Genetic Regulation

1. Staffs and Students (in 2009)

Professor	Akinori KIMURA	
Associate Professor (Graduate School of Biomedical Science)		
	Toshiaki NAKAJIMA	
Assistant Professor	Takuro ARIMURA	
Research Associate	Taeko NARUSE	
Graduate Student	Daisuke SHICHI,	Taisuke ISHIKAWA,
	Haruna OHTSUKA	
Graduate Student (Biomedical Science PhD program)		
	Hitoshi OHTANI,	Makiko KONISHI,
	Jianbo ANN,	Yukiko OKUDA,
	Yusuke SAITO	
Visiting Graduate Student	Marika TAKAHASHI	
Visiting Post-doc	Zhiyong CHEN	
Visiting Student (Nangyang Technology University)		
	Hui Chiang LAU	

2. Purpose of Education

Genetic factors, i.e. structural and/or functional diversity of human genome, are more or less involved in the etiology and pathogenesis of human diseases. Main objective of Genetic Regulation is to identify the gene mutations or polymorphisms and to decipher the molecular mechanisms involved in the etiology and pathogenesis of intractable diseases, in order to develop new strategies for diagnosis, treatment and/or prevention of the diseases. Current research is focused on the cardiovascular diseases (e.g. idiopathic cardiomyopathy, idiopathic arrhythmia, and coronary heart disease), autoimmune diseases (e.g. Burger disease, Behcet disease, rheumatoid arthritis, and chronic thromboembolic pulmonary hypertension) and infectious diseases (e.g. HIV/AIDS). In addition, genome diversity in immune-related genes is investigated from the view-point of primate evolution.

3. Research Subjects

1) Identification and functional analysis of disease-related genes for cardiovascular diseases
2) Identification and functional analysis of disease-related genes for autoimmune diseases
3) Identification and functional analysis of disease-related genes for infectious diseases
4) Structural, functional and evolutional analyses of MHC and immune-related genes in vaccination

4. Publications

Original Article

1. Arimura T, Hayashi YK, Murakami T, Oya Y, Funabe S, Hirasawa EA, Hattori N, Nishino I, Kimura A. Mutational analysis of fukutin gene in dilated cardiomyopathy and hypertrophic cardiomyopathy. Circ J. 2009; 73(1): 158-161.
2. Kominami S, Tanabe N, Ota M, Naruse T, Katsuyama Y, Nakanishi N, Tomoike H, Sakuma M, Shirato K, Takahashi M, Shibata H, Yasunami M, Chen Z, Kasahara Y, Tatsumi K, Kuriyama T, Kimura A. HLA-DPB1 and NFKBIL1 may confer the susceptibility to chronic thromboembolic pulmonary hypertension in the absence of deep vein thrombosis. J Hum Genet. 2009; 54(2): 108-114.
3. Ueda K, Hirano Y, Higashiuesato Y, Aizawa Y, Hayashi T, Inagaki N, Tana T, Ohya Y, Takishita S, Muratani H, Hiraoka M, Kimura A. Role of HCN4 channel in preventing ventricular arrhythmia. J Hum Genet. 2009; 54(2): 115-121.
4. Shichi D, Ota M, Katsuyama Y, Inoko H, Naruse T, Kimura A. Complex divergence at a microsatellite marker C1_2_5 in lineage of HLA-Cw/-B haplotype. J Hum Genet. 2009; 54(4): 224-229.
5. Hinohara K, Nakajima T, Sasaoka T, Sawabe M, Lee BS, Ban J, Park JE, Izumi T, Kimura A. Replication studies for the association of PSMA6 polymorphism with coronary artery disease in East Asian populations. J Hum Genet. 2009; 54(4): 248-251.
6. Kikkawa EF, Tsuda TT, Sumiyama D, Naruse TK, Fukuda M, Kurita M, Wilson RP, LeMaho Y, Miller GD, Tsuda M, Murata K, Kulski JK, Inoko H. Trans-species polymorphism of the Mhc class II DRB-like gene in banded

