M, Watanabe Y, Bartley J, Abdenur JE, Wang RY, Chang R, Tsurusaki Y, Doi H, Nakashima M, Saitsu H, Ogata K, Goto Y, Matsumoto N. Mitochondrial complex III deficiency caused by a homozygous UQCRC2 mutation presenting with neonatal-onset recurrent metabolic decompensation. ***Hum Mutat.*** 2013 Mar;34(3):446-52.

17. Miyake N, Koshimizu E, Okamoto N, Mizuno S, Ogata T, Nagai T, Kosho T, Ohashi H, Kato M, Sasaki G, Mabe H, Watanabe Y, Yoshino M, Matsuishi T, Takanashi J, Shotelersuk V, Tekin M, Ochi N, Kubota M, Ito N, Ihara K, Hara T, Tonoki H, Ohta T, Saito K, Matsuo M, Urano M, Enokizono T, Sato A, Tanaka H, Ogawa A, Fujita T, Hiraki Y, Kitanaka S, Matsubara Y, Makita T, Taguri M, Nakashima M, Tsurusaki Y, Saitsu H, Yoshiura K, Matsumoto N, Niikawa N. MLL2 and KDM6A mutations in patients with Kabuki syndrome. ***Am J Med Genet A.*** 2013 Sep;161(9):2234-2243.
18. Nakashima S, Watanabe Y, Okada J, Ono H, Nagata E, Fukami M, Ogata T. Critical role of Yp inversion in PRKX/PRKY-mediated Xp;Yp translocation in a patient with 45,X testicular disorder of sex development. ***Endocr J*** 2013. 60(12):1329-1334.
19. Yatsuki H, Higashimoto K, Jozaki K, Koide K, Okada J, Watanabe Y, Okamoto N, Tsuno Y, Yoshida Y, Ueda K, Shimizu K, Ohashi H, Mukai T, Soejima H. Novel mutations of CDKN1C in Japanese patients with Beckwith-Wiedemann syndrome. ***Genes Genom*** (2013) 35:141–147
20. Sasaki K, Mishima H, Miura K, Yoshiura KI. Uniparental disomy analysis in trios using genome-wide SNP array and whole-genome sequencing data imply segmental uniparental isodisomy in general populations. ***Gene*** 2013. 512(2):267-274.
21. Yamada A, Ishikawa T, Ota I, Kimura M, Shimizu D, Tanabe M, Chishima T, Sasaki T, Ichikawa Y, Morita S, Yoshiura KI, Takabe K, Endo I. High expression of ATP-binding cassette transporter ABCC11 in breast tumors is associated with aggressive subtypes and low disease-free survival. ***Breast Cancer Res Treat*** 137(3): 773-782, 2013.
22. Ishikawa T, Toyoda Y, Yoshiura K, Niikawa N. Pharmacogenetics of human ABC transporter ABCC11: new insights into apocrine gland growth and metabolite secretion. 2013. ***Front Genet*** 3: 306.
23. Higashijima A, Miura K, Mishima H, Kinoshita A, Jo O, Abe S, Hasegawa Y, Miura S, Yamasaki K, Yoshida A, Yoshiura K, Masuzaki H. Characterization of placenta-specific microRNAs in fetal growth restriction pregnancy. ***Prenat Diagn*** 2013. 33(3): 214-222.
24. Higashimoto K, Maeda T, Okada J, Ohtsuka Y, Sasaki K, Hirose A, Nomiyama M, Takayanagi T, Fukuzawa R, Yatsuki H, Koide K, Nishioka K, Joh K, Watanabe Y, Yoshiura KI, Soejima H. Homozygous deletion of DIS3L2 exon 9 due to non-allelic homologous recombination between LINE-1s in a Japanese patient with Perlman syndrome. ***Eur J Hum Genet*** 2013. 21(11):1316-1319.
25. Abe S, Miura K, Kinoshita A, Mishima H, Miura S, Yamasaki K, Hasegawa Y, Higashijima A, Jo O, Sasa K, Yoshida A, Yoshiura K, Masuzaki H. Copy number variation of the antimicrobial-gene, defensin beta 4, is associated with susceptibility to cervical cancer. ***J Hum Genet*** 58(5): 250-253. 2013.
26. Hamaguchi D, Miura K, Abe S, Kinoshita A, Miura S, Yamasaki K, Yoshiura KI, Masuzaki H. Initial viral load in cases of single human papillomavirus 16 or 52 persistent infection is associated with progression of later cytopathological findings in the uterine cervix. ***J Med Virol*** 2013 85(12): 2093-2100.
27. Nakao K, Oikawa M, Arai J, Mussazhanova Z, Kondo H, Shichijo K, Nakashima M, Hayashi T, Yoshiura K, Hatachi T, Nagayasu T. A Predictive Factor of the Quality of Microarray Comparative Genomic Hybridization Analysis for Formalin-fixed Paraffin-embedded Archival Tissue. 2013. ***Diagn Mol Pathol*** 22(3):174-180.
28. Hasegawa Y, Miura K, Furuya K, Yoshiura K, Masuzaki H. Identification of Complete Hydatidiform

Mole Pregnancy-Associated MicroRNAs in Plasma. ***Clin Chem*** 2013. 59(9):1410-1412.

29. Nakazawa Y, Sasaki K, Mitsutake N, Matsuse M, Shimada M, Nardo T, Takahashi Y, Ohyama K, Ito K, Mishima H, Nomura M, Kinoshita A, Ono S, Takenaka K, Masuyama R, Kudo T, Slor H, Utani A, Tateishi S, Yamashita S, Stefanini M, Lehmann AR, Yoshiura KI, Ogi T. Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase IIo processing in transcription-coupled nucleotide-excision repair. ***Nat Genet*** 44(5): 586-592, 2012
30. Matsuse M, Sasaki K, Nishihara E, Minami S, Hayashida C, Kondo H, Suzuki K, Saenko V, Yoshiura K, Mitsutake N, Yamashita S. Copy number alteration and uniparental disomy analysis categorizes Japanese papillary thyroid carcinomas into distinct groups. ***PLoS One*** 7(4): e36063, 2012 April.
31. Ono S, Yoshiura K, Kinoshita A, Kikuchi T, Nakane Y, Kato N, Sadamatsu M, Konishi T, Nagamitsu S, Matsuura M, Yasuda A, Komine M, Kanai K, Inoue T, Osamura T, Saito K, Hirose S, Koide H, Tomita H, Ozawa H, Niikawa N, Kurotaki N. Mutations in PRRT2 responsible for paroxysmal kinesigenic dyskinesias also cause benign familial infantile convulsions. ***J Hum Genet*** 57(5): 338-341, 2012 May.
32. Arai J, Tsuchiya T, Oikawa M, Mochinaga K, Hayashi T, Yoshiura KI, Tsukamoto K, Yamasaki N, Matsumoto K, Miyazaki T, Nagayasu T. Clinical and molecular analysis of synchronous double lung cancers. ***Lung Cancer*** 77(2): 281-287, 2012 Aug.
33. Mishima H, Aerts J, Katayama T, Bonnal RJ, Yoshiura K. The Ruby UCSC API: accessing the UCSC genome database using Ruby. ***BMC Bioinformatics*** 13: 240, 2012 Sep.
34. Hikida M, Tsuda M, Watanabe A, Kinoshita A, Akita S, Hirano A, Uchiyama T, Yoshiura KI. No evidence of association between 8q24 and susceptibility to nonsyndromic cleft lip with or without palate in Japanese population. ***Cleft Palate Craniofac J*** 49(6): 714-717, 2012 Nov.
35. Ogi T, Walker S, Stiff T, Hobson E, Limsirichaikul S, Carpenter G, Prescott K, Suri M, Byrd P, Matsuse M, Mitsutake N, Nakazawa Y, Vasudevan P, Barrow M, Stewart G, Taylor M, O'Driscoll M, and Jeggo P. Identification of the first ATRIP-deficient patient and novel mutations in ATR define a clinical spectrum for ATR-ATRIP Seckel syndrome. ***PLoS Genetics***, **8**: e1002945, 2012.
36. Jinam T, Nishida N, Hirai M, Kawamura S, OOta H, Umetsu K, Kimura R, Ohashi J, Tajima A, Yamamoto T, Tanabe H, Mano S, Suto Y, Kaname T, Naritomi K, Yanagi K, Niikawa N, Omoto K, Tokunaga K, Saitou N. The history of human populations in the Japanese Archipelago inferred from genomewide SNP data with a special reference to the Ainu and the Ryukyuan populations. ***J Hum Genet***, 57: 787-795, 2012.
37. Yanagi K, Kaname T, Wakui K, Hashimoto O, Fukushima Y, Naritomi K. Identification of four novel synonymous substitutions in the X-linked genes neuroligin 3 and neuroligin 4X in Japanese patients with autistic spectrum disorder. ***Autism Res Treat***, 2012 : 724072, 2012.
38. Cheung W, Kotzamanis G, Abdulrazzak H, Goussard S, Kaname T, Kotsinas A, Gorgoulis VG, Grillot-Courvalin C, Huxley C. Bacterial delivery of large intact genomic-DNA-containing BACs into mammalian cells. ***Bioeng Bugs***, 3(2): 86-92, 2012.
39. Kaname T, Yanagi K, Maehara H. Osteosarcoma and midkine. '***Midkine: From embryogenesis to pathogenesis and medication***' Ergüven M, Billir A, Muramatsu T. Eds. Springer, New York, pp313-319, 2012.
40. Tsurusaki Y, Okamoto N, Ohashi H, Kosho T, Imai Y, Hibi-Ko Y, Kaname T, Naritomi K, Kawame H, Wakui K, Fukushima Y, Homma T, Kato M, Hiraki Y, Yamagata T, Yano S, Mizuno S, Sakazume S, Ishii T, Nagai T, Shiina M, Ogata K, Ohta T, Niikawa N, Miyatake S, Okada I, Mizuguchi T, Doi H, Saitsu H, Miyake N, Matsumoto N. Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. ***Nat Genet,*** 44(4): 376-378, 2012.
41. He Y, Wang WR, Xu S, Jin L; Pan-Asia SNP Consortium. Paleolithic Contingent in Modern Japanese: Estimation and Inference using Genome-wide Data. ***Sci Rep***, 2:355, 2012.

