

Publication List

ICON KEY



Biomarker Detection



Hematology



MRD



Order of Mutations



Solid Tumor



Clinical Profiling



Mouse



Single-cell Multi-omics



Single-cell DNA Sequencing



Therapy Resistance

2022



Guess, T. et al., *Blood Cancer Discovery*

Distinct patterns of clonal evolution drive myelodysplastic syndrome progression to secondary acute myeloid leukemia

- Used DNA + protein multi-omics to characterize the clonal architecture and evolution in MDS patients who progress to sAML.



Droskea, L. et al., *BioRxiv preprint*

High-throughput single-cell sequencing for retroviral reservoir characterization

- Described a scDNA-seq method optimized for sequencing of proviral and host DNA for retroviral reservoir characterization.



Bianchi, A. et al., *BioRxiv preprint*

scTAM-seq enables targeted high-confidence analysis of DNA methylation in single cells

- Developed scTAM-seq, a targeted bisulfite-free method for analyzing DNA methylation at single-CpG resolution across thousands of single cells.



Maslah, N. et al., *Blood Advances*

Single-cell analysis reveals selection of TP53-mutated clones after MDM2 inhibition

- Demonstrated that MDM2 treatment can directly favor the emergence of TP53-mutated subclones in chronic myeloproliferative neoplasms (MPN).




 **Meyers, S. et al., HemaSphere**

Monitoring of Leukemia Clones in B-cell Acute Lymphoblastic Leukemia at Diagnosis and During Treatment by Single-cell DNA Amplicon Sequencing

- *Characterized the mutational heterogeneity and evolution in B-ALL, both at diagnosis and during treatment.*



 **Zhang, H. et al., BioRxiv preprint**

Application of high-throughput, high-depth, targeted single-nucleus DNA sequencing in pancreatic cancer

- *Optimized a highly automated nuclei extraction workflow that achieved fast and reliable targeted single-nucleus DNA library preps from pancreatic adenocarcinoma (PDAC) patients*




 **Snellings, D.A. et al., Nature Cardiovascular Research**

Developmental Venous Anomalies are a Genetic Primer for Cerebral Cavernous Malformations

- *Adapted sc-DNA sequencing to evaluate the relationship between developmental venous anomalies (DVAs) and cerebral cavernous malformations (CCMs)*



 **Leighton, J. et al., BioRxiv preprint**

Reconstructing mutational lineages in breast cancer by multi-patient targeted single cell DNA sequencing

- *Developed a Multi-Patient-Targeted (MPT) scDNA-sequencing method to study tumor evolution in breast cancer*



 **Kim, T. et al., British Society for Hematology**

Single cell proteogenomic sequencing identifies a relapse-fated AML subclone carrying FLT3-ITD with CN-LOH at chr13q

- *Used DNA + protein multi-omics to detect a relapse-fated subclone at the time of initial diagnosis that was enriched with homozygous 21 bp FLT3-ITDs.*



 **Blombery, P E. et al., Blood**

Clonal hematopoiesis, myeloid disorders and BAX-mutated myelopoiesis in patients receiving venetoclax for CLL

- *Sc-DNA sequencing revealed patterns of co-occurring BAX mutations in the myeloid compartments of CLL patients receiving venetoclax therapy*



2022 cont.



Wang, E. et al., *The New England Journal of Medicine*

Mechanisms of Resistance to Noncovalent Bruton's Tyrosine Kinase Inhibitors. *The New England Journal of Medicine*

- *Identified clonal architecture of resistance mutations to a new class of non-covalent BTK inhibitors in CLL patients*



Zhang, Q. et al., *Signal Transduction and Target Therapy*

Activation of RAS/MAPK pathway confers MCL-1 mediated acquired resistance to BCL-2 inhibitor venetoclax in acute myeloid leukemia

- *Identified the activation of RAS/MAPK pathway, leading to increased stability and higher levels of MCL-1 protein, as a major acquired mechanism of VEN resistance in AML.*



Rodriguez, S. et al., *Science Advances*

Preneoplastic somatic mutations including MYD88 L265P in lymphoplasmacytic lymphoma