penguins (genus Spheniscus). Immunogenetics.2009; 61(5): 341-352.
7. Tomoyasu Y, Yamaguchi T, Tajima A, Nakajima T, Inoue I, Maki K. Further evidence for an association between mandibular height and the growth hormone receptor gene in a Japanese population. Am J Orthod Dentofacial Orthop. 2009; 136(4): 536-541.
8. Kang EH, Yamaguchi T, Tajima A, Nakajima T, Tomoyasu Y, Watanabe M, Yamaguchi M, Park SB, Maki K, Inoue I. Association of the growth hormone receptor gene polymorphisms with mandibular height in a Korean population. Arch Oral Biol. 2009; 54(6): 556-562.
9. Romphruk AV, Romphruk A, Naruse TK, Raroengjai S, Puapairoj C, Inoko H, Leelayuwat C. Polymorphisms of NKG2D ligands: diverse RAET1/ULBP genes in Northeastern Thais. Immunogenetics. 2009; 61(9): 611-617.
10. Park YE, Hayashi YK, Bonne G, Arimura T, Noguchi S, Nonaka I, Nishino I. Autophagic degradation of nuclear components in mammalian cells. Autophagy. 2009; 5(6): 795-804.
11. Arimura T, Inagaki N, Hayashi T, Shichi D, Sato A, Hinohara K, Vatta M, Towbin JA, Chikamori T, Yamashina A, Kimura A. Impaired binding of ZASP/Cypher with phosphoglucomutase 1 is associated with dilated cardiomyopathy. Cardiovasc Res. 2009; 83(1): 80-88.
12. Moulik M, Vatta M, Witt SH, Alora AM, Murphy RT, McKenna WJ, Boriek A, Oka K, Labeit S, Bowles NE, Arimura T, Kimura A, Towbin JA. ANKRD -the gene encoding cardiac ankyrin repeat protein- is a novel dilated cardiomyopathy gene. J Am Coll Cardiol. 2009; 54(4):325-333.
13. Arimura T, Bos MJ, Sato A, Kubo T, Okamoto H, Nishi H, Harada H, Koga Y, Moulik M, Doi YL, Towbin JA, Ackerman MJ, Kimura A. Cardiac ankyrin repeat protein gene (ANKRD1) mutations in hypertrophic cardiomyopathy. J Am Coll Cardiol. 2009; 54(4): 334-342.
14. Kano T, Mori T, Kimura A. Gender ratio distortion in abortuses and live births from patients with recurrent spontaneous abortion. Am J Reprod Immunol. 2009; 62(3): 125-127.
15. Hinohara K, Nakajima T, Yasunami M, Houda S, Sasaoka T, Yamamoto K, Lee BS, Shibata H, Takahashi TY, Arimura T, Sato A, Naruse T, Ban J, Inoko H, Yamada Y, Sawabe M, Park JE,, Izumi T, Kimura A. Megakaryoblastic leukemia factor-1gene in the susceptibility to coronary artery disease. Hum Genet. 2009; 126(4): 539-547.
16. Hinohara K, Nakajima T, Sasaoka T, Sawabe M, Lee BS, Ban J, Park JE, Izumi T, Kimura A. Validation of the association between AGTRL1 polymorphism and coronary artery disease in Japanese and Korean populations. J Hum Genet. 2009; 54(9): 554-556.
17. Nakajima T, Nakayama EE, Kaur G, Terunuma H, Mimaya J, Ohtani H, Mehra N, Shioda Y, Kimura A. Impact of novel TRIM5-alpha variants, Gly110Arg and G176del, on the anti-HIV-1 activity and the susceptibility to HIV-1 infection. AIDS. 2009; 23(16): 2091-2100.
18. Hinohara K, Ohtani H, Nakajima T, Sasaoka T, Sawabe M, Lee BS, Ban J, Park JE, Izumi T, Kimura A. Validation of eight genetic risk factors in East Asian populations replicated the association of BRAP with coronary artery disease. J Hum Genet. 2009; 54(11), 642–646.
19. Sato A, Arimura T, Makita N, Ishikawa T, Aizawa Y, Ushinohama H, Aizawa Y, Kimura A. Novel mechanisms of trafficking defect caused by KCNQ1 mutations found in long QT syndrome. J Biol Chem. 2009; 284(50): 35122-35133.

Review Article

1. Nakajima T, Kimura A. Comparative genomics: insight into human health and disease. In The HLA Complex in Biology and Medicine: a resource book. (Mehra N, ed), pp566-588, Jaypee Brothers Medical Publishers Ltd., New Delhi, 2010
2. Kimura A. Does a gene polymorphism predisposing to the intermediate phenotype predict the risk of disease? Circ J. 2009; 73(6): 1016-1017.