42. Migita K, Uehara R, Nakamura Y, Yasunami M, Tsuchiya-Suzuki A, Yazaki M, Nakamura A, Masumoto J, Yachie A, Furukawa H, Ishibashi H, Ida H, Yamazaki K, Kawakami A, Agematsu K. Familial Mediterranean Fever in Japan. ***Medicine (Baltimore)*** 91: 337-343, 2012.
43. Migita K, Ida H, Moriuchi H, Agematsu K. Clinical relevance of MEFV gene mutations in Japanese patients with unexplained fever. J Rheumatol. 39: 875-877, 2012.
44. Higashimoto K, Nakabayashi K, Yatsuki H, Yoshinaga H, Jozaki K, Okada J, Watanabe Y, Aoki A, Shiozaki A, Saito S, Koide K, Mukai T, Hata K, Soejima H. Aberrant methylation of H19-DMR acquired after implantation was dissimilar in soma versus placenta of patients with Beckwith-Wiedemann syndrome. ***Am J Med Genet A.*** 158A(7): 1670-1675, July 2012.
45. Higashimoto K, Maeda T, Okada J, Ohtsuka Y, Sasaki K, Hirose A, Nomiyama M, Takayanagi T, Fukuzawa R, Yatsuki H, Koide K, Nishioka K, Joh K, Watanabe Y, Yoshiura K, Soejima H. Homozygous deletion of *DIS3L2* exon 9 due to non-allelic homologous recombination between LINE-1s in a Japanese patient with Perlman syndrome. ***European Journal of Human Genetics.*** in press 2013 Mar 13. doi: 10.1038/ejhg.2013.45. [Epub ahead of print]
46. Miyake N, Yano S, Sakai C, Hatakeyama H, Matsushima Y, Shiina M, Watanabe Y, Bartley J, Abdenur JE, Wang RY, Chang R, Tsurusaki Y, Doi H, Nakashima M, Saitsu H, Ogata K, Goto YI, Matsumoto N. Source. Mitochondrial Complex III Deficiency Caused by a Homozygous UQCRC2 Mutation Presenting with Neonatal-Onset Recurrent Metabolic Decompensation. ***Hum Mutat.*** 34(3): 446-452. 2013, Mar.
47. Okada I, Hamanoue H., Terada K, Tohma T, Megarbane A, Chouery E, Abou-Ghoch J, Jalkh N, Cogulu O, Ozkinay F, Horie K, Takeda J, Furuichi T, Ikegawa S, Nishiyama K, Miyatake S, Nishimura A, Mizuguchi T, Niikawa N, Hirahara F, Kaname T, Yoshiura K, Tsurusaki Y, Doi H, Noriko Miyake N, Furukawa T, Matsumoto N, and Saitsu H. SMOC1 Is Essential for Ocular and Limb Development in Humans and Mice. ***Am J Hum Genet*** Jan;88(1): 1-12, 2011.
48. Amani D, Ravangard F, Niikawa N, Yoshiura KI, Karimzadeh M, Dehaghani AS, Ghaderi A. Coding region polymorphisms in the indoleamine 2,3-dioxygenase (INDO) gene and recurrent spontaneous abortion. ***J Reprod Immunol*** Jan;88(1): 42-47, 2011.
49. Ono S, Yoshiura K, Kurotaki N, Kikuchi T, Niikawa N, Kinoshita A. Mutation and Copy Number Analysis in Paroxysmal Kinesigenic Dyskinesia Families. ***Movement Disorders*** Mar; 26(4):762-764.
50. Miura K, Higashijima A, Shimada T, Miura S, Yamasaki K, Abe S, Jo O, Kinoshita A, Yoshida A, Yoshimura S, Niikawa N, Yoshiura K, Masuzaki H. Clinical application of fetal sex determination using cell-free fetal DNA in pregnant carriers of X-linked genetic disorders. ***J Hum Genet*** 2011 Apr;56(4):296-299.
51. Yamasaki K, Miura K, Shimada T, Miura S, Abe S, Murakami M, Sameshima T, Fujishita A, Kotera K, Kinoshita A, Yoshiura K, Masuzaki H. Epidemiology of human papillomavirus genotypes in pregnant Japanese women. ***J Hum Genet*** 2011 Apr;56(4):313-315.
52. Kurotaki N, Tasaki S, Mishima H, Ono S, Imamura A, Kikuchi T, Nishida N, Tokunaga K, Yoshiura K, Hiroki Ozawa H. Identification of Novel Schizophrenia Loci by Homozygosity Mapping Using DNA Microarray Analysis. ***PLos One*** May 2011;6(5):e20589
53. Oikawa M, Nagayasu T, Yano H, Hayashi T, Abe K, Kinoshita A, Yoshiura KI. Intracystic Papillary Carcinoma of Breast Harbors Significant Genomic Alteration Compared with Intracystic Papilloma: Genome-wide Copy Number and LOH Analysis Using High-Density Single-Nucleotide Polymorphism Microarrays. ***Breast J*** 2011 Jul-Aug;17(4):427-430.
54. Hannibal MC, Buckingham KJ, Ng SB, Ming JE, Beck AE, McMillin MJ, Gildersleeve HI, Bigham AW, Tabor HK, Mefford HC, Cook J, Yoshiura K, Matsumoto T, Matsumoto N, Miyake N, Tonoki H, Naritomi K, Kaname T, Nagai T, Ohashi H, Kurosawa K, Hou JW, Ohta T, Liang D, Sudo A, Morris CA, Banka S, Black GC, Clayton-Smith J, Nickerson DA, Zackai EH, Shaikh TH, Donnai D,

Niikawa N, Shendure J, Bamshad MJ. Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. ***Am J Med Genet A.*** 2011 155A(7):1511-1516.

