Human Genome Center

Laboratory of Functional Analysis *In Silico*
機能解析イン・シリコ分野

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Laboratory of Genome Database
ゲノムデータベース分野

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Our laboratory's mission is to conduct computational ("in silico") studies on the functional aspects of genome information. At present, we mainly focus on analyzing regulatory information about gene expression in the non-coding region using various next-generation sequencing (NGS) data. We also actively collaborate with researchers from various fields.

1. A graph-embedding approach to dissecting proximal and distal gene regulators

Sung-Joon Park and Kenta Nakai

The spatial organization of the genome plays a critical role in mediating the functional effects of distal chromosomal interactions. In particular, enhancer-promoter interactions have been intensively studied using advanced computational algorithms. However, our understanding of how enhancer signals are transmitted to their target promoters through complex regulatory networks remains limited. In this study, we developed a novel computational framework that combines a regression model, which predicts gene expression by identifying key promoter-distal and -proximal regulators, with a graph-embedding algorithm designed to detect cell-type-specific and conserved regulatory interactions within complex gene regulatory networks. We applied this method to human naïve and germinal center B cells and, as a result, identified sets of promoter-distal transcription factors and architectural cofactor proteins, which are co-regulated to maintain cellular stability and prevent malignancy. These findings emphasize the importance of understanding both cis- and trans-regulatory interactions in the transcriptional machinery. Our approach provides a valuable alternative for studying enhancer biology and its mediation through protein-protein interactions within the context of 3D genome organization.

2. Epigenetic profiling of housekeeping core promoters in the human genome

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This research investigates the role of HK-CREs in gene regulation and cancer development. We identify approximately 11,000 HK-CREs across the human genome, demonstrating their widespread influence beyond housekeeping gene expression. These elements are enriched in unmethylated CpG sites and exhibit extensive interactions with other genes. Our analysis reveals that aberrant methylation of HK-CREs, particularly those associated with zinc finger genes (ZNFGs), is prevalent in various cancer types. We observe a strong correlation between the expression of genes linked to these HK-CREs and patient survival, suggesting their critical role as tumor suppressors. Then, we use HiC data to explore the relationship between HK-CREs and zinc finger genes (ZNFGs). Despite transcriptional differences likely due to heterochromatin marks, HK-CREs and ZNFGs exhibit remarkable similarities in their epigenetic profiles. Both gene sets display extensive promoter-promoter interactions, suggesting a previously unrecognized relationship between them.

3. TF-EPI: an interpretable enhancer-promoter interaction detection method based on Transformer

Bowen Liu, Weihang Zhang, Xin Zeng, Martin Loza, Sung-Joon Park and Kenta Nakai

In this study, we developed TF-EPI, a deep-learning model based on a Transformer architecture to detect enhancer-promoter interactions solely from DNA sequences. The performance of TF-EPI surpassed that of other state-of-the-art methods on multiple benchmark datasets. Importantly, by utilizing the attention mechanism of the Transformer, we identified distinct cell type-specific motifs and sequences in enhancers and promoters, which were validated against databases such as JASPAR and UniBind, highlighting the potential of our method in discovering new biological insights. Moreover, our analysis of the transcription factors (TFs) corresponding to these motifs and short sequence pairs revealed the heterogeneity and commonality of gene regulatory mechanisms and demonstrated the ability to identify TFs relevant to the source information of the cell line. Overall, our work unveils important sequence information for the investigation of enhancer-promoter pairs based on the attention mechanism of the Transformer, providing an important milestone in the investigation of cis-regulatory grammar.