- *DNA + protein multi-omics demonstrated that mutation in a gene relevant to B cell function, such as MYD88, represents a preneoplastic event that requires additional genetic alterations to drive lymphomagenesis*



2021



Thompson E.R. et al., *Blood Advances*

Single-cell sequencing demonstrates complex resistance landscape in CLL and MCL treated with BTK and BCL2 inhibitors

- *Identified clonal architecture of acquired genomic resistance to BTK and BCL2 inhibitors in CLL and MCL patients.*



Zhao Y. et al., *Nature*

Diverse alterations associated with resistance to KRAS(G12C) inhibition

- *Revealed a heterogeneous pattern of resistance involving many genes and multiple subclonal events emerging during KRAS (G12C) inhibitor treatment for lung and colorectal cancer.*



Sharma R. et al., *Blood*

Gain-of-function mutations in RPA1 cause a syndrome with short telomeres and somatic genetic rescue

- *Single-cell DNA+protein multi-omics revealed somatic genetic rescue events that evolved independently in HSPCs, myeloid cells, and B-cells in patients with telomere disorders.*





Sahoo S.S. et al., *Nature Medicine*

Clinical evolution, genetic landscape, and trajectories of clonal hematopoiesis in SAMD9/SAMD9L syndromes

- *Uncovered diversity of somatic genetic rescue events in pediatric instances of SAMD9/9L syndromes, some of which serve as “natural gene therapy.”*



Dillon L.W. et al., *Blood Cancer Discovery*

Personalized single-cell proteogenomics to distinguish acute myeloid leukemia from nonmalignant clonal hematopoiesis

- *Distinguished malignant variants of AML from age-related clonal hematopoiesis by resolving immunophenotypic identity of clonal architecture.*



Marin-Bejar, O. et al., *Cancer Cell*

Evolutionary predictability of genetic versus nongenetic resistance to anticancer drugs in melanoma

- *An analysis of genetic and nongenetic mechanisms of therapy resistance in melanoma.*



Thijssen, R. et al., *Blood*

Intact TP-53 function is essential for sustaining durable responses to BH3-mimetic drugs in leukemias

- *Revealed the effect of TP-53 mutation status on treatment of AML with the BH3-mimetic drugs.*



Wang, F. et al., *Nature Communications*

Leukemia stemness and co-occurring mutations drive resistance to IDH inhibitors in acute myeloid leukemia

- *Investigated clones that enabled relapse in AML patients who received IDH inhibitor treatment.*



Ren, A. et al., *Nature*

PIK3CA and CCM mutations fuel cavernomas through a cancer-like mechanism

- *Single-nuclei sequencing revealed patterns of co-mutation that promote cerebral cavernous malformations (CCMs).*



Lim, K.H. et al., *Blood Advances*

Clonal evolution and heterogeneity in advanced systemic mastocytosis revealed by single-cell DNA sequencing

- *Investigation of resistance to the multikinase inhibitor, midostaurin, in patients with advanced systemic mastocytosis (advSM).*





Demaree, B. et al., *Nature Communications*

Joint profiling of DNA and proteins in single cells to dissect genotype-phenotype associations in leukemia

- *Genotype & immunophenotype “decoupling” in leukemias samples was revealed using single-cell multi-omics.*



Peretz, C.A. et al., *Blood Advances*

Single cell DNA sequencing reveals complex mechanisms of resistance to quizartinib

- *Investigated mechanisms of resistance to quizartinib in patients with FLT3-ITD mutated AML.*



Iacobucci, I. et al., *Blood*

Modeling and targeting of erythroleukemia by hematopoietic genome editing

- *Used scDNA-seq analyze CRISPR-edited cells in preclinical models of acute erythroid leukemia (AEL).*



Kennedy, A.L. et al., *Nature Communications*

Distinct genetic pathways define pre-malignant versus compensatory clonal hematopoiesis in Shwachman-Diamond syndrome

- *Zygosity and CN-LOH are reliably measured with high sensitivity (0.1%) and predict progression to leukemia in patients with Shwachman-Diamond syndrome.*



Alberti-Servera, L. et al., *Blood*

Single-cell DNA amplicon sequencing reveals clonal heterogeneity and evolution in T-cell acute lymphoblastic leukemia