55. Arima K, Kinoshita A, Mishima H, Kanazawa N, KanekoT, Mizushima T, Ichinose K, Nakamura H, Tsujino A, Kawakami A, Matsunaka M, Kasagi S, Kawano S, Kumagai S, Ohmura K, Mimori T, Hirano M, Ueno S, Tanaka K, Tanaka M, Toyoshima I, Sugino H, Yamakawa A, Tanaka K, Niikawa N, Furukawa F, Shigeo Murata S, Eguchi K, Ida H, Yoshiura K. An assembly defect due to a *PSMB8* mutation reduces proteasome activity and causes autoinflammatory disorder, Nakajo-Nishimura syndrome. *Proc Natl Acad Sci* 2011 Sep6;108(36):14914-14919.
56. Mishima H, Sasaki K, Tanaka M, Tatebe O, Yoshiura KI. Agile parallel bioinformatics workflow management using Pwrake. *BMC Res Notes* 2011 Sep 8;4(1):331.
57. Yamasaki K, Miura K, Shimada T, Ikemoto R, Miura S, Murakami M, Sameshima T, Fujishita A, Kotera K, Kinoshita A, Yoshiura KI, Masuzaki H.Pre-vaccination epidemiology of human papillomavirus infections in Japanese women with abnormal cytology. ***J Obstet Gynaecol Res*** 2011
58. Ono S, Tanaka T, Ishida M, Kinoshita A, Fukuoka J, Takaki M, Sakamoto N, Ishimatsu Y, Kohno S, Hayashi T, Senba M, Yasunami M, Kubo Y, Yoshida LM, Kubo H, Ariyoshi K, Yoshiura K, Morimoto K. Surfactant protein C G100S mutation causes familial pulmonary fibrosis in Japanese kindred. *Eur Respir J* 38(4): 861-869. 2011.
59. Sasaki K, Okamoto N, Kosaki K, Yorifuji T, Shimokawa O, Mishima H, Yoshiura K-i, Harada N. Maternal uniparental isodisomy and heterodisomy on chromosome 6 encompassing a *CUL7* gene mutation causing 3M syndrome. *Clin Genet* 80: 478-483. 2011.
60. Oikawa M, Yoshiura KI, Kondo H, Miura S, Nagayasu T, Nakashima M. Significance of Genomic Instability in Breast Cancer in Atomic Bomb Survivors: Analysis of Microarray-Comparative Genomic Hybridization. ***Radiat Oncol*** 2011 6(1):168.
61. Hikida M, Tsuda M, Watanabe A, Kinoshita A, Akita S, Hirano A, Uchiyama T, Yoshiura KI. No evidence of association between 8q24 and susceptibility to nonsyndromic cleft lip with or without palate in Japanese population. ***Cleft Palate Craniofac J*** 2011
62. Kaname T. A commentary on Implication of gene copy number variation in health and diseases. ***J Hum Genet***, 2012. 57:79-80.
63. Yoneda Y, Saitsu H, Touyama M, Makita Y, Miyamoto A, Hamada K, Kurotaki N, Tomita H, Nishiyama K, Tsurusaki Y, Doi H, Miyake N, Ogata K, Naritomi K, Matsumoto N. Missense mutations in the DNA-binding/dimerization domain of NFIX cause Sotos-like features. ***J Hum Genet***, 2012. 57:207-211.
64. Hatin WI, Nur-Shafawati AR, Zahri MK, Xu S, Jin L, Tan SG, Rizman-Idid M, Zilfalil BA; HUGO Pan-Asian SNP Consortium. Population genetic structure of peninsular Malaysia Malay sub-ethnic groups. ***PLoS One***, 2011. 6:e18312.
65. Koga T, Okada A, Kawashiri S, Kita J, Suzuki T, Nakashima Y, Tamai M, Satoh K, Origuchi T, Iwamoto N, Yamasaki S, Nakamura H, Migita K, Ida H, Ueki Y, Eguchi K, Kawakami A. Soluble urokinase plasminogen activator receptor as a useful biomarker to predict the response to adalimumab in patients with rheumatoid arthritis in a Japanese population. ***Clin Exp Rheumatol.*** 2011. 29: 811-815.
66. Takei S, Hoshino T, Matsunaga K, Sakazaki Y, Sawada M, Oda H, Takenaka SI, Imaoka H, Kinoshita T, Honda S, Ida H, Fukuda TA, Aizawa H. Soluble interleukin-18 receptor complex is a novel biomarker in rheumatoid arthritis. ***Arthritis Res Ther.*** 2011 13(R52)
67. To H, Yoshimatsu H, Tomonari M, Ida H, Tsurumoto T, Tsuji Y, Sonemoto E, Shimasaki N, Koyanagi S, Sasaki H, Ieiri I, Higuchi S, Kawakami A, Ueki Y, Eguchi K. Methotrexate chronotherapy is effective against rheumatoid arthritis. ***Chronobiol Int.*** 2011 28:267-274.
68. Komano Y, Tanaka M, Nanki T, Koike R, Sakai R, Kameda H, Nakajima A, Saito K, Takeno M, Atsumi T, Tohma S, Ito S, Tamura N, Fujii T, Sawada T, Ida H, Hashiramoto A, Koike T, Ishigatsubo Y, Eguchi K, Tanaka Y, Takeuchi T, Miyasaka N, Harigai M. Incidence and risk factors for serious infection in patients

with rheumatoid arthritis treated with tumor necrosis factor inhibitors: a report from the registry of Japanese rheumatoid arthritis patients for longterm safety. J Rheumatol. 2011 38: 1258-1264.
69. Origuchi T, Iwamoto N, Kawashiri SY, Fujikawa K, Aramaki T, Tamai M, Arima K, Nakamura H, Yamasaki S, Ida H, Kawakami A, Ueki Y, Matsuoka N, Nakashima M, Mizokami A, Kawabe Y, Mine M, Fukuda T, Eguchi K. Reduction in serum levels of substance P in patients with rheumatoid arthritis by etanercept, a tumor necrosis factor inhibitor. ***Mod Rheumatol.*** 2011. 21: 244-250.
70. Kawashiri SY, Kawakami A, Iwamoto N, Fujikawa K, Satoh K, Tamai M, Nakamura H, Okada A, Koga T, Yamasaki S, Ida H, Origuchi T, Eguchi K. The power Doppler ultrasonography score from 24 synovial sites or 6 simplified synovial sites, including the metacarpophalangeal joints, reflects the clinical disease activity and level of serum biomarkers in patients with rheumatoid arthritis. Rheumatology (Oxford). 2011. 50: 962-965.
71. Hida A, Akahoshi M, Takagi Y, Imaizumi M, Sera N, Soda M, Maeda R, Nakashima E, Ida H, Kawakami A, Nakamura T, Eguchi K. Lipid infiltration in the parotid glands: a clinical manifestation of metabolic syndrome. ***Exp Clin Endocrinol Diabetes.*** 2012. 120: 110-115.
72. Ichinose K, Origuchi T, Kawashiri SY, Iwamoto N, Fujikawa K, Aramaki T, Kamachi M, Arima K, Tamai M, Nakamura H, Ida H, Kawakami A, Eguchi K. Long-term follow-up of adalimumab monotherapy for rheumatoid arthritis in Japanese patients: a report of six cases. ***Rheumatol Int.*** 2012 32: 483-487.
73. Kawashiri SY, Kawakami A, Okada A, Koga T, Tamai M, Yamasaki S, Nakamura H, Origuchi T, Ida H, Eguchi K. CD4+CD25highCD127low/- Treg cell frequency from peripheral blood correlates with disease activity in patients with rheumatoid arthritis. ***J Rheumatol.*** 2011 38: 2517-2521.
74. Kita J, Tamai M, Arima K, Nakashima Y, Suzuki T, Kawashiri SY, Iwamoto N, Okada A, Koga T, Yamasaki S, Nakamura H, Origuchi T, Ida H, Aoyagi K, Uetani M, Eguchi K, Kawakami A. Treatment discontinuation in patients with very early rheumatoid arthritis in sustained simplified disease activity index remission after synthetic disease-modifying anti-rheumatic drug administration. ***Mod Rheumatol.*** 2012 22(3): 346-352.
75. Kita J, Tamai M, Arima K, Nakashima Y, Suzuki T, Kawashiri SY, Okada A, Koga T, Yamasaki S, Nakamura H, Origuchi T, Aramaki T, Nakashima M, Fujikawa K, Tsukada T, Ida H, Aoyagi K, Uetani M, Eguchi K, Kawakami A. Delayed treatment with tumor necrosis factor inhibitors in incomplete responders to synthetic disease-modifying anti-rheumatic drugs shows an excellent effect in patients with very early rheumatoid arthritis with poor prognosis factors. ***Mod Rheumatol.*** 2012 22(2): 195-201.
76. Tamai M, Kawakami A, Uetani M, Fukushima A, Arima K, Fujikawa K, Iwamoto N, Aramaki T, Kamachi M, Nakamura H, Ida H, Origuchi T, Aoyagi K, Eguchi K. Magnetic resonance imaging (MRI) detection of synovitis and bone lesions of the wrists and finger joints in early-stage rheumatoid arthritis: comparison of the accuracy of plain MRI-based findings and gadolinium-diethylenetriamine pentaacetic acid-enhanced MRI-based findings. ***Mod Rheumatol.*** 2011. 29(6):1062-1063
77. Iwamoto N, Kawakami A, Tamai M, Arima K, Nakamura H, Kawashiri SY, Kita J, Okada A, Koga T, Kamachi M, Yamasaki S, Ichinose K, Ida H, Origuchi T, Uetani M, Eguchi K. Magnetic resonance imaging of wrist and finger joints distinguishes secondary Sjögren's syndrome with rheumatoid arthritis from primary Sjögren's syndrome with articular manifestations. ***Clin Exp Rheumatol.*** 2011 29:1062-1063.
78. Watanabe Y, Yano S, Niihori T, Aoki Y, Matsubara Y, Yoshino M, Matsuishi T. A familial case of LEOPARD syndrome associated with a high-functioning autism spectrum disorder. ***Brain Dev.*** 2011 33: 576-579.
79. Segawa Y, Matsufuji M, Itokazu N, Utsunomiya H, Watanabe Y, Yoshino M, Takashima S. A long-term survival case of arginase deficiency with severe multicystic white matter and compound mutations. ***Brain Dev.*** 2011 33: 45-48.

