Advanced Clinical Research Center

Division of Hematopoietic Disease Control
造血病態制御学分野

Professor	Yasuhito Nannya, M.D., Ph.D.	教　授	博士(医学)	南　谷　泰　仁
Associate Professor	Takaaki Konuma, M.D., Ph.D.	准教授	博士(医学)	小　沼　貴　晶
Assistant Professor	Koji Jimbo, M.D., Ph.D.	助　教	博士(医学)	神　保　光　児

The main goal of our research is to elucidate the pathogenesis of hematopoietic diseases and to study the development of therapeutic strategies for these diseases. We will continue to develop the therapeutic targets that have already been identified and advance them to the next stage for clinical application. First, as part of Japan's Whole Genome Project for Blood Disorders, we are advancing analyses of ALL and lymphoma. We have discovered multiple novel mutations and are progressing with their functional analyses. Regarding clonal progression in PNH, we performed whole-genome analysis of single-cell-derived colonies and analyzed the roles indicated by immune attacks and additional genetic mutations. We also analyzed ctDNA after hematopoietic stem cell transplantation and evaluated its role in predicting relapse. To evaluate the role of MSCs in post-transplant GVHD, we performed proteomics analysis and discovered a novel mechanism: MSCs suppress GVHD by enhancing the function of MDSCs. Additionally, we are developing diagnostic tools for MDS using peripheral blood via Ghost cytometry and developing AI to predict the significance of genomic mutations. Furthermore, we developed an efficient in vitro amplification system for AML cells, comparing it with PDX models, and are exploring its application for genome editing and drug sensitivity testing.

1. *Genomic Landscape of Newly Diagnosed Adult Acute Lymphoblastic Leukemia*

Authors: Hidehito Fukushima[1], Kaito Mimura[2], Koji Okazaki[3], Hiroyuki Takamori[1], Ryunosuke Saiki[3], Yotaro Ochi[3], Kazuaki Yokoyama[1], Kenichi Yoshida[2], Motohiro Kato[4], Seishi Ogawa[3], and Yasuhito Nannya[1]

Affiliations:

1. Division of Hematopoietic Disease Control, Institute of Medical Science, The University of Tokyo, Tokyo, Japan

2. Division of Genome Analysis Platform Development, National Cancer Center Research Institute, Tokyo, Japan

3. Department of Pathology and Tumor Biology, Graduate School of Medicine, Kyoto University, Kyoto, Japan

4. Department of Pediatrics, The University of Tokyo, Tokyo, Japan

While pediatric acute lymphoblastic leukemia (ALL) has been extensively characterized through multi-omics sequencing, the genomic architecture of newly diagnosed adult ALL remains incompletely defined. We performed whole-genome sequencing on 156 *de-novo* adult cases (126 B-cell/MPAL, 30 T-cell) using matched buccal swab germline controls. Compared to pediatric ALL, canonical subtypes such as *ETV6::RUNX1*, hyperdiploid, and hypodiploid are far less frequent in adult ALL. Notably, we identified recurrent structural variants in three relatively under-

studied genes, with recurrence rates of 45%, 18%, and 12% respectively, following well-characterized alterations in *IKZF1*, *CDKN2A*, and *PAX5*.

Alterations in these genes exhibit specific patterns of mutual exclusivity and coexistence with other drivers, were associated with distinct transcriptional states, and correlated with survival. These findings underscore heterogeneity in adult ALL and highlight the potential value of incorporating these genes' status into adult risk models and the design of future clinical trials.

2. Mesenchymal Stromal Cells-mediated Neutrophil Functional Reprogramming in Acute Graft-versus-host disease

Tomokazu Seki[1], Kazuaki Yokoyama[2,3], Xinyu Huang[2], Seiko Kato[3], Yuho Najima[4], Noriko Doki[4], Tokiko Nagamura-Inoue[2,3], Yasuhito Nannya[1,3]
1. Division of Hematopoietic Disease Control, The Institute of Medical Science, The University of Tokyo, Tokyo, Japan.
2. Department of Cell Processing and Transfusion, Research Hospital, The Institute of Medical Science, The University of Tokyo, Tokyo, Japan.
3. Department of Hematology and Oncology, Research Hospital, The Institute of Medical Science, The University of Tokyo, Tokyo, Japan.
4. Hematology Division, Tokyo Metropolitan Cancer and Infectious Diseases Center, Komagome Hospital, Tokyo, Japan.