- *First study to assess the clonality and order of mutation acquisition of T-cell acute lymphoblastic leukemia (T-ALL) patient samples.*



Patel, B.A. et al., *Haematologica*

Detectable mutations precede late myeloid neoplasia in aplastic anemia

- *Identified evolution of mutations in a rare case where an severe aplastic anemia (SAA) patient who received immunosuppressive therapy developed MDS/AML with normal cytogenetics.*



Thompson E.R. et al., *Haematologica*

Clonal independence of *JAK2* and *CALR* or *MPL* mutations in comutated myeloproliferative neoplasms demonstrated by single cell DNA sequencing

- *First publication in T-cell acute lymphoblastic leukemia (T-ALL) that highlights SNVs/indels and MRD detection.*




Miles, L.A. et al., *Nature*

Single-cell mutation analysis of clonal evolution in myeloid malignancies

- *Used DNA + protein multi-omics to assess clonal architecture and evolution of AML, MPN, and clonal hematopoiesis.*


Morita, K. et al., *Nature Communications*

Clonal evolution of acute myeloid leukemia revealed by high-throughput single-cell genomics

- *Extensive study investigating genotype and immunophenotype associated with clonal evolution in AML.*


Ten Hacken, E. et al., *Genome Biology*

High throughput single-cell detection of multiplex CRISPR-edited gene modifications

- *Analyzed single and multiplexed CRISPR edits in individual cells in models of chronic lymphocytic leukemia (CLL).*


Xiao, W. et al., *Blood Advances*

A JAK2/IDH1-mutant MPN clone unmasked by ivosidenib in an AML patient without antecedent MPN

- *Clonal architecture of a relapsed AML patient, in which treatment with an IDH1 inhibitor promoted outgrowth of a minor myeloid neoplasm (MPN) clone.*


Taylor, J. et al., *Blood*

Single-cell genomics reveals the genetic and molecular bases for escape from mutational epistasis in myeloid neoplasms

- *Studied a rare phenomenon in biology where 2 splicing mutations co-occur in the patient sample.*


Maia, C. et al., *Blood*

Biological and clinical significance of dysplastic hematopoiesis in patients with newly-diagnosed multiple myeloma

- *Identified biomarkers from multiple myeloma samples that develop into MDS using multidimensional flow cytometry and single-cell DNA-seq.*


Choe, S. et al., *Blood Advances*

Molecular mechanisms mediating relapse following ivosidenib monotherapy in IDH1-mutant relapsed or refractory AML

- *Biopharma study that showed molecular resistance mechanisms in 174 patients following ivosidenib monotherapy for R/R AML.*



2020 cont.

DiNardo, C.D. et al., *Blood*

Molecular patterns of response and treatment failure after frontline venetoclax combinations in older patients with AML



- *Molecular basis for treatment resistance or durable remission in older patients with AML given venetoclax combination therapy.*

Ediriwickrema, A. et al., *Blood Advances*

Single-cell mutational profiling enhances the clinical evaluation of AML MRD



- *MRD detection identified clones at remission that expanded into the dominant clone at relapse in patients with AML.*

2019

Xu, L. et al., *Scientific Reports*

Clonal evolution and changes in two AML patients detected with a novel single-cell DNA sequencing platform



- *Clonal remodeling in patients with AML after bone marrow transplant revealed donor chimerism and unique clones.*

Gao, Y. et al., *Cancer Discovery*

V211D mutation in MEK1 causes resistance to MEK inhibitors in colon cancer



- *Treatment resistance mechanism revealed in colon cancer PDX model after treatment with MEK inhibitor binimetinib and EGFR antibody panitumumab.*

McMahon, C. et al., *Cancer Discovery*

Clonal selection with Ras pathway activation mediates secondary clinical resistance to selective FLT3 inhibition in acute myeloid leukemia



- *Treatment resistance mechanism revealed in AML with co-occurring FLT3 and RAS mutations after treatment with the FLT3-inhibitor gilteritin in patients with AML.*

2018

Pellegrino, M. et al., *Genome Research*

High-throughput single-cell DNA sequencing of acute myeloid leukemia tumors with droplet microfluidics



- *First publication that showcased single-cell DNA sequencing of thousands of cells using droplets in longitudinal AML sample.*

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