学会発表

1. **American Society of Human Genetics, 63rd Annual Meeting, Boston Convention Center & Exhibition Center, October 22-26, 2013, Boston.** An infantile case of hepatomegaly, lactic acidosis, hypoglycemia, ketosis, and hyperlipidemia of unknown etiology. Watanabe Y, Seki Y, Yanagi T, Mizouchi T, Takeuchi T, Iwamoto J, YoshinoM, Watanabe S, Inokuchi T, Yano S, Yoshiura K, Matsuishi T.
2. **IEIIS 2012 Homeostatic Inflammation Symposium 2012.10.23-26 Tokyo**
   Hiroaki Ida, Shinjiro Kaieda, Seiyo Honda, Kazuhiko Arima, Koh-ichiro Yoshiura, Nobuo Kanazawa, Takaaki Fukuda
   Proteasome disability syndrome: a novel autoinflammatory syndrome
3. **The 55th Annual Meeting of Jte Japanese Society for Inherited Metabolic Disease (JSIMD), The 3rd Asian Congress for Inherited Metabolic Diseases (ACIMD) 2013.11.27-29 (Urayasu, Chiba)**
   Watanabe Y, Seki Y, Yanagi T, Mizuochi T, Takeuchi T, Iwamoto J, Yoshino M, Watanabe S, Inokuchi T, Yano S, Yoshiura K, Matsuishi T. : An infantile case of hepatomegaly, lactic acidosis, hypoglycemia, ketosis, and hyperlipidemia of unknown etiology.
4. **The 12th International Congress of Inborn Errors of Metabolism 2013.9.3-9.6 (Barcelona)**
   Yano S, Miyake N, Watanabe Y, Bartley J, Abdenur JE, Wang RY, Chang R, Goto Y, Shiina M, Ogata K, Matsumoto N: Mitochondrial ubiquinol-cytochrome c reductase core protein II defects may affect multiple metabolic pathways.
5. **ESHG2012 (EUROPEAN Human Genetics CONFERENCE 2012), 6 月 23 日-26 日, Nürnberg, Germany**
   P12.131: Tadashi Kaname, Kumiko Yanagi, Yukako Muramatsu, Takaya Tohma, Hiroaki Hanafusa, Konomi Morita, Shinya Ikematsu, Yusuke Itagaki, Hiroko Taniai, Kenji Kurosawa, Seiji Mizuno, Koichiro Yoshiura, Kenji Naritomi. A mutation detected by exome sequencing and phenotypic variability in a family with Lenz microphthalmia syndrome.
   P02.150: Ganaha A., Kaname T., Yanagi K., Narutomi K., Usami S., Suzuki M. Distinct pathogenic substitution of IVS15+5G＞A in the SLC26A4 gene in patients with enlarged vestibular aqueduct and Pendred syndrome in Okinawa islands.
6. **Annual Symposium of the Society for the Study of Inborn Errors of Metabolism 2012.8.30-9.2 (Birmingham)**
   Watanabe Y, Tashiro K, Aoki K, Seki Y, Yanagi T, Okada J, Mizuochi T, Inokuchi T, Yoshino M, Matsuishi T. Two cases of neonatal onset type II citrullinemia diagnosed by urine organic acids based newborn screening.
7. **The American Society of Human Genetics, 62nd Annual Meeting, San Francisco, CA, USA, November 6-10, 2012.**
   2758W: Kaname T., Yanagi K., Okamoto N., Kurosawa K., Izumikawa Y., Fukushima Y., Makita Y., Tsukahara M., Altincik A., Mizuno S., Naritomi K. Novel mutations of the FGD1 gene in 16 patients with Aarskog-Scott syndrome.
   724T: Watanabe Y, Tashiro K, Aoki K, Inokuchi T, Seki Y, Yanagi T, Mizuochi T, Yoshino M, Matsuishi T. Two cases of neonatal onset type II citrullinemia diagnosed by urine organic acids based newborn screening.
8. **10th Korean PWS Symposium, Seoul, Korea, September 18, 2012.**
   (invited): Kaname T. A practical approach to genetic diseases by next-generation sequencing technologies. :
9. **The 13th Annual Bioinformatics Open Source Conference, 2012年7月13-14日, Long Beach Convention Center, Long Beach, CA, USA.**
   Hiroyuki Mishima, Raoul J.P. Bonnal, Naohisa Goto, Francesco Strozzi, Toshiaki Katayama, Pjotr Prins: Biogem, Ruby UCSC API, and Bioruby. (口演)
   Hiroyuki Mishima, Raoul J.P. Bonnal, Naohisa Goto, Francesco Strozzi, Toshiaki Katayama, Pjotr Prins: Biogem, Ruby UCSC API, and Bioruby. (ポスター)
10. **20th Annual International Conference on Intelligent Systems for Molecular Biology, 2012年7月15-17日, Long Beach Convention Center, Long Beach, CA, USA.**
    (06) Hiroyuki Mishima, Jan Aerts, Toshiaki Katayama, Raoul J.P. Bonnal, Koh-ichiro Yoshiura: The Ruby UCSC API:

accessing the UCSC Genome Database using Ruby. (ポスター)

11. **IEIIS 2012 Homeostatic Inflammation Symposium 2012.10.23-26 Tokyo**
Ida H, Kaieda S, Honda S, Arima K, Yoshiura K, Kanazawa N, Fukuda T. Proteasome disability syndrome: a novel autoinflammatory syndrome

12. **Annual Symposium of the Society for the Study of Inborn Errors of Metabolism 2011.8.30-9.2 (Geneva)**
Long-term survival and restoration of GLCNAC-1-phosphotransferase activity in peripheral lymphocytes of a patient with I-cell disease who received allogenic bone marrow transplantation. Yoshino M, Watanabe Y, Yabe H, Kato S, Otomo T, Sakai N, Gasa S, Hayasaka-Sukegawa K.

