Human Genome Center

Laboratory of Genome Technology
シークエンス技術開発分野

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The major goal of our group is to identify genes of medical importance, and to develop new diagnostic and therapeutic tools. We have been attempting to isolate genes involving in carcinogenesis and also those causing or predisposing to various diseases as well as those related to drug efficacies and adverse reactions. By means of technologies developed through the genome project including a high-resolution SNP map, a large-scale DNA sequencing, and proteome/metabolome analysis, we have isolated a number of biologically and/or medically important genes, and are developing novel diagnostic and therapeutic tools.

1. Analysis of host genetic factors of various diseases

Germline TP53 variants have been predominantly studied within Li-Fraumeni syndrome (LFS) families. Growing evidence suggests that their broader contribution to cancer susceptibility is emerging as a public health concern, underscoring the need to redefine their frequency, functional spectrum, and clinical significance in large and diverse population cohorts. This study included 73,854 individuals with 23 cancer types and 38,288 controls, identified 276 germline TP53 variants in the Japanese population and conducted case-control association analyses. ClinVar pathogenic/likely pathogenic (P/LP) variants exhibit low carrier frequency in the population (approximately 1 in 3,738), but carriers demonstrate cancer penetrance approaching 90%, with markedly elevated risks for multiple cancers including female breast cancer, bone tumors, lymphoma, lung cancer, and gastric cancer. Two East Asian-enriched variants, demonstrated significant protective effects. In burden tests integrating multidimensional annotations, we observed distinct cancer risk patterns across variant categories. These findings provide novel insights into the diversity and clinical significance of TP53 germline mutations in East Asian populations, while underscoring the importance of incorporating population genetic background into risk stratification for TP53 variant carriers.

2. Large-scale omics analysis for disease biomarker screening

Tumor-associated autoantibodies (TAAbs) are promising biomarkers for lung cancer detection, yet their induction and clinical significance remain unclear. Serum samples from 140 healthy controls, 90 pulmonary fibrosis (PF), 90 small cell lung cancer (SCLC), and 365 lung adenocarcinoma (LUAD) cases were analyzed by protein array and two-stage ELISA validation. Distinct TAAb panels for SCLC and LUAD were identified. TAAb-based models showed excellent diagnostic accuracy (AUC > 0.8), outperforming ProGRP (AUC = 0.621) and CEA (AUC = 0.493) in distinguishing cancer from PF. TAAb induction was associated with antigen overexpression, somatic mutations, HLA class II polymorphisms, and immune infiltration. TAAb seropositivity correlated with older age and advanced stage, predicting poor survival in SCLC but favorable prognosis in advanced LUAD. In prediagnostic sera, TAAb levels increased progressively, detectable up to two years before diagnosis. These findings underscore TAAbs as accurate, early-detection biomarkers reflecting tumor–immune crosstalk and disease progression in lung cancer.

Publications

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