Acute graft-versus-host disease (GVHD) remains a life-threatening complication following allogeneic hematopoietic stem cell transplantation. Neutrophils have traditionally been regarded as short-lived effector cells that exacerbate tissue inflammation and injury in acute GVHD; however, emerging evidence suggests that neutrophils exhibit functional plasticity and may serve as inducible immunoregulatory cells under specific inflammatory contexts. Mesenchymal stromal cell (MSC) therapy has demonstrated clinical efficacy in steroid-refractory GVHD, primarily attributed to immunomodulation of various immune cells, especially aberrantly activated T cells. However, whether MSCs modulate GVHD through functional reprogramming of neutrophils remains poorly understood. To investigate MSCs-mediated neutrophil immunomodulation, we performed targeted plasma proteomics using a proximity extension assay, stress-conditioned in vitro co-culture experiments, and carboxyfluorescein diacetate succinimidyl ester–based mixed lymphocyte reaction (MLR) assays. In steroid-refractory GVHD patients treated with umbilical cord–derived MSCs, plasma proteome profiling revealed significant upregulation of neutrophil-associated pathways such as neutrophil degranulation and vesicle. Consistently, in vitro co-culture of stress-primed neutrophils with umbilical cord–derived MSCs induced the expression of myeloid-derived suppressor cells (MDSC)–associated markers on neutrophils, such as CD66b and LOX-1. Functionally, these MSC-conditioned neutrophils exhibited elevated reactive oxygen species production and suppressed lymphocyte proliferation in MLR assays. Collectively, these findings suggest that MSCs mitigate GVHD by functionally reprogramming neutrophils toward an immunosuppressive, MDSC–like phenotype.

3. Ghost Cytometry–based Assessment of Morphological Abnormalities in Myelodysplastic Syndromes

Tomokazu Seki[1], Sadao Ota[2], Yasuhito Nannya[1,3]
1. Division of Hematopoietic Disease Control, The Institute of Medical Science, The University of Tokyo, Tokyo, Japan.
2. ThinkCyteK.K., Tokyo, Japan.
3. Department of Hematology and Oncology, Research Hospital, The Institute of Medical Science, The University of Tokyo, Tokyo, Japan.

Myelodysplastic syndromes (MDS) are clonal hematologic malignancies characterized by ineffective dysplastic hematopoiesis, peripheral cytopenia, and a risk of progression to acute myeloid leukemia. Although therapeutic options for lower-risk MDS have expanded in recent years, the disease remains underdiagnosed due to the reliance on invasive bone marrow examination. To address this limitation, we sought to develop a peripheral blood–based screening assay capable of identifying patients with MDS based on morphological abnormalities. We employed ghost cytometry, a label-free single-cell analysis platform that captures high-dimensional cellular morphological signals from peripheral blood without the use of antibodies or stains. Peripheral blood cells from patients with MDS (n=10) exhibited reproducible morphological signals distinct from those of healthy individuals (n=9). To establish machine learning-based model identifying patients with MDS, the validation study is ongoing among four cohorts: cytopenic patients without MDS (n=20), patients with lower-risk MDS (n=20), patients with higher-risk MDS (n=20), and healthy controls (n=20). Peripheral blood–based morphological profiling using ghost cytometry enables discrimination of MDS from non-MDS cytopenia without the need for invasive procedures. This screening approach may reduce unnecessary bone marrow examinations and allow diagnostic resources to be focused on patients most likely to benefit.