13. **12th Unternational Congress of Human Genetics and The American Society of Human Genetics, 61st Annual Meeting, Montoreal, Canada, October 11-15, 2011.**
556T: Identification of novel schizophrenia loci by homozygosity mapping using DNA microarrayanalysos. N. Kurotaki, H. Mishima, S. Ono, A. Imamura, S. Tasaki, T. Kikuchi, N. Nishida, K. Tokunaga, K. Yoshiura, H. Ozawa.
1036/F: Ehlers-Danlos syndrome and bilateral periventricular heterotopia due to the novel mutation of p.Gly132Val in the CHD1 in FLNA: An infantile case presented with congenital tracheobronchopulmonary disease. Watanabe Y, Okada J, Kimura K, Okamatsu Y, Oya T, Yoshino M, Yagi H, Sato M.
1095T: Detection of a mutation in Lents micropthalmia family by exome sequencing. T. Kaname, K. Yanagi, Y. Muramatsu, T. Tohma, H. Hanafusa, K. Morita, S. Ikematsu, Y. Itagaki, S. Mizuno, K. Yoshiura, K. Naritomi.

14. **10th World Congress on Inflammation 2011.06.25-29 Paris.**
Ida H, Arima K, Kinoshita A, Mishima H, Kanazawa N, Furukawa F, Murata S, Yoshiura K-I, Eguchi K A novel mutation of proteasome subunit causes decrease of proteasome activity in Nakajo-Nishimura syndrome (familial Japanese fever).
Kanazawa N, Kunimoto K, Mikita N, Furukawa F, Yoshiura K-I, Ida H Nakajo-Nishimura syndrome (familial Japanese fever) and related autoinflammatory disorders accompanied with lipodystrophy.

15. **Annual European Congress of Rheumatology (EULAR 2011) 2011.5.25-28 London.**
Arima K, Kanazawa N, Mishima H, Kinoshita A, Ida H, Yoshiura K, Eguchi K Decrease of proteasome activity is associated with a novel mutation of the proteasome catalytic subunit in an autoinflammatory disorder, Nakajo-Nishimura syndrome.

16. **75th annual meeting of the American College of Rheumatology(ACR 2011) 2011.11.5-9 Chicago.**
Ida H, Arima K, Kanazawa N, Yoshiura KI Proteasome disability syndrome : an analysis of the pathogenesis of a new autoinflammatory syndrome, Nakajo-Nishimura syndrome.

H. 知的所有権の出願・取得状況

1 特許取得

日本国特許出願 特願2011-71082 筆頭発明者:荻朋男 発明の名称:日焼けの原因遺伝子 出願人：長崎大学 出願日:2011年3月28日

「自己炎症疾患又は自己免疫疾患関連遺伝子及びその利用」発明者(長崎大学：吉浦孝一郎，久留米大学：井田弘明，和歌山県立医科大学：金澤伸雄) 出願番号：特願 2011-177269

2 実用新案登録
なし

3 その他
なし

II. 研究成果の刊行に関する一覧表

平成２３～２５年度研究成果の刊行に関する一覧表

書籍

Kaname T, Yanagi K, Maehara H. Osteosarcoma and midkine. '***Midkine: From embryogenesis to pathogene sis and medication***' Ergüven M, Billir A, Muramatsu T. Eds. Springer, New York, pp313-319, 2012.

雑誌

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Kaname T, Yanagi K, Naritomi K.	A ommentary on the promise of whole-exome sequencing in medical genetics.	*J Hum Genet*	In press		2014
Sasaki K, Mishima H, Miura K, Yoshiura KI.	Uniparental disomy analysis in trios using genome-wide SNP array and whole-genome sequencing data imply segmental uniparental isodisomy in general populations.	*Gene*	512(2)	267-274	2013
Yamada A, Ishikawa T, Ota I, Kimura M, Shimizu D, Tanabe M, Chishima T, Sasaki T, Ichikawa Y, Morita S, Yoshiura KI, Takabe K, Endo I.	High expression of ATP-binding cassette transporter ABCC11 in breast tumors is associated with aggressive subtypes and low disease-free survival.	*Breast Cancer Res Treat*	137(3)	773-782	2013
Ishikawa T, Toyoda Y, Yoshiura K, Niikawa N.	Pharmacogenetics of human ABC transporter ABCC11: new insights into apocrine gland growth and metabolite secretion.	*Front Genet*	3	306	2013
Higashijima A, Miura K, Mishima H, Kinoshita A, Jo O, Abe S, Hasegawa Y, Miura S, Yamasaki K, Yoshida A, Yoshiura K, Masuzaki H.	Characterization of placenta-specific microRNAs in fetal growth restriction pregnancy.	*Prenat Diagn*	33(3)	214-222	2013
Higashimoto K, Maeda T, Okada J, Ohtsuka Y, Sasaki K, Hirose A, Nomiyama M, Takayanagi T, Fukuzawa R, Yatsuki H, Koide K, Nishioka K, Joh K, Watanabe Y, Yoshiura KI, Soejima H.	Homozygous deletion of DIS3L2 exon 9 due to non-allelic homologous recombination between LINE-1s in a Japanese patient with Perlman syndrome.	*Eur J Hum Genet*	21(11)	1316-1319	2013

Abe S, Miura K, Kinoshita A, Mishima H, Miura S, Yamasaki K, Hasegawa Y, Higashijima A, Jo O, Sasa K, Yoshida A, Yoshiura K, Masuzaki H.	Copy number variation of the antimicrobial-gene, defensin beta 4, is associated with susceptibility to cervical cancer.	*J Hum Genet*	58(5)	250-253	2013
Hamaguchi D, Miura K, Abe S, Kinoshita A, Miura S, Yamasaki K, Yoshiura KI, Masuzaki H.	Initial viral load in cases of single human papillomavirus 16 or 52 persistent infection is associated with progression of later cytopathological findings in the uterine cervix.	*J Med Virol*	85(12)	2093-2100	2013
Nakao K, Oikawa M, Arai J, Mussazhanova Z, Kondo H, Shichijo K, Nakashima M, Hayashi T, Yoshiura K, Hatachi T, Nagayasu T.	A Predictive Factor of the Quality of Microarray Comparative Genomic Hybridization Analysis for Formalin-fixed Paraffin-embedded Archival Tissue.	*Diagn Mol Pathol*	22(3)	174-180	2013
Hasegawa Y, Miura K, Furuya K, Yoshiura K, Masuzaki H.	Identification of Complete Hydatidiform Mole Pregnancy-Associated MicroRNAs in Plasma.	*Clin Chem*	59(9)	1410-1412	2013
Miyake N, Koshimizu E, Okamoto N, Mizuno S, Ogata T, Nagai T, Kosho T, Ohashi H, Kato M, Sasaki G, Mabe H, Watanabe Y, Yoshino M, Matsuishi T, Takanashi J, Shotelersuk V, Tekin M, Ochi N, Kubota M, Ito N, Ihara K, Hara T, Tonoki H, Ohta T, Saito K, Matsuo M, Urano M, Enokizono T, Sato A, Tanaka H, Ogawa A, Fujita T, Hiraki Y, Kitanaka S, Matsubara Y, Makita T, Taguri M, Nakashima M, Tsurusaki Y, Saitsu H, Yoshiura K, Matsumoto N, Niikawa N.	MLL2 and KDM6A mutations in patients with Kabuki syndrome.	*Am J Med Genet A.*	161(9)	2234-2243	2013