4. HyGAnno: Hybrid graph neural network-based cell type annotation for single-cell ATAC sequencing data

Weihang Zhang, Yang Cui, Bowen Liu, Martin Loza, Sung-Joon Park and Kenta Nakai

Reliable cell type annotations are crucial for investigating cellular heterogeneity in single-cell omics data. Although various computational approaches have been proposed for single-cell RNA sequencing (scRNA-seq) annotation, high-quality cell labels are still lacking in single-cell ATAC sequencing (scATAC-seq) data because of extreme sparsity and inconsistent chromatin accessibility between datasets. Here, we present a novel automated cell annotation method called HyGAnno that transfers cell type information from a well-labeled scRNA-seq reference to an unlabeled scATAC-seq target via a parallel graph neural network in a semi-supervised manner. Unlike existing methods that utilize only gene expression or gene activity features, HyGAnno integrates genome-wide accessibility peak features to facilitate the training process. In addition, HyGAnno reconstructs a reference-target cell graph that can be used to detect cells with low prediction reliability according to their specific graph connectivity patterns. HyGAnno was tested using large datasets and demonstrated the advantages of accurate cell annotation, interpretable cell embedding, robustness to noisy reference data, and adaptability to tumor tissues.

5. Spatial transcriptomics analysis via image-aided graph contrastive learning for domain exploration and alignment-free integration

Yitao Yang, Yang Cui, Xin Zeng, Yubo Zhang, Martin Loza, Sung-Joon Park and Kenta Nakai

Spatial transcriptomics is an essential application for investigating cellular structures and interactions and requires multimodal information to study spatial domains precisely. Here, we propose STAIG, a novel deep-learning model that integrates gene expression, spatial coordinates, and histological images using graph-contrastive learning coupled with high-performance feature extraction. STAIG can integrate tissue slices without pre-alignment and remove batch effects. Moreover, it was designed to accept data acquired from various platforms, with or without histological images. By performing extensive benchmarks, we demonstrated the capability of STAIG to recognize spatial regions with high precision and uncover new insights into tumor microenvironments, highlighting its promising potential in deciphering spatial biological intricates.

6. SCOIGET: a tool for predicting spatial tumor evolution patterns by inferring spatial copy number variation distributions

Yujia Zhang[2], Yitao Yang, Kenta Nakai and Hui Lu[2,3]

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A comprehensive spatiotemporal map of tumor heterogeneity is essential for understanding tumor evolution, with copy number variation (CNV) as a significant feature. Existing studies often rely on tools originally developed for single-cell data, which fail to utilize spatial information. Here, we introduce SCOIGET (Spatial COpy number Inference by Graph on Evolution of Tumor), a novel framework using graph neural networks with graph attention layers to learn spatial neighborhood gene expression features and infer copy number variations. Our model significantly improves the efficiency and accuracy of depicting tumor evolution, capturing detailed spatial and temporal changes within the tumor microenvironment. It is highly versatile, showing strong generalizability across various spatial omics technologies and cancer types, making it applicable to diverse downstream tasks. This performance enhances research efficiency and offers valuable insights into tumor progression. In conclusion, SCOIGET integrates multiple features with advanced algorithms to provide a detailed and accurate representation of tumor heterogeneity and evolution, supporting the development of personalized cancer treatment strategies.

7. Comparative single-cell transcriptomic analysis reveals putative differentiation drivers and potential origin of vertebrate retina

Xin Zeng, Fuki Gyoja[4], Yang Cui, Martin Loza, Takehiro Kusakabe[4] and Kenta Nakai
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Despite known single-cell expression profiles in vertebrate retinas, understanding their developmental and evolutionary expression patterns among homologous cell types remains limited. We examined and compared approximately 240,000 retinal cells from four species and found significant similarities among homologous cell types, indicating inherent regulatory patterns. To understand these shared patterns, we constructed gene regulatory networks for each developmental stage for three of these species. We identified 690 regulons governed by 530 regulators across three species, 10 common cell class-specific regulators, and 16 highly preserved regulons. RNA velocity analysis pinpointed conserved putative driver genes and regulators to retinal cell differentiation in both mouse and zebrafish. Investigation of the origins of retinal cells by examining conserved expression patterns between vertebrate retinal cells and invertebrate *Ciona intestinalis* photoreceptor-related cells implied functional similarities in light transduction mechanisms. Our findings offer insights into the evolutionarily conserved regulatory frameworks and differentiation drivers of vertebrate retinal cells.

