



Naoyuki Tsuchiya

Faculty of Medicine

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Theme

- Identification of genomic variants associated with susceptibility and clinical characteristics of autoimmune rheumatic diseases through human genome analysis

Keyword ANCA-associated vasculitis, systemic lupus erythematosus, human genomics

Highlight

Major Scientific Interests of the Group

Our laboratory is interested in genetics and genomics of human autoimmune rheumatic diseases such as ANCA-associated vasculitis, systemic lupus erythematosus and systemic sclerosis. We are taking advantage of genome-wide approach and candidate gene approach in combination to identify genomic variants associated with development of the diseases, as well as those associated with their serious complications such as interstitial lung disease.

Currently, we are specifically interested in susceptibility genes to ANCA-associated vasculitis in east Asian populations, whose epidemiology greatly differs from that in the European populations, and largely unexplored. We are also interested in the role of variations in the genes whose analysis

remains challenging, such as multigene families and those with genomic structural variations.

Projects for Regular Students in Doctoral or Master's Programs

1. Identification of genomic variations associated with autoimmune rheumatic diseases
2. Biologic and bioinformatic analyses of the variations associated with autoimmune rheumatic diseases

Study Programs for Short Stay Students (one week – one trimester)

Genome database (tutorial), SNV genotyping (laboratory).

Other Faculty Members

Assistant Professor Aya Kawasaki

Applications and Prospects

- Our studies are expected to lead to better understanding of the pathogenesis of these enigmatic intractable diseases, as well as to identification of molecular targets and biomarkers valuable in establishing future precision medicine for human autoimmune rheumatic diseases.

Literature, intellectual property, work

- 1) Kawasaki A et al. Association of *TERT* and *DSP* variants with microscopic polyangiitis and myeloperoxidase-ANCA positive vasculitis in a Japanese population: a genetic association study. *Arthritis Res Ther* 2020; 22: 246.
- 2) Yamashita K, et al. Association of functional (GA)_n microsatellite polymorphism in the *FLI1* gene with susceptibility to human systemic sclerosis. *Rheumatology* 2020;59:3553-62.
- 3) Yokoyama N, et al.. Association of *NCF1* polymorphism with systemic lupus erythematosus and systemic sclerosis, but not with ANCA-associated vasculitis in a Japanese population. *Sci Rep* 2019;9:16366.
- 3) Namba N, et al.. Association of *MUC5B* promoter polymorphism with interstitial lung disease in myeloperoxidase-antineutrophil cytoplasmic antibody - associated vasculitis. *Ann Rheum Dis* 2019;78:1144-6.
- 5) Juge P-A, et al. *MUC5B* promoter variant and rheumatoid arthritis with interstitial lung disease. *N Engl J Med* 2018;379: 2209-19.