Kashiyama K, Nakazawa Y, Pilz D, Guo C, Shimada M, Sasaki K, Fawcett H, Wing J, Lewin S, Carr L, Yoshiura K, Utani A, Hirano A, Yamashita S, Greenblatt D, Nardo T, Stefanini M, McGibbon D, Sarkany R, Fassihi H, Takahashi Y, Nagayama Y, Mitsutake N, Lehmann AR, and Ogi T.	Malfunction of Nuclease ERCC1-XPF Results in Diverse Clinical Manifestations and Causes Cockayne Syndrome, Xeroderma Pigmentosum, and Fanconi Anemia.	*Am J Hum Genet*	92(5)	807-819	2013
Woodbine L, Neal JA, Sasi NK, Shimada M, Deem K, Coleman H, Dobyns WB, Ogi T, Meek K, Davies EG, Jeggo PA.	PRKDC mutations in a SCID patient with profound neurological abnormalities.	*The Journal of Clinical Investigation*	123(7)	2969-2980	2013
Matsuse M, Mitsutake N, Tanimura S, Ogi T, Nishihara E, Hirokawa M, Fuziwara CS, Saenko VA, Suzuki K, Miyauchi A, Yamashita S.	Functional characterization of the novel BRAF complex mutation, BRAF (V600delinsYM), identified in papillary thyroid carcinoma.	*International Journal of Cancer*	132(3)	738-743	2013
Suzumori N, Kaname T, Muramatsu Y, Yanagi K, Kumagai K, Mizuno S, Naritomi K, Saitho S, Sugiura M.	Prenatal diagnosis of X-linked recessive Lenz microphthalmia syndrome.	*J Obstetr Gyaecol Res*,	39(11)	1545-1547	2013
Ganaha A, Kaname T, Yanagi K, Naritomi K, Tono T, Usami S, Suzuki M.	Pathogenic substitution of IVS15 + 5G > A in SLC26A4 in patients of Okinawa Islands with enlarged vestibular aqueduct syndrome or Pendred syndrome.	*BMC Med Genet*	14	56	2013
Altıncık A, Kaname T, Demir K, Böber E.	A novel mutation in a mother and a son with Aarskog-Scott syndrome.	*J Pediatr Endocrinol Metab*,	26(3-4)	385-388	2013
Kosho T, Okamoto N, Ohashi H, Tsurusaki Y, Imai Y, Hibi-Ko Y, Kawame H, Homma T, Tanabe S, Kato M, Hiraki Y, Yamagata T, Yano S, Sakazume S, Ishii T, Nagai T, Ohta T, Niikawa N, Mizuno S, Kaname T, Naritomi K, Narumi Y, Wakui K, Fukushima Y, Miyatake S, Mizuguchi T, Saitsu H, Miyake N, Matsumoto N.	Clinical correlations of mutations affecting six components of the SWI/SNF complex: detailed description of 21 patients and a review of the literature.	*Am J Med Genet A*,	161A(6)	1221-1237	2013

Migita K, Agematsu K, Masumoto J, Ida H, Honda S, Jiuchi Y, Izumi Y, Maeda Y, Uehara R, Nakamura Y, Koga T, Kawakami A, Nakashima M, Fujieda Y, Nonaka F, Eguchi K, Furukawa H, Nakamura T,Nakamura M, Yasunami M.	The contribution of SAA1 polymorphisms to Familial Mediterranean fever susceptibility in the Japanese population.	*PLoS One*	8(2)	e55227	2013
Ihara K, Yoshino M, Hoshina T, Harada N, Kojima-Ishii K, Makimura M, Hasegawa Y, Watanabe Y, Yamaguchi S, Hara T.	Coagulopathy in patients with late-onset ornithine transcarbamylase deficiency in remission state: a previously unrecognized complication.	*Pediatrics,*	131(1)	e327-330	2013
Miyake N, Yano S, Sakai C, Hatakeyama H, Matsushima Y, Shiina M, Watanabe Y, Bartley J, Abdenur JE, Wang RY, Chang R, Tsurusaki Y, Doi H, Nakashima M, Saitsu H, Ogata K, Goto Y, Matsumoto N.	Mitochondrial complex III deficiency caused by a homozygous UQCRC2 mutation presenting with neonatal-onset recurrent metabolic decompensation.	*Hum Mutat*	34(3)	446-452	2013
Okano Y, Kobayashi K, Ihara K, Ito T, Yoshino M, Watanabe Y, Kaji S, Ohura T, Nagao M, Noguchi A, Mushiake S, Hohashi N, Hashimoto-Tamaoki T.	Fatigue and quality of life in citrin deficiency during adaptation and compensation stage.	*Mol Genet Metab*	109(1)	9-13	2013
Nakashima S, Watanabe Y, Okada J, Ono H, Nagata E, Fukami M, Ogata T.	Critical role of Yp inversion in PRKX/PRKY-mediated Xp;Yp translocation in a patient with 45,X testicular disorder of sex development.	*Endocr J.*	60(12)	1329-1334	2013
Ikewaki N, Sonoda T, Inoko K.	Unique properties of cluster of differentiation 93 in the umbilical cord blood of neonates.	*Microbiol Immunol*	57(12)	822-832	2013
Ogi T, Nakazawa Y, Sasaki K, Guo C, Yoshiura K, Utani A, Nagayama Y.	Molecular cloning and characterisation of UVSSA, the responsible gene for UV-sensitive syndrome.	Journal of Biochemistry (Tokyo).	85	133-144	2013
吉浦孝一郎	遺伝性疾患におけるエクソーム解析の有用性と近将来.	医学のあゆみ	245(5)	363-368	2013
三嶋博之	全エクソーム解析における情報処理.	医学のあゆみ	245(5)	345-351	2013

要　匡	常染色体優性遺伝性疾患のエクソーム解析.	医学のあゆみ	245(5)	381-385	2013
黒滝直弘，小野慎二，小澤寛樹，吉浦孝一郎	発作性運動誘発性舞踏アテトーゼの分子メカニズム.	神経内科	79(6)	718-725	2013
右田清志、野中文陽、和泉泰衛、江口勝美、中村正、井田弘明、上松一永	家族性地中海熱の臨床	炎症と免疫	21	40-46	2013
川上純、右田清志、井田弘明	自己炎症疾患	medicina	50	458-462	2013
井田弘明	遺伝性発熱性疾患の遺伝子診断ガイドライン	リウマチ科	50	507-511	2013
井田弘明	自己炎症症候群	最新医学社	68	2561-2569	2013
Nakazawa Y, Sasaki K, Mitsutake N, Matsuse M, Shimada M, Nardo T, Takahashi Y, Ohyama K, Ito K, Mishima H, Nomura M, Kinoshita A, Ono S, Takenaka K, Masuyama R, Kudo T, Slor H, Utani A, Tateishi S, Yamashita S, Stefanini M, Lehmann AR, Yoshiura KI, Ogi T.	Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase IIo processing in transcription-coupled nucleotide-excision repair.	*Nature Genetics*	44(5)	586-592	2012
Matsuse M, Sasaki K, Nishihara E, Minami S, Hayashida C, Kondo H, Suzuki K, Saenko V, Yoshiura K, Mitsutake N, Yamashita S.	Copy number alteration and uniparental disomy analysis categorizes Japanese papillary thyroid carcinomas into distinct groups.	*PLoS One*	74(4)	e36063	2012
Arai J, Tsuchiya T, Oikawa M, Mochinaga K, Hayashi T, Yoshiura KI, Tsukamoto K, Yamasaki N, Matsumoto K, Miyazaki T, Nagayasu T.	Clinical and molecular analysis of synchronous double lung cancers.	*Lung Cancer*	77(2):	281-287	2012
Ono S, Yoshiura K, Kinoshita A, Kikuchi T, Nakane Y, Kato N, Sadamatsu M, Konishi T, Nagamitsu S, Matsuura M, Yasuda A, Komine M, Kanai K, Inoue T, Osamura T, Saito K, Hirose S, Koide H, Tomita H, Ozawa H, Niikawa N, Kurotaki N.	Mutations in PRRT2 responsible for paroxysmal kinesigenic dyskinesias also cause benign familial infantile convulsions.	*J Hum Genet*	57(5):	338-341	2012