4. Two mechanistic archetypes of paroxysmal nocturnal hemoglobinuria evolution revealed by lifelong, time-scaled single-cell-based phylogenetics

Hiroyuki Takamori1), 2), Yasutaka Ueda1), Yoshikazu Matsuoka3), Tatsuya Fujioka3), Nozomi Yusa4), Kazuaki Yokoyama5), Hideki Makishima6), Yoshiko Murakami7), Taroh Kinoshita7), 8), Seiya Imoto9), Satoru Miyano10), Yuzuru Kanakura1), 11), Jun-Ichi Nishimura1), Naoki Hosen1), Seishi Ogawa12), and Yasuhito Nannya2),5)

1. Department of Hematology and Oncology, The University of Osaka Graduate School of Medicine
2. Division of Hematopoietic Disease Control, The Institute of Medical Science, The University of Tokyo
3. Department of iPS Stem Cell Regenerative Medicine, Kansai Medical University
4. Department of Laboratory Medicine, The Institute of Medical Science, The University of Tokyo, Japan
5. Department of Hematology and Oncology, The Institute of Medical Science Research Hospital, The University of Tokyo, Tokyo, Japan
6. Department of Hematology and Clinical Oncology, Shinshu University School of Medicine
7. Laboratory of Immunoglycobiology, Research Institute for Microbial Diseases, The University of Osaka
8. Center for Infectious Disease Education and Research, The University of Osaka;
9. Division of Health Medical Intelligence, Human Genome Center, the Institute of Medical Science, The University of Tokyo
10. M&D Data Science Center, Tokyo Medical and Dental University
11. Department of Hematology, Sumitomo Hospital
12. Department of Pathology and Tumor Biology, Kyoto University

Paroxysmal nocturnal hemoglobinuria (PNH) requires substantial expansion of PIGA-mutated hematopoietic stem cell clones, yet the lifelong clonal histories leading to disease onset remain largely unknown. We integrated time-scaled single-cell–based whole-genome phylogenetic analysis with long-term clinical follow-up to dissect how immune-mediated selection and driver mutations shape PIGA-mutated clonal expansion. We analyzed 202 hematopoietic colonies from two PNH patients representing contrasting evolutionary archetypes. In Patient 1, eight independent PIGA-mutated clones lacking additional driver mutations expanded only after onset of aplastic anemia (AA), in parallel with a marked loss of wild-type clonal diversity. In Patient 2, a PIGA-mutated clone carrying an HMGA2 rearrangement expanded slowly for more than a decade before onset of AA. The PIGA/HMGA2–mutated clone gradually increased its effective size encompassing the period of AA/PNH while the size of wild type clone was suppressed low compared with healthy individuals. These observations indicate that clonal dominance in this case was driven primarily by AA-associated immune selection rather than by an intrinsic proliferative advantage conferred by HMGA2. Together, these results define two mechanistic routes to PNH—pure immune-selection and driver-assisted evolution—and reveal that autoimmune bottlenecks are the decisive events transforming indolent PIGA-mutated hematopoiesis into clinical disease.

5. Deciphering the Genetic Landscape of Hairy Cell Leukemia: The Japanese Variant Perspective

Yokoyama K1, Kamitani I2,3, Kidoguchi K4, Ishii H2, Shimizu E5, Imoto S5, Takizawa J6,7,Ogawa S8, Nannya Y1,3, Kimura S4

1. Department of Hematology/Oncology, IMSUT Hospital
2. Division of Clinical Oncology/Hematology, Department of Internal Medicine, The Jikei University School of Medicine
3. Division of Hematopoietic Disease Control, IMSUT
4. Division of Hematology, Respiratory Medicine and Oncology, Department of Internal Medicine, Faculty of Medicine, Saga University, Saga, Japan
5. Human Genome Center, IMSUT
6. Department of Hematology, Endocrinology and Metabolism, Niigata University Faculty of Medicine, Niigata
7. Prospective observation study of chronic lymphocytic leukemia, hairy cell leukemia and related diseases in Japan (CLLRSG-01)
8. Department of Pathology and Tumor Biology, Graduate School of Medicine

