab	2014	国外
Cytochrome P450 oxidoreductase deficiency: Rare congenital disorder leading to skeletal malformations and steroidogenic defects.	Fukami M*, **Ogata T**:	*Pediatr Int* 2014 Oct 8.		国外
Anti-tumor effects of suberoylanilide hydroxamic acid on Epstein-Barr virus-associated T- and natural killer- cell lymphoma	Siddiquey MN, Nakagawa H, Iwata S, Kanazawa T, Suzuki M, Imadome KI, Fujiwara S, Goshima F, Murata T, Kimura H.	*Cancer Sci*	2014; 105(6):713-22.	国外
CD137 expression is induced by Epstein-Barr virus infection through LMP1 in T or NK cells and mediates survival promoting signals.	Yoshimori M, Imadome KI, Komatsu H, Wang L, Saitoh Y, Yamaoka S, Fukuda T, Kurata M, Koyama T, Shimizu N, Fujiwara S, Miura O, Arai A.	*PLoS ONE*	2014 Nov 19;9(11):e112564.	国外

Modeling EBV infection and pathogenesis in new-generation humanized mice	Fujiwara S, Imadome K, and Takei M.	*Exp Mol Med*	47, e136; doi:10.1038/emm.2014.102 Published online 23 January 2015	国外
Bilateral giant coronary aneurysms in a 40-year-old male with Noonan syndrome caused by a KRAS germline mutation.	Fujimoto N, Nakajima H, Sugiura E, Dohi K, Kanemitsu S, Yamada N, Aoki Y, Nakatani K, Shimpo H, Nobori T, Ito M.	Int J Cardiol.	173(3):e63-6, 2014	国外
Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis.	Flex E, Jaiswal M, Pantaleoni F, Martinelli S, Strullu M, Fansa EK, Caye A, De Luca A, Lepri F, Dvorsky R, Pannone L, Paolacci S, Zhang SC, Fodale V, Bocchinfuso G, Rossi C, Burkitt-Wright EM, Farrotti A, Stellacci E, Cecchetti S, Ferese R, Bottero L, Castro S, Fenneteau O, Brethon B, Sanchez M, Roberts AE, Yntema HG, van der Burgt I, Cianci P, Bondeson ML, Digilio MC, Zampino G, Kerr B, Aoki Y, Loh ML, Palleschi A, Di Schiavi E, Carè A, Selicorni A, Dallapiccola B, Cirstea IC, Stella L, Zenker M, Gelb BD, Cavé H, Ahmadian MR, Tartaglia M.	Hum Mol Genet.	23:4315-27, 2014	国外

Identification of acquired mutations by whole-genome sequencing in GATA-2 deficiency evolving into myelodysplasia and acute leukemia	Fujiwara T, Fukuhara N, Funayama R, Nariai N, Kamata M, Nagashima T, Kojima K, Onishi Y, Sasahara Y, Ishizawa K, Nagasaki M, Nakayama K, Harigae H	Ann Hematol	**平成24年4月**	**国外**
HCV infection enhances Th17 commitment, which could affect the pathogenesis of autoimmune diseases	Kondo Y, Ninomiya M, Kimura O, Machida K, Funayama R, Nagashima T, Kobayashi K, Kakazu E, Kato T, Nakayama K, Lai M. M, Shimosegawa T	PLoS One	**平成24年6月**	**国外**
GNE myopathy associated with congenital thrombocytopenia: A report of two siblings	Izum, R, Niihori T, Suzuki N, Sasahara Y, Rikiishi T, Nishiyama A, Nishiyama S, Endo K, Kato M, Warita H, Konno H, Takahashi T, Tateyama M, Nagashima T, Funayama R, Nakayama K, Kure S, Matsubara Y, Aoki Y, Aoki M	Neuromuscul Disord	**平成24年12月**	**国外**
CRL4(VprBP) E3 Ligase Promotes Monoubiquitylation and Chromatin Binding of TET Dioxygenases.	Nakagawa T, Lv L, Nakagawa M, Yu Y, Yu C, D'Alessio A. C, Nakayama K, Fan H. Y, Chen X, Xiong Y.	Mol Cell	**平成25年1月**	**国外**
舞踏運動を呈したdysferlin**異常症の**1**例**.	**高橋俊明、今井尚志、田中洋康、吉岡勝、今野秀彦、日向野修一、小野寺好明、齋藤博、木村格、糸山泰人、武田篤、青木正志**	JMDD（**運動障害**）	2014	**国内**
GNE myopathy associated with congenital thrombocytopenia: a report of two siblings.	Izumi R, Niihori T, Suzuki N, Sasahara Y, Rikiishi T, Nishiyama A, Nishiyama S, Endo K, Kato M, Warita H, Konno H, Takahashi T, Tateyama M, Nagashima T, Funayama R, Nakayama K, Kure S, Matsubara Y, Aoki Y, Aoki M.	Neuromuscul Disord.	2014	**国外**

