

BIOGRAPHICAL SKETCH

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NAME: Ju, Young Seok

eRA COMMONS USER NAME (credential, e.g., agency login): YOUNGSEOK

POSITION TITLE: Associate Professor (tenured), Graduate School of Medical Science and Engineering, Korea Advanced Institute of Science and Technology (KAIST)

EDUCATION/TRAINING (*Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable. Add/delete rows as necessary.*)

INSTITUTION AND LOCATION	DEGREE (if applicable)	Completion Date MM/YYYY	FIELD OF STUDY
Seoul National University, Seoul, Korea	M.D.	02/2007	Medicine
Seoul National University, Seoul, Korea	Ph.D.	02/2010	Biochemistry
Life Science Institute, Macrogen Inc, Korea	Research	03/2013	Genomic Medicine
Wellcome Trust Sanger Institute, Hinxton, UK	Research	10/2015	Cancer Genomics

A. Personal Statement

Young Seok Ju, M.D., Ph.D. is an Associate Professor at the Graduate School of Medical Science and Engineering, Korea Advanced Institute of Science and Technology (KAIST), Daejeon, South Korea, and the Director of the Center for Somatic Mutation and Mosaicism (SMM), KAIST. His goal is to understand the genomic mosaicism and cellular heterogeneity in somatic and germline tissues, and to trace clonal dynamics in normal and cancer tissues using acquired mutations as cellular barcodes. He started whole-genome analyses in 2007 (Seoul National University College of Medicine, Ph.D.) and completed the first Korean genome sequence (Nature 2009). Then he used his genome techniques to explore cancer genome mutations. He identified KIF5B-RET fusion oncogene in lung cancer (Genome Res 2011), clonal evolution of lung adenocarcinomas to small-cell carcinomas (J Clin Oncol 2017), complex genomic arrangements for fusion oncogene generation in lung cancer (Cell 2019), and mitochondrial DNA mutations in cancer (Nature Genetics 2020). In 2015, his interest expanded to mutations in normal tissues, to systematically delineate somatic mosaicism at single-cell resolution, and to reconstruct the clonal dynamics of normal cells from the earliest stage of human life using the mutations (Nature 2017, Nature 2021, Nature 2023). For the last 7 years, his lab has produced and explored whole-genomes of over 1,000 single-cell expanded normal clones (often using organoid culture techniques) to extensively decompose somatic mosaicism. The laboratory has trained 7 MD-PhDs and 2 PhDs, and now has 9 graduate students. In 2020, Dr. Ju founded Genome Insight Inc. (San Diego, CA) with the goal of revolutionizing healthcare through translating cancer and rare disease whole-genome data for personalized medical care. Dr. Ju brings expertise in NGS, organoids, bioinformatics, and computational biology, as well as his cross-disciplinary knowledge from working in both academia and industry.

Major ongoing grants

Leader Researcher Grant Ju (PI) 06/01/2020-02/28/2029
 National Research Foundation (NRF) of Korea (total KRW 7B; ~USD 5.4M)
 "MosaiClone: tracing somatic mosaicism and clonal expansion in human lifetime"

The primary goal of this project is to extensively characterize the landscape of somatic mosaicism by all types of genomic variants in numerous human cell types at single-cell and nucleotide resolution.

Role: PI

SUHF Investigator Program Ju (PI) 01/01/2019-12/31/2023
The Suh Kyungbae Foundation (SUHF) (total KRW 2.5B; ~USD 2M)
"Origins and functional consequences of complex genomic rearrangements in cancer cells"
The major goal of this project is to accurately uncover the molecular mechanism and precisely measure the transforming effect of complex genome rearrangements by integrating cutting-edge technologies.
Role: PI

B. Positions, Scientific Appointments, and Honors

Positions and Scientific Appointments

2010-2013 Postdoctoral Researcher, Life Science Institute Macrogen Inc., Korea (subs. of military service)
2013-2015 Postdoctoral Fellow, Cancer Genome Project, Wellcome Trust Sanger Institute, Hinxton, UK
2015-2019 Assistant, Associate Professor, Grad School of Med Sciences and Engineering, KAIST, Korea
2021- Director, Center for Somatic Mutation and Mosaicism, KAIST, Korea
2020- Genomics co-founder, Genome Insight Inc., San Diego, CA
2022-2023 Visiting Scholar, University of California San Diego Neurosciences, La Jolla, CA

Honors and