Hairy cell leukemia (HCL) is a rare, indolent mature B-cell neoplasm characterized by villous lymphocytes proliferating primarily in the bone marrow and spleen. Three major subtypes exist: classical HCL (HCLc), prevalent in Western countries with near-universal BRAF V600E mutations responsive to purine analogs; Western-type variant (HCLv), harboring MAP2K1 mutations in approximately 50% of cases with frequent purine analog resistance; and Japanese-type variant (HCLjv), predominantly observed in Japan, lacking BRAF V600E mutations, often refractory to purine analogs, with undefined molecular pathogenesis and no established standard therapy. Through whole-genome sequencing of more than 20 HCLc/HCLv/HCLjv cases, we identified novel genetic alterations: structural variants positioning oncogene X near the immunoglobulin heavy chain enhancer (Eμ), loss-of-function mutations in non-canonical NFκB pathway suppressor gene Y, and epigenetic regulator gene Z mutations. Patient samples demonstrated X protein overexpression with Akt pathway activation and NFκB activation upon Y deletion. Functional studies confirmed these findings, and compound screening identified the proteasome inhib-

itor bortezomib as a promising therapeutic candidate through dual Akt/NFκB pathway inhibition. This study aims to establish mouse models recapitulating HCLjv pathogenesis and elucidate the cooperative mechanisms between X and Y mutations in disease development.

6. Post-transplant circulating tumor DNA monitoring predicts relapse and survival in AML and MDS patients undergoing allogeneic hematopoietic stem cell transplantation: A KSGCT1702 prospective multicenter study

Miho Ogawa[1], Kazuaki Yokoyama[2], Nozomi Yusa[3], Mika Ito[2], Eigo Shimizu[4], Takaaki Konuma[1], Seiko Kato[1], Seiya Imoto[4], Yasuhito Nannya[1,2], Satoshi Takahashi[5], and Yoshinobu Kanda[6-8]

1. Division of Hematopoietic Disease Control, IMSUT

2. Department of Hematology/Oncology, IMSUT Hospital

3. Department of Laboratory Medicine, IMSUT Hospital

4. Human Genome Center, IMSUT

5. Division of Precision Research Platform, IMSUT

6. Division of Hematology, Saitama Medical Center, Jichi Medical University

7. Division of Hematology, Department of Medicine, Jichi Medical University

8. Kanto Study & Group for Cell Therapy (KSGCT)

Background: Disease relapse remains the leading cause of mortality following allogeneic hematopoietic stem cell transplantation (allo-HSCT) for acute myeloid leukemia (AML) and myelodysplastic syndromes (MDS). Minimally invasive measurable residual disease (MRD) detection methods are needed.

Methods: In this prospective multicenter study within the Kanto Hematopoietic Stem Cell Transplantation Group, we analyzed circulating tumor DNA (ctDNA) in 50 AML/MDS patients undergoing myeloablative allo-HSCT. Patient-specific digital PCR assays targeting driver mutations identified by comprehensive genomic profiling were performed at pretransplant and post-transplant landmarks (Days 30, 60, 90, 120).

Results: ctDNA-MRD positivity at Days 60–90 strongly predicted relapse and survival. Day 90 achieved the highest discrimination (C-index 0.789), while Day 60 provided optimal specificity. In multivariable analysis, ctDNA positivity remained independently prognostic (adjusted subdistribution hazard ratio 2.03–3.10), superseding conventional risk factors including ELN 2022 classification. Dynamic kinetic analysis identified patients with increasing ctDNA between Days 60–90 as having extremely high relapse risk.

Conclusions: Post-transplant ctDNA-MRD monitoring at Days 60–90 provides powerful, independent risk stratification, supporting its integration into post-HSCT surveillance and preemptive intervention trials.

7. Development of Explainable AI Technology for Cancer Genome Variant Interpretation Support

Katsuhiko Murakami 1, Shin-ichiro Tago1, Rikuhiro Kojima1, Sho Takishita1, Hiroaki Morikawa1, Miho Ogawa 2,, Hidehito Fukushima 2 , Hiroyuki Takamori 2 , Kazuaki Yokoyama 3, Yasuhito Nannya 2,3 , Seiya Imoto 4 and Masaru Fuji 11.