Mishima H, Aerts J, Katayama T, Bonnal RJ, Yoshiura K.	The Ruby UCSC API: accessing the UCSC genome database using Ruby.	*BMC Bioinformatics*	13	240	2012
Hikida M, Tsuda M, Watanabe A, Kinoshita A, Akita S, Hirano A, Uchiyama T, Yoshiura K.	No evidence of association between 8q24 and susceptibility to nonsyndromic cleft lip with or without palate in Japanese population.	*Cleft Palate Craniofac J*	49(6)	714-717	2012
Kawakami A, Migita K, Ida H, Yoshiura K, Arima K, Eguchi K.	[109th Scientific Meeting of the Japanese Society of Internal Medicine: educational lecture: 14. Autoinflammatory syndrome].	*Nihon Naika Gakkai Zasshi*	101(9)	2733-2739,	2012
Ogi T, Walker S, Stiff T, Hobson E, Limsirichaikul S, Carpenter G, Prescott K, Suri M, Byrd P, Matsuse M, Mitsutake N, Nakazawa Y, Vasudevan P, Barrow M, Stewart G, Taylor M, O'Driscoll M, and Jeggo P.	Identification of the first ATRIP-deficient patient and novel mutations in ATR define a clinical spectrum for ATR-ATRIP Seckel syndrome.	*PLoS Genetics*	8	e1002945	2012
Jinam T, Nishida N, Hirai M, Kawamura S, OOta H, Umetsu K, Kimura R, Ohashi J, Tajima A, Yamamoto T, Tanabe H, Mano S, Suto Y, Kaname T, Naritomi K, Yanagi K, Niikawa N, Omoto K, Tokunaga K, Saitou N.	The history of human populations in the Japanese Archipelago inferred from genomewide SNP data with a special reference to the Ainu and the Ryukyuan populations.	*J Hum Genet*, 57: 787-795, 2012.	57(12)	787-795	2012
Yanagi K, Kaname T, Wakui K, Hashimoto O, Fukushima Y, Naritomi K.	Identification of four novel synonymous substitutions in the X-linked genes neuroligin 3 and neuroligin 4X in Japanese patients with autistic spectrum disorder.	*Autism Res Treat*	2012	724072	2012
Cheung W, Kotzamanis G, Abdulrazzak H, Goussard S, Kaname T, Kotsinas A, Gorgoulis VG, Grillot-Courvalin C, Huxley C.	Bacterial delivery of large intact genomic-DNA-containing BACs into mammalian cells.	*Bioeng Bugs*	3(2)	86-92	2012
He Y, Wang WR, Xu S, Jin L; Pan-Asia SNP Consortium.	Paleolithic Contingent in Modern Japanese: Estimation and Inference using Genome-wide Data.	*Sci Rep*	2	355	2012

Tsurusaki Y, Okamoto N, Ohashi H, Kosho T, Imai Y, Hibi-Ko Y, Kaname T, Naritomi K, Kawame H, Wakui K, Fukushima Y, Homma T, Kato M, Hiraki Y, Yamagata T, Yano S, Mizuno S, Sakazume S, Ishii T, Nagai T, Shiina M, Ogata K, Ohta T, Niikawa N, Miyatake S, Okada I, Mizuguchi T, Doi H, Saitsu H, Miyake N, Matsumoto N.	Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome.	*Nat Genet*	44(4)	376-378	2012
Migita K, Uehara R, Nakamura Y, Yasunami M, Tsuchiya-Suzuki A, Yazaki M, Nakamura A, Masumoto J, Yachie A, Furukawa H, Ishibashi H, Ida H, Yamazaki K, Kawakami A, Agematsu K.	Familial Mediterranean Fever in Japan.	*Medicine (Baltimore)*	91(6)	337-343	2012
Migita K, Ida H, Moriuchi H, Agematsu K.	Clinical relevance of MEFV gene mutations in Japanese patients with unexplained fever.	*J Rheumatol*	39(4)	875-877	2012
Higashimoto K, Nakabayashi K, Yatsuki H, Yoshinaga H, Jozaki K, Okada J, Watanabe Y, Aoki A, Shiozaki A, Saito S, Koide K, Mukai T, Hata K, Soejima H.	Aberrant methylation of H19-DMR acquired after implantation was dissimilar in soma versus placenta of patients with Beckwith-Wiedemann syndrome.	*Am J Med Genet A.*	158A(7)	1670-1675	2012
井田弘明，有馬和彦，金澤伸雄，吉浦孝一郎	中條-西村症候群の原因遺伝子とプロテアソーム機能異常	リウマチ科（科学評論社）	47(6)	654-660	2012
井田弘明，有馬和彦，金澤伸雄，吉浦孝一郎	プロテアソーム病	炎症と免疫（先端医学社）	20(6)	609-614	2012
井田弘明，福田孝昭	自己炎症症候群の定義と分類	九州リウマチ	32(2)	75-78	2012
井田弘明，福田孝昭	自己炎症症候群	日本臨床	70(suppl 8)	561-568,	2012
井田弘明	自己炎症症候群	日本医事新報	4615	78-83	2012

Okada I, Hamanoue H., Terada K, Tohma T, Megarbane A, Chouery E, Abou-Ghoch J, Jalkh N, Cogulu O, Ozkinay F, Horie K, Takeda J, Furuichi T, Ikegawa S, Nishiyama K, Miyatake S, Nishimura A, Mizuguchi T, Niikawa N, Hirahara F, Kaname T, Yoshiura K, Tsurusaki Y, Doi H, Noriko Miyake N, Furukawa T, Matsumoto N, and Saitsu H.	SMOC1 Is Essential for Ocular and Limb Development in Humans and Mice.	*American Journal of Human Genetics*	88(1)	1-12	2011
Amani D, Ravangard F, Niikawa N, Yoshiura KI, Karimzadeh M, Dehaghani AS, Ghaderi A.	Coding region polymorphisms in the indoleamine 2,3-dioxygenase (INDO) gene and recurrent spontaneous abortion.	*Jounal of Reproductive Immunology*	88(1)	42-47	2011
Ono S, Yoshiura K, Kurotaki N, Kikuchi T, Niikawa N, Kinoshita A.	Mutation and Copy Number Analysis in Paroxysmal Kinesigenic Dyskinesia Families.	*Movement Disorders*	26(4)	762-764	2011
Miura K, Higashijima A, Shimada T, Miura S, Yamasaki K, Abe S, Jo O, Kinoshita A, Yoshida A, Yoshimura S, Niikawa N, Yoshiura K, Masuzaki H.	Clinical application of fetal sex determination using cell-free fetal DNA in pregnant carriers of X-linked genetic disorders.	*Journal of Human Genetics*	56(4)	296-299	2011
Yamasaki K, Miura K, Shimada T, Miura S, Abe S, Murakami M, Sameshima T, Fujishita A, Kotera K, Kinoshita A, Yoshiura K, Masuzaki H.	Epidemiology of human papillomavirus genotypes in pregnant Japanese women.	*Journal of Human Genetics*	56(4)	313-315	2011
Kurotaki N, Tasaki S, Mishima H, Ono S, Imamura A, Kikuchi T, Nishida N, Tokunaga K, Yoshiura K, Hiroki Ozawa H.	Identification of Novel Schizophrenia Loci by Homozygosity Mapping Using DNA Microarray Analysis.	*PLoS One*	6(5)	e20589	2011
Oikawa M, Nagayasu T, Yano H, Hayashi T, Abe K, Kinoshita A, Yoshiura KI.	Intracystic Papillary Carcinoma of Breast Harbors Significant Genomic Alteration Compared with Intracystic Papilloma: Genome-wide Copy Number and LOH Analysis Using High-Density Single-Nucleotide Polymorphism Microarrays.	*Breast Journal*	17(4)	427-430	2011