Targeted next-Generation sequencing effectively analyzed the cystic fibrosis transmembrane conductance regulator gene in pancreatitis.	Nakano E, Masamune A, Niihori T, Kume K, Hamada S, Aoki Y, Matsubara Y, Shimosegawa T.	Dig Dis Sci.	2014 Dec 10.	**国外**
Variants in the interferon regulatory factor-2 gene are not associated with pancreatitis in Japan.	Nakano E, Masamune A, Kume K, Kakuta Y, Shimosegawa T.	Pancreas.	2014;43:1125-1126.	**国外**
膵炎の原因遺伝子探索	**正宗　淳、中野絵里子、粂潔、新堀哲也、青木洋子、下瀬川徹.**	**膵臓**	2014;29:51-58.	**国内**
膵炎の原因遺伝子はどこまで解ったか.	**正宗　淳、中野絵里子、粂潔、新堀哲也、青木洋子、下瀬川徹.**	**肝胆膵**	2014;69:1115-1121.	**国内**
遺伝性膵炎・家族性膵炎	**正宗　淳、下瀬川徹.**	**胆と膵**	2014;25:1137-1141.	**国内**
A novel heterozygous MAP2K1 mutation in a patient with Noonan syndrome with multiple lentigines.	Nishi E, Mizuno S, Nanjo Y, Niihori T, Fukushima Y, Matsubara Y, Aoki Y, Kosho T	Am J Med Genet A.	167(2):407-11, 2015	**国外**

X-Linked Agammaglobulinemia Associated with B-Precursor Acute Lymphoblastic Leukemia.	Hoshino A, Okuno Y, Migita M, Ban H, Yang X, Kiyokawa N, Adachi Y, Kojima S, Ohara O, Kanegane H.	J Clin Immunol. 2015 Jan 16. [Epub ahead of print]	2015, Jan 16	**国外**
Aicardi-Goutières syndrome is caused by IFIH1 mutations.	Oda H, Nakagawa K, Abe J, Awaya T, Funabiki M, Hijikata A, Nishikomori R, Funatsuka M, Ohshima Y, Sugawara Y, Yasumi T, Kato H, Shirai T, Ohara O, Fujita T, Heike T.	Am J Hum Genet. 2014 Jul 3;95(1):121-5.	2014, July	**国外**
A complement factor B mutation in a large kindred with atypical hemolytic uremic syndrome.	Funato M, Uemura O, Ushijima K, Ohnishi H, Orii K, Kato Z, Yamakawa S, Nagai T, Ohara O, Kaneko H, Kondo N.	J Clin Immunol. 2014 Aug;34(6):691-5.	2014, Aug	**国外**
Clinical and genetic characterization of Japanese sporadic cases of periodic Fever, aphthous stomatitis, pharyngitis and adenitis syndrome from a single medical center in Japan.	Kubota K, Ohnishi H, Teramoto T, Kawamoto N, Kasahara K, Ohara O, Kondo N.	J Clin Immunol. 2014 Jul;34(5):584-93.	2014, July	**国外**
Somatic NLRP3 mosaicism in Muckle-Wells syndrome. A genetic mechanism shared by different phenotypes of cryopyrin-associated periodic syndromes.	Nakagawa K, Gonzalez-Roca E, Souto A, Kawai T, Umebayashi H, Campistol JM, Cañellas J, Takei S, Kobayashi N, Callejas-Rubio JL, Ortego-Centeno N, Ruiz-Ortiz E, Rius F, Anton J, Iglesias E, Jimenez-Treviño S, Vargas C, Fernandez-Martin J, Calvo I, Hernández-Rodríguez J, Mendez M, Dordal MT, Basagaña M, Bujan S,	Ann Rheum Dis. 2015 Mar;74(3):603-10.	2015, March	**国外**