1. Computing Laboratories, Fujitsu Research, Fujitsu Ltd.,

2. Division of Hematopoietic Disease Control, IMSUT

3. Department of Hematology/Oncology, IMSUT Hospital

4. Human Genome Center, IMSUT

Background: Interpreting the clinical significance of genetic variants detected by next-generation sequencing remains a time-consuming bottleneck in cancer genomic medicine. We developed explainable artificial intelligence (XAI) technologies to support variant interpretation for expert panel preparation.

Methods: We constructed a large-scale knowledge graph integrating approximately 15.5 billion triples from multiple genomic databases (COSMIC, ClinVar, dbNSFP, Mitelman, Pfam). Using Deep Tensor deep learning and large language models (LLMs), we developed systems for: (1) automated expert panel report generation using stepwise prompts mimicking physician reasoning, (2) pathogenicity prediction with natural language explanations for single nucleotide variants (SNVs), and (3) fusion gene pathogenicity interpretation with mechanistic explanations.

Results: For fusion gene report generation, our method achieved 79% disease coverage and 93% drug coverage, outperforming LLM such as ChatGPT alone (71% and 46%, respectively). The SNV pathogenicity predictor achieved 0.94 accuracy and 0.98 AUC on ClinVar validation data. The fusion gene XAI attained an F1 score of 84.5% while providing mechanistic explanations consistent with published literature.

Conclusions: Knowledge graph-based XAI enables high-accuracy pathogenicity prediction with interpretable explanations, potentially reducing physician workload while maintaining transparency in clinical decision-making. These technologies represent a significant step toward AI-assisted precision oncology.

8. Development of novel ex vivo culture systems for hematological malignancies

Chao LI[1,2], Kimihito Cojin Kawabata[3], Satoshi Yamazaki[4], Satoshi Takahashi[1,3], Yasuhito Nannya[1]

1. Division of Hematopoietic Disease Control, Insti-

tute of Medical Science, The University of Tokyo, Tokyo, Japan
2. Department of Medical Genome Sciences, Graduate School of Frontier Sciences, The University of Tokyo, Tokyo, Japan
3. Project Division of Clinical Precision Research Platform, Institute of Medical Science, The University of Tokyo, Tokyo, Japan,
4. Division of Cell Regulation, Institute of Medical Science, The University of Tokyo, Tokyo, Japan

In the field of biological assays using primary tumor specimens derived from patients with hematological malignancies, how to conduct ex vivo culture of tumor cells, maintaining their fundamental characteristics plays significant roles. We conducted 21-day ex vivo culture experiments using a total of 56 primary tumor specimens (PTS) derived from AML patients, evaluating the efficacy of 21 different culture systems. The performance of these culture systems was assessed using multicolor flow cytometry to monitor leukemic cell viability, number of cells, and stem-like characteristics (defined by CD45, CD34, CD38, HLA-DR, and other lineage markers).

Through many trials and errors, , we identified a superior serum-free condition supplemented with a specific combination of polymers, a cytokine receptor activator, cord blood expansion agents, and a small molecule. By day 21, this optimized condition outperformed traditional OP-9 feeder cell systems in the enrichment and expansion of leukemic stem cells. The stem-like, or repopulating phenotypes of our ex vivo expanded PTS were validated using both flow cytometry, showing either maintained or expanded prematurity, increased colony-forming capacity, using specimens harvested after 21 days of ex vivo culture.

Furthermore, this medium facilitated successful CRISPR-mediated gene knockout while sustaining leukemic cell viability, as well as their prematurity, over extended periods. Drug sensitivity screening performed on long-term cultured PTS yielded results consistent with pre-culture specimens in many points.

Our results are suggestive of the extended applicability of PTS in biological experiments, drug screening, and gene etiding Use of the protocol in drug screening experiments could cover a much longer term than the current 3-day standards. Furthermore, basic research on leukemia will be comprehensively conducted using PTS with extended flexibility in wet assays.