8. Computational analysis reveals MHC-II expressing tumor cells influence immune surveillance and prognostic outcomes in triple-negative breast

Yang Cui, Weihang Zhang, Xin Zeng, Yitao Yang, Sung-Joon Park and Kenta Nakai

Triple-negative breast cancer (TNBC) is an aggressive subtype with poor prognosis and therapy resistance, driven partly by the tumor microenvironment (TME). In this study, we combined scRNA-seq and bulk RNA-seq data from TNBC patients and identified two TME-based subtypes: tumor-dense (TD) and non-tumor-dense (nonTD). TD is associated with a more malignant phenotype, reduced immune cell infiltration, and poor prognosis, while nonTD shows an active immune response and better outcomes. We identified a tumor cell subgroup, C3, with high MHC-II pathway activity, which interacts with CD4+ T cells. C3 cells were almost absent in TD but prevalent in nonTD, suggesting their role in immune responses. Spatial transcriptomics revealed that C3 cells co-localize with immune-infiltrated regions, indicating their role in recruiting immune cells to tumors. Finally, we developed survival and immune infiltration prediction models based on C3 gene signatures, achieving high accuracy. This work enhances understanding of TNBC biology and offers potential for improving clinical outcomes.

9. Integrative analysis of cancer multimodality data identifying COPS5 as a novel biomarker of diffuse large B-cell lymphoma

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Accurate clinical biomarkers are essential for preventing, diagnosing, and treating diseases; however, identifying them remains challenging. Advanced computational methods have accelerated biomarker discovery from complex multimodal data, but managing sparse data with missing information still limits performance and interpretability. To address this, we developed a pipeline that combines joint non-negative matrix factorization (JNMF) to identify key features in sparse, high-dimensional data with biological pathway analysis to interpret these features by detecting activated pathways. Applying this pipeline to large-scale cancer datasets, we identified genomic features relevant to specific cancers as common pat-

tern modules (CPMs) of JNMF. We found COPS5 to be a potential upstream regulator of pathways linked to diffuse large B-cell lymphoma (DLBCL). COPS5 demonstrated co-overexpression with DLBCL markers MYC, TP53, and BCL2, and its high expression correlated with lower survival probabilities in patients. Using CRISPR-Cas9, we confirmed COPS5's role in promoting tumor growth, suggesting it as a novel prognostic biomarker for DLBCL. This work demonstrates that integrating and simplifying complex data can uncover hidden biological insights, advancing the discovery of clinical biomarkers.

10. In-silico analysis revealed Marco (SR-A6) and Abca1/2 as potential regulators of lipid metabolism in M1 macrophage hysteresis

Yubo Zhang, Wenbo Yang, Yutaro Kumagai[6], Martin Loza, Yitao Yang, Sung-Joon Park and Kenta Nakai
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Macrophages undergo polarization, resulting in distinct phenotypes. These transitions, including de-/repolarization, lead to hysteresis, where cells retain genetic and epigenetic signatures of previous states, influencing macrophage function. We previously identified a set of interferon-stimulated genes (ISGs) associated with high lipid levels in macrophages that exhibited hysteresis following M1 polarization, suggesting potential alterations in lipid metabolism. In this study, we applied weighted gene co-expression network analysis (WGCNA) and conducted comparative analyses on 162 RNA-seq samples from de-/repolarized and lipid-loaded macrophages, followed by functional exploration. Our results demonstrate that during M1 hysteresis, the sustained high expression of Marco (SR-A6) enhances lipid uptake, while the suppression of Abca1/2 reduces lipid efflux, collectively leading to elevated intracellular lipid levels. This accumulation may compensate for reduced cholesterol biosynthesis and provide energy for sustained inflammatory responses and interferon signaling. Our findings elucidate the relationship between M1 hysteresis and lipid metabolism, contributing to understanding the underlying mechanisms of macrophage hysteresis.