	Yashiro M, Kubota T, Koike R, Akuta N, Shimoyama K, Iwata N, Saito MK, Ohara O, Kambe N, Yasumi T, Izawa K, Kawai T, Heike T, Yagüe J, Nishikomori R, Aróstegui JI.			
Merkel Cell Polyomavirus-positive Merkel Cell Carcinoma in a Patient with Epidermodysplasia Verruciformis.	Mizuno Y, Kato G, Shu E, Ohnishi H, Fukao T, Ohara O, Fukumoto H, Katano H, Seishima M.	Acta Derm Venereol. 2015 Jan 15;95(1):98-99.	2015, Jan	**国外**
Age-related decrease of meiotic cohesins in human oocytes.	Tsutsumi M, Fujiwara R, Nishizawa H, Ito M, Kogo H, Inagaki H, Ohye T, Kato T, Fujii T, Kurahashi H.	PLoS One 2014: 9(5): e96710.	2014	**国外**
Dual roles for the telomeric repeats in chromosomally integrated human herpesvirus-6.	Ohye T, Inagaki H, Ihira M, Higashimoto Y, Kato K, Oikawa J, Yagasaki H, Niizuma T, Takahashi Y, Kojima S, Yoshikawa T, Kurahashi H.	Sci Rep 2014: 4: 4559.	2014	**国外**
Breakpoint analysis of the recurrent constitutional t(8;22)(q24.13;q11.21) translocation.	Mishra D, Kato T, Inagaki H, Kosho T, Wakui K, Kido Y, Sakazume S, Taniguchi-Ikeda M, Morisada N, Iijima K, Fukushima Y, Emanuel BS, Kurahashi H.	Mol Cytogenet 2014: 7: 55.	2014	**国外**
Signature of backward replication slippage at the copy number variation junction.	Ohye T, Inagaki H, Ozaki M, Ikeda T, Kurahashi H.	J Hum Genet 2014: 59(5): 247-250.	2014	**国外**

Analysis of the t(3;8) of hereditary renal cell carcinoma: A palindrome mediated translocation.	Kato T, Franconi CP, Sheridan MB, Hacker AM, Inagakai H, Glover TW, Arlt MF, Drabkin HA, Gemmill RM, Kurahashi H, Emanuel BS.	Cancer Genet 2014: 207(4): 133-140.	2014	**国外**
Recurrent Pregnancy loss.	Nisizawa H, Kurahashi H.	Clinical Genomics: Practical Applications in Adult Patient Care, Michael Murray, Mark Babyatski, and Monica Giovanni, McGraw-Hill Professional,	2014	**国外**
Prevalence of Emanuel syndrome: theoretical frequency and surveillance result.	Ohye T, Inagaki H, Kato T, Tsutsumi M, Kurahashi H.	Pediatr Int 2014: 56(4): 462-466.	2014	**国外**
Molecular and virological evidence of viral activation from chromosomally integrated HHV-6A in a patient with X-SCID.	Endo A, Watanabe K, Ohye T, Suzuki K, Matsubara T, Shimizu N, Kurahashi H, Yoshikawa T, Katano H, Inoue N, Imai K, Takagi M, Morio T, Mizutani S.	Clin Infect Dis 2014: 59(4): 545-548.	2014	**国外**
A patient of Autosomal Recessive Alport syndrome due to Segmental Maternal Isodisomy.	Fu XJ, Morisada N, Hashimoto F, Taniguchi-Ikeda M, Hashimura Y, Ohtsubo H, Ninchoji T, Kaito H, Nozu K, Takahashi E, Nakanishi K, Kurahashi H, Iijima K.	Hum Genome Variation 2014, 1, 14006.	2014	**国外**