Publications

1. Gurnari, C., H. Makishima, A. Durmaz, E. Attardi, R. Saiki, A. Bataller, G. Sapinho, L. Gondek, Y. Nannya, S. Best, P. Krishnamurthy, K.L. Kong, Y. Atsuta, S. Kasahara, K. Ohyashiki, Y. Miyazaki, N. Kanemura, N. Hiramoto, F. Versino, M.J. Montoro, S. Torres-Esquius, A. Jerez Cayuela, M. Lopez-Esteban, C. Martinez-Laperche, H. Awada, V. Visconte, C.D. DiNardo, M.T. Voso, A.E. DeZern, G. Garcia-Manero, A.G. Kulasekararaj, J.P. Maciejewski, and S. Ogawa, DDX41-mutant myeloid neoplasms defy current prognostic schemes and require a dedicated risk scoring system: a multicenter, retrospective study. Leukemia, 2025.
2. Hata, R., S. Funakoshi, T. Makiyama, T. Kato, M. Narita, Y. Matsumura, R. Hirohata, Y. Naka, K. Ida, H. Ito, A. Yoshizawa, Y. Nannya, H. Haga, S. Ogawa, K. Ono, T. Kimura, and Y. Yoshida, Integrative Transcriptomic-Histological Analysis in Dilated Cardiomyopathy Unveils FGFR1 Inhibition as Anti-Cardiac Fibrotic and Cardioprotective Therapy. JACC Basic Transl Sci, 2025. 10(10): p. 101363.
3. Ikoma, Y., N. Nakamura, Y. Kaneda, H. Takamori, T. Seki, N. Hiramoto, J. Kitagawa, J. Kanda, K. Fujita, T. Morishita, Y. Ochi, S. Chiba, N. Sasaki, M. Ichii, K. Imada, M. Watanabe, M. Hishizawa, Y. Miyahara, Y. Maesako, Y. Tanaka, S. Oka, M. Tsuji, S. Yoshihara, K. Mitani, Y. Ueda, T. Kitano, M. Watanabe, N. Sezaki, T. Kondo, S. Kasahara, A. Takaori-Kondo, N. Kanemura, S. Ogawa, and Y. Nannya, Impact of myelodysplasia-related gene mutations and residual mutations at remission in venetoclax/azacitidine for AML. Leukemia, 2025. 39(6): p. 1362-1367.
4. Jimbo, K., M. Nojima, K. Toriuchi, M. Yamagishi, M. Nakashima, Y. Yamano, K. Uchimaru, and Y. Nannya, Long-term kinetics of proviral load in HTLV-1 carriers: defining risk for the development of adult T-cell leukemia/lymphoma. Biomark Res, 2025. 13(1): p. 34.
5. Konuma, T., M. Hamatani-Asakura, S. Kato, S. Andoh, H. Ichimura, J. Kuroda, K. Kondo, K. Yokoyama, M. Monna-Oiwa, Y. Nannya, and S. Takahashi, Recipient polymorphism of the C-reactive protein (CRP) gene rs2794521 stratified disease-free survival, but did not influence maximum serum CRP levels during neutropenia after single-unit cord blood transplantation in adults. Leuk Lymphoma, 2025: p. 1-4.
6. Konuma, T., M. Hamatani-Asakura, M. Monna-Oiwa, S. Kato, S. Andoh, H. Ichimura, K. Yokoyama, Y. Nannya, and S. Takahashi, Clinical impact of glucocorticoid responsiveness-related gene polymorphism on graft-versus-host disease and survival after single-unit cord blood transplantation. Int J Hematol, 2025.
7. Konuma, T., M. Hamatani-Asakura, M. Mon-