11. RNA secondary structure prediction by conducting multi-class classifications

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Predicting the RNA secondary structure based on the RNA sequence is challenging, as valid predictions should follow various constraints. While several deep learning methods have been developed for predicting RNA secondary structures, they commonly adopt post-processing steps to adjust the model output to produce valid predictions, which are complicated and could limit performance. In this research, we propose a simple method by considering RNA secondary structure prediction as multiple multi-class classifications, eliminating the need for those complicated post-processing steps. We use this method to train and evaluate our model based on the attention mechanism and the convolutional neural network. Besides, we introduce two additional methods, including data augmentation to improve further the within-RNA-family performance and a method to alleviate the performance drop in the cross-RNA-family evaluation. We could produce valid predictions and perform better without complex post-processing steps. We show that our additional methods benefit the performance of within-RNA-family and cross-RNA-family evaluations.

12. Characterization of trans-spliced chimeric RNAs: insights into the mechanism of trans-splicing

Rui Yokomori, Takehiro G. Kusakabe[4] and Kenta Nakai

Trans-splicing is a post-transcriptional processing event that joins exons from separate RNAs to produce a chimeric RNA. However, the detailed mechanism of *trans*-splicing remains poorly understood. Here, we characterize *trans*-spliced genes and provide insights into the mechanism of *trans*-splicing in the tunicate *Ciona*. Tunicates are the closest invertebrates to humans, and their genes frequently undergo trans-splicing. Our analysis revealed that, in genes that give rise to both trans-spliced and non-trans-spliced messenger RNAs, trans-splice acceptor sites were preferentially located at the first functional acceptor site, and their paired donor sites were weak in both *Ciona* and humans. Additionally, we found that *Ciona trans*-spliced genes had GU- and AU-rich 5′ transcribed regions. Our data and findings are not only useful for the *Ciona* research community but may also aid in a better understanding of the *trans*-splicing mechanism, potentially advancing the development of gene therapy based on *trans*-splicing.

13. Developing an open-access repository for the multi-dimensional genome structure data

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The community-wide effort to characterize 3D genome organization has underscored the significance of functional connections between genetic and epigenetic processes and the physical properties of DNA, such as stiffness, torsion, and supercoiling. However, the mechanisms that establish functional genome structures remain poorly understood, highlighting the need for comprehensive and integrative approaches. In this context, we are developing a data repository system, the Genome Modality Suite, as part of the research project Genome Modality. The system is designed to handle heterogeneous and multi-dimensional data, including RNA-seq and ChIP-seq signal tracks (1D data), Hi-C contact matrices (2D data), and XYZ-coordinate genome structures (3D data). Additionally, we have successfully launched the system for public use, leveraging PHP, MySQL, and JavaScript libraries, and we are continuously refining its features. Our system aims to accelerate progress in understanding the multi-dimensional properties of the genome.

14. Single-cell transcriptome analysis of ocular-like cell lineages derived from human pluripotent stem cells

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The generation of self-formed, ectodermal, autonomous multi-zone structures, known as SEAM, from human induced pluripotent stem cells (hiPSCs) provides a unique opportunity to explore the dynamics of ocular cell differentiation. In this study, we used single-cell transcriptomics to study ectodermally-derived ocular cell populations that emerge during SEAM formation. Our analysis reveals the interdependence between early eye tissues and outlines the sequential formation of specific cell types over a 12-week period. We demonstrate a progression from pluripotency to ocular tissue specification and differentiation, including cornea, conjunctiva, lens, and retina. These findings not only enhance our understanding of ocular development in a human stem cell-based model but also establish a robust methodology for investigating cellular and molecular dynamics during SEAM formation at single-cell resolution. Furthermore, they underscore the potential of hiPSC-derived systems as powerful platforms for modeling human eye development and disease.