Mishima H, Sasaki K, Tanaka M, Tatebe O, Yoshiura KI.	Agile parallel bioinformatics workflow management using Pwrake.	*BMC Res Notes*	4(1)	331	2011
Hannibal MC, Buckingham KJ, Ng SB, Ming JE, Beck AE, McMillin MJ, Gildersleeve HI, Bigham AW, Tabor HK, Mefford HC, Cook J, Yoshiura K, Matsumoto T, Matsumoto N, Miyake N, Tonoki H, Naritomi K, Kaname T, Nagai T, Ohashi H, Kurosawa K, Hou JW, Ohta T, Liang D, Sudo A, Morris CA, Banka S, Black GC, Clayton-Smith J, Nickerson DA, Zackai EH, Shaikh TH, Donnai D, Niikawa N, Shendure J, Bamshad MJ.	Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome.	*American Journal of Medical Genetics, Part A*	155A(7)	1511-1516	2011
Arima K, Kinoshita A, Mishima H, Kanazawa N, KanekoT, Mizushima T, Ichinose K, Nakamura H, Tsujino A, Kawakami A, Matsunaka M, Kasagi S, Kawano S, Kumagai S, Ohmura K, Mimori T, Hirano M, Ueno S, Tanaka K, Tanaka M, Toyoshima I, Sugino H, Yamakawa A, Tanaka K, Niikawa N, Furukawa F, Shigeo Murata S, Eguchi K, Ida H, Yoshiura K.	An assembly defect due to a *PSMB8* mutation reduces proteasome activity and causes autoinflammatory disorder, Nakajo-Nishimura syndrome.	*Procedings National Academy of Sci*ence, USA	108(36)	14914-14919	2011
Yamasaki K, Miura K, Shimada T, Ikemoto R, Miura S, Murakami M, Sameshima T, Fujishita A, Kotera K, Kinoshita A, Yoshiura KI, Masuzaki H.	Pre-vaccination epidemiology of human papillomavirus infections in Japanese women with abnormal cytology.	*Jouna; of Obstetrics and Gynaecological Research*	37(11)	1666-1670.	2011
Ono S, Tanaka T, Ishida M, Kinoshita A, Fukuoka J, Takaki M, Sakamoto N, Ishimatsu Y, Kohno S, Hayashi T, Senba M, Yasunami M, Kubo Y, Yoshida LM, Kubo H, Ariyoshi K, Yoshiura K, Morimoto K.	Surfactant protein C G100S mutation causes familial pulmonary fibrosis in Japanese kindred.	*European Respiratory Journal*	38(4)	861-869	2011

Sasaki K, Okamoto N, Kosaki K, Yorifuji T, Shimokawa O, Mishima H, Yoshiura K-i, Harada N.	Maternal uniparental isodisomy and heterodisomy on chromosome 6 encompassing a *CUL7* gene mutation causing 3M syndrome.	*Clinical Genetics*	80(5)	478-483	2011
Oikawa M, Yoshiura KI, Kondo H, Miura S, Nagayasu T, Nakashima M.	Significance of Genomic Instability in Breast Cancer in Atomic Bomb Survivors: Analysis of Microarray-Comparative Genomic Hybridization.	*Radiation Oncology*	6(1)	168	2011
Sosonkina N, Nakashima M, Ohta T, Niikawa N, Starenki D.	Down-regulation of ABCC11 protein (MRP8) in human breast cancer.	*Expimental Oncology*	33(1)	42-46	2011
Hatin WI, Nur-Shafawati AR, Zahri MK, Xu S, Jin L, Tan SG, Rizman-Idid M, Zilfalil BA; HUGO Pan-Asian SNP Consortium.	Population genetic structure of peninsular Malaysia Malay sub-ethnic groups.	*PLoS One,*	6	e18312	2011
Koga T, Okada A, Kawashiri S, Kita J, Suzuki T, Nakashima Y, Tamai M, Satoh K, Origuchi T, Iwamoto N, Yamasaki S, Nakamura H, Migita K, Ida H, Ueki Y, Eguchi K, Kawakami A.	Soluble urokinase plasminogen activator receptor as a useful biomarker to predict the response to adalimumab in patients with rheumatoid arthritis in a Japanese population.	*Clininal and Experimental Rheumatology*	29(5)	811-815	2011
Takei S, Hoshino T, Matsunaga K, Sakazaki Y, Sawada M, Oda H, Takenaka SI, Imaoka H, Kinoshita T, Honda S, Ida H, Fukuda TA, Aizawa H.	Soluble interleukin-18 receptor complex is a novel biomarker in rheumatoid arthritis.	*Arthritis Research and Therapy*	13(2)	R52	2011
To H, Yoshimatsu H, Tomonari M, Ida H, Tsurumoto T, Tsuji Y, Sonemoto E, Shimasaki N, Koyanagi S, Sasaki H, Ieiri I, Higuchi S, Kawakami A, Ueki Y, Eguchi K.	Methotrexate chronotherapy is effective against rheumatoid arthritis.	*Chronobiol International*	28(3)	267-274	2011

Komano Y, Tanaka M, Nanki T, Koike R, Sakai R, Kameda H, Nakajima A, Saito K, Takeno M, Atsumi T, Tohma S, Ito S, Tamura N, Fujii T, Sawada T, Ida H, Hashiramoto A, Koike T, Ishigatsubo Y, Eguchi K, Tanaka Y, Takeuchi T, Miyasaka N, Harigai M.	Incidence and risk factors for serious infection in patients with rheumatoid arthritis treated with tumor necrosis factor inhibitors: a report from the registry of Japanese rheumatoid arthritis patients for longterm safety.	*Journal of Rheumatology*	38	1258-1264	2011
Origuchi T, Iwamoto N, Kawashiri SY, Fujikawa K, Aramaki T, Tamai M, Arima K, Nakamura H, Yamasaki S, Ida H, Kawakami A, Ueki Y, Matsuoka N, Nakashima M, Mizokami A, Kawabe Y, Mine M, Fukuda T, Eguchi K.	Reduction in serum levels of substance P in patients with rheumatoid arthritis by etanercept, a tumor necrosis factor inhibitor.	*Modern Rheumatology*	21(3)	244-250	2011
Kawashiri SY, Kawakami A, Iwamoto N, Fujikawa K, Satoh K, Tamai M, Nakamura H, Okada A, Koga T, Yamasaki S, Ida H, Origuchi T, Eguchi K.	The power Doppler ultrasonography score from 24 synovial sites or 6 simplified synovial sites, including the metacarpophalangeal joints, reflects the clinical disease activity and level of serum biomarkers in patients with rheumatoid arthritis.	*Rheumatology (Oxford)*	50(4)	962-965	2011
Kawashiri SY, Kawakami A, Okada A, Koga T, Tamai M, Yamasaki S, Nakamura H, Origuchi T, Ida H, Eguchi K.	CD4+CD25highCD127low/-Treg cell frequency from peripheral blood correlates with disease activity in patients with rheumatoid arthritis.	*Journal of Rheumatology*	38(12)	2517-2521	2011
Iwamoto N, Kawakami A, Tamai M, Arima K, Nakamura H, Kawashiri SY, Kita J, Okada A, Koga T, Kamachi M, Yamasaki S, Ichinose K, Ida H, Origuchi T, Uetani M, Eguchi K.	Magnetic resonance imaging of wrist and finger joints distinguishes secondary Sjögren's syndrome with rheumatoid arthritis from primary Sjögren's syndrome with articular manifestations.	*Clininal and Experimental Rheumatology*	29(6)	1062-1063	2011
井田弘明、吉浦孝一郎、金澤伸雄	特集□　自己炎症症候群 中條—西村症候群の臨床と病態	炎症と免疫（先端医学社）	19(2)	153-157	2011
井田弘明	自己炎症症候群の遺伝子異常	内科（南江堂）	107(4)	645-652	2011

井田弘明	医学用語解説　プロテアソーム	炎症と免疫（先端医学社）	19(3)	297-299	2011
井田弘明	自己炎症症候群と自然免疫	リウマチ科（科学評論社）	46(2)	149-154	2011
金澤伸雄、有馬和彦、井田弘明、吉浦孝一郎、古川福実	中條-西村症候群	日本臨床免疫学会誌	34(5)	388-400	2011