na-Oiwa, S. Kato, S. Andoh, Y. Oda, K. Yokoyama, Y. Nannya, and S. Takahashi, Risk of acute graft-versus-host disease in recipients with IL-10 promoter polymorphism rs1800872, but not IL-10 receptor beta polymorphism rs2834167, after single-unit cord blood transplantation. Hum Immunol, 2025. 86(4): p. 111322.
8. Konuma, T., M. Hamatani-Asakura, M. Monna-Oiwa, S. Kato, M. Isobe, K. Yokoyama, Y. Nannya, and S. Takahashi, Higher relapse and worse overall survival in recipients with CTLA-4 AA genotype of rs231775 following single-unit cord blood transplantation in adults. Leuk Lymphoma, 2025. 66(4): p. 733-743.
9. Konuma, T., M. Monna-Oiwa, S. Kato, M. Isobe, S. Takahashi, and Y. Nannya, Prognostic Value of the Pretransplant Fibrosis-4 Index on Non-Relapse and Overall Mortality following Unrelated Single-Unit Cord Blood Transplantation in Adults. Acta Haematol, 2025. 148(3): p. 258-268.
10. Kynning, M.K., E. Westerberg, L. Forsell, M. Creignou, D.M. Berggren, B. Tesi, E. Bernard, E. Papaemmanuil, Y. Nannya, L.C. Franco, D. Valentini, E.H. Lindberg, S. Ogawa, E. Ejerblad, and J. Ungerstedt, Comorbidities and mutations including single- and multihit TET2 mutations in relation to outcome in chronic myelomonocytic leukaemia-A population-based study. Br J Haematol, 2025.
11. Nakashima, M., K. Nagai, N. Takao, N. Araya, Y. Kuze, J. Mizuike, S. Tosaka, S. Aratani, N. Yagishita, E. Horibe, T. Watanabe, T. Sato, Y. Nannya, Y. Suzuki, K. Uchimaru, M. Yamagishi, and Y. Yamano, Chromatin remodeling enhances MAP3K8 expression in HAM: a key pathogenesis for therapeutic intervention. Nat Commun, 2025. 16(1): p. 9874.
12. Oshima, M., N. Takayama, Y. Nakajima-Takagi, D. Shinoda, N. Itokawa, S. Kurosawa, S. Kaito, T. Kamiya, Y. Yamada, S. Andoh, K. Kayamori, S.K. Paul, M.A. Kanashiro, T. Muto, S. Tsukamoto, E. Sakaida, E. Sato, N. Yusa, K. Yokoyama, Y. Nannya, S. Imoto, B. Rahmutulla, A. Kaneda, K. Yamaguchi, Y. Furukawa, N. Doki, K. Eto, K. Nishikawa, Y. Ding, T. Myojo, Y. Harada, H. Harada, and A. Iwama, Chromatin accessibility in stem cells unveils progressive transcriptional alterations in myelodysplastic syndrome. Nat Commun, 2025. 16(1): p. 10726.
13. Suzuki, K., S. Kusayanagi, Y. Kuze, M. Hata, S. Kozuma, K. Jimbo, Y. Nannya, Y. Suzuki, K. Uchimaru, and M. Yamagishi, Bidirectional anti-tumor and immunological strategies by targeting GARP-TGF-beta axis in adult T-cell leukemia/lymphoma. Leukemia, 2025. 39(10): p. 2465-2476.
14. Takamori, H., Y.J. Huang, H. Fukushima, K. Yokoyama, T.Y. Huang, M.C. Kuo, S. Ogawa, Y. Nannya, and L.Y. Shih, Whole-genome sequencing of myeloproliferative neoplasms revealed dynamic clonal changes in the fibrotic or leukemic transformation and novel FOXP1 mutations in the fibrotic transformation. Leukemia, 2025.
15. Yanada, M., S. Yamasaki, S. Mizuno, J. Sugita, T. Fujino, Y. Misaki, M. Tanaka, N. Uchida, M. Onizuka, N. Doki, S. Ota, M. Sawa, T. Kawakita, Y. Hasegawa, H. Nakamae, K. Ishiwata, N. Hiramoto, F. Ishimaru, J. Kanda, M. Ohbiki, Y. Atsuta, H. Nakasone, and T. Konuma, Comparison of single-unit umbilical cord blood transplantation and haploidentical transplantation using posttransplant cyclophosphamide during first complete remission of acute myeloid leukemia. Bone Marrow Transplant, 2025.
16. Yokoyama, A., K. Watanabe, Y. Inoue, T. Hirano, M. Tamaoki, K. Hirohashi, S. Kawaguchi, Y. Ishida, Y. Takeuchi, Y. Kishimoto, S.K. Kim, C. Katada, Y. Nannya, H. Seno, S. Ogawa, M. Muto, and N. Kakiuchi, Somatic mosaicism in the buccal mucosa reflects lifestyle and germline risk factors for esophageal squamous cell carcinoma. Sci Transl Med, 2025. 17(796): p. eadq6740.
17. Zaimoku, Y., T. Imi, T. Hatada, H. Mizumaki, H. Mura, H. Yoshino, Y. Kano, M. Kobayashi, E. Morishita, N. Fushida, T. Matsushita, K. Mizuguchi, H. Ikeda, Y. Nannya, S. Ogawa, K. Hosomichi, N. Doki, Y. Katayama, T. Koike, K.I. Matsuoka, T. Nishida, Y. Takahashi, K. Kataoka, H. Nakazawa, Y. Ueda, T. Fukuda, T. Ichinohe, F. Ishimaru, M. Onizuka, Y. Atsuta, T. Miyamoto, T. Japanese Society of, and T. Cellular, Prevalence and outcome of VEXAS syndrome in unrelated hematopoietic cell transplantation for bone marrow failure. Clin Exp Med, 2025. 25(1): p. 300.