15. Advances in immunology: insights into B cell dynamics, antibody discovery, and TCR repertoires in autoimmunity

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This research explores immune cell behavior and innovative approaches to address infectious and autoimmune diseases. In COVID-19 patients, single-cell transcriptomics revealed proinflammatory signatures in B cells correlated with disease severity and uncovered a developmental pathway linking atypical memory B cells to conventional memory B cells. Key genes, such as ZFP36 and DUSP1, were identified as drivers of differentiation and activation, shedding light on their roles in immune recovery and response to SARS-CoV-2. A bioinformatics pipeline was developed to accelerate antibody discovery using machine learning and AI, integrating sequencing data to predict neutralizing antibodies. This scalable approach reduces the resources needed for therapeutic development and offers potential applications for other diseases. In parallel, analysis of T cell receptor (TCR) repertoires in ZAP-70 mutant mice revealed how attenuated TCR signaling promotes self-reactive Th17 cells in inflamed joints. This disruption shifts regulatory T cells (Tregs) toward a conventional T cell-like repertoire, increasing autoimmune susceptibility. These findings provide critical insights into the mechanisms of self-tolerance breakdown, with implications for rheumatoid arthritis and related autoimmune diseases. Together, these studies enhance our understanding of immune dynamics and demonstrate how advanced technologies can drive new therapeutic interventions in immunology.

16. Exploration for the critical quality attributes (CQA) of chondrocytes derived from polydactyly patients with guaranteed efficacy

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Osteoarthritis of the knee (OAK) is a degenerative disease commonly affecting middle-aged and elderly women over 40. It progresses gradually due to factors such as lower limb deformation (e.g., bow legs), genetic predisposition, aging, obesity, muscle weakening, joint inflammation, or trauma from heavy work. OAK significantly reduces activities of daily living and quality of life, presenting a major challenge in aging societies by shortening healthy life expectancy. Despite its high prevalence, no curative treatment exists, and advanced cases often require artificial joint replacement. To address this, Tokai University has pioneered articular cartilage regeneration by transplanting chondrocyte sheets in OAK patients. Clinical studies have shown that both autologous and allogeneic chondrocyte sheet transplants can repair and regenerate OAK cartilage defects with hyaline cartilage, restoring original joint function. As part of this initiative, our research focuses on developing Critical Quality Attributes (CQAs) for allogeneic raw cells using bioinformatics tools, leveraging data provided by Tokai University. These CQAs are vital for ensuring the consistency, comparability, and scalability of allogeneic cell sheets for mass production. This approach aims to provide a curative treatment for OAK, enabling patients to preserve their natural joints throughout life without artificial replacements.

17. Resting heart rate and risk of dementia: a mendelian randomization study in the international genomics of Alzheimer's project and UK biobank

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Observational studies suggest a higher resting heart rate (RHR) is linked to an increased risk of dementia, but the causal relationship remains unclear. This study used two-sample Mendelian randomization to assess whether genetically predicted higher RHR influences Alzheimer's disease (AD) risk. Summary statistics from genome-wide association studies (GWAS) were analyzed using the generalized summary Mendelian randomization (GSMR) approach. Outcomes included AD diagnosis, maternal and paternal family history of AD (from UK Biobank), and a combined meta-analysis of these GWAS results. Further adjustments were made to account for RHR-modifying medications. The GSMR analysis found no significant causal relationship between genetically predicted higher RHR and AD risk (β_{GSMR} = 0.12, P = 0.30), maternal family history (β_{GSMR} = −0.18, P = 0.13), or paternal family history (β_{GSMR} = −0.14, P = 0.39). These findings remained consistent after adjusting for medication effects (β_{GSMR} = −0.03, P = 0.72). This study concludes that RHR does not causally influence dementia risk, suggesting that previous observational associations likely result from shared correlations between RHR and cardiovascular conditions.

Publication list

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