Novel FATP4 mutations responsible for ichthyosis prematurity syndrome in a Japanese patient.	Tsuge I, Morishita M, Kato T, Tsutsumi M, Inagaki H, Mori Y, Yamawaki K, Inuo C, Ieda K, Ohye T, Hayakawa A, Kurahashi H.	Hum Genome Variation, in press.	2015	**国外**
Acute eosinophilic pneumonia occurring in a dedicator of cytokinesis 8 (DOCK8) deficient patient.	Tsuge I, Ito K, Ohye T, Kando N, Kondo Y, Nakajima Y, Inuo C, Kurahashi H, Urisu A.	Pediatric Pulmonol 2014: 49(3): E52-55.	2014	**国外**
An immunocompetent child with chromosomally integrated human herpes virus 6B accidentally identified during the care of Mycoplasma pneumoniae infection.	Oikawa J, Tanaka J, Yoshikawa T, Morita Y, Hishiki H, Ishiwada N, Ohye T, Kurahashi H, Kohno Y.	J Infect Chemother 2014: 20(1): 65-67.	2014	**国外**
Serum liver-type fatty acid-binding protein predicts recovery of graft function after kidney transplantation from donors after cardiac death.	Kawai A, Kusaka M, Kitagawa F, Ishii J, Fukami N, Maruyama T, Sasaki H, Shiroki R, Kurahashi H, Hoshinaga K.	Clin Transplant 2014:2 8(6): 749-754.	2014	**国外**
Nine-year follow-up in a child with chromosomal integration of human herpesvirus 6 transmitted from an unrelated donor through the Japan Marrow Donor Program.	Yagasaki H, Shichino H, Shimizu N, Ohye T, Kurahashi H, Yoshikawa T, Takahashi S.	Transplant Infect Dis, in press.	2015	**国外**
Pheochromocytoma as the first manifestation of MEN2A with RET mutation S891A: report of a case.	Hibi Y, Ohye T, Ogawa K, Shimizu Y, Shibata M, Kagawa C, Mizuno Y, Uchino S, Kosugi S, Kurahashi H, Iwase K.	Surgery Today 2014: 44(11): 2195-2200.	2014	**国外**

A MEN2A family with two asymptomatic carriers affected by unilateral renal agenesis.	Hibi Y, Ohye T, Ogawa K, Shimizu Y, Shibata M, Kagawa C, Mizuno Y, Kurahashi H, Iwase K.	Endocr J 2014: 61(1): 19-23.	2014	**国外**
Accurate prediction for the stage of histological chorioamnionitis before delivery by amniotic fluid IL-8 level.	Yoneda S, Shiozaki A, Ito M, Yoneda N, Inada K, Yonezawa R, Kigawa M, Saito S.	Am J Reprod Immunol.	in press	**国外**
Extensive serum biomarker analysis in patients with enterohemorrhagic Escherichia coli O111-induced hemolytic-uremic syndrome.	Shimizu M, Kuroda M, Inoue N, Konishi M, Igarashi N, Taneichi H, Kanegane H, Ito M, Saito S, Yachie A.	Cytokine. 66:1-6	2014	**国外**
Whole exome sequencing revealed biallelic IFT122 mutations in a family with CED1 and recurrent pregnancy loss.	Tsurusaki Y, Yonezawa R, Furuya M, Nishimura G, Pooh RK, Nakashima M, Saitsu H, Miyake N, Saito S, Matsumoto N.	Clin Genet 85:592-594	2014	**国外**
Multiple pregnancy,short cervix, part-time worker, steroid use, low educational level, and male fetus are risk factors for preterm birth in Japan: A multicenter, prospective study.	Shiozaki A, Yoneda S, Nakabayashi M, Takeda Y, Takeda S, Sugimura M, Yoshida K, Tajima A, Manabe M, Akagi K, Nakagawa S, Tada K, Imafuku N, Ogawa M, Mizunoe T, Kanayama N, Itoh H, Minoura S, Ogino M, Saito S.	J Obstet Gynaecol Res 40: 53–61.	2014	**国外**