和文業績

1. 南谷泰仁　白血球数・白血球分画　臨床検査ガイド2025年度版 文光堂 p480-483
2. 南谷泰仁　骨髄異形成症候群　今日の治療指針2025年度 医学書院　p667-669
3. 小川弥穂、南谷泰仁　がんゲノム情報に基づくAML診療の現状と展望　EBM血液疾患の治療2025　中学医学社　p110-114
4. 南谷泰仁　白血病・リンパ腫におけるゲノム医療 Medical Practice　2025/2　Vol42(2) p262-265
5. 南谷泰仁　造血器腫瘍におけるゲノム異常に基づく治療薬アクセス　週刊医学のあゆみ　ゲノム解析時代の血液腫瘍学　医歯薬出版株式会社 Vol292(1)　p7-11
6. 南谷泰仁　骨髄系腫瘍診療におけるパネル検査の臨床的有用性　週刊医学のあゆみ　造血器腫瘍の遺伝子パネル検査の臨床的意義　医歯薬出版株式会社　Vol293(12)　p1132-1134
7. 南谷泰仁　造血器腫瘍に対するがん遺伝子パネル

検査 がん最新の薬物療法　2025-2026　南江堂 P6-10
8. 南谷泰仁　骨髄異形成症候群の染色体異常・遺伝子異常：概論　新・白血病学　日本臨床社　p71-78
9. 南谷泰仁　固形腫瘍における腫瘍浸潤クローン性造血の影響 腫瘍内科　科学評論社
10. 南谷泰仁　骨髄異形成症候群におけるゲノム情報　腫瘍内科　科学評論社 vol 35(5) p 456-462, 2025
11. 南谷泰仁　モダンメディア　ヘムサイト診断薬®を用いた造血器腫瘍のゲノム解析　栄研化学株式会社
12. 南谷泰仁　診療の参照ガイド　骨髄異形成症候群診療の参照ガイド　令和7年改訂版　特発性造血障害に関する調査研究班
13. 南谷泰仁　クローン性造血とその原因･影響 日本内科学会雑誌　日本内科学会
14. 南谷泰仁　骨髄異形成症候群（MDS）分子病態と予後　白血病治療マニュアル　南江堂
15. 小沼貴晶　臍帯血移植の現状と将来展望　臨床血液　2025;66(5):331-338. Japanese. doi: 10.11406/rinketsu.66.331. PMID: 40467462.
16. 小沼貴晶　臍帯血移植とHLA半合致移植の比較 EBM血液疾患の治療2025　中学医学社　p537-543
17. 小沼貴晶　造血幹細胞移植の前処置　血液疾患最新の治療2026-2028　南江堂　p247-252
18. 神保光児　成人T細胞白血病/リンパ腫におけるCD7およびCADM1を用いたフローサイトメトリープロファイルの有用性　臨床血液　2025 年 66 巻 4 号 p. 209-219　doi: 10. 11406/rinketsu. 66. 209. PMID: 40350270
19. 神保光児、南谷泰仁　ヒトT細胞白血病ウイルス1型　白血病と言われたら第7版下巻　全国骨髄バンク推進連絡協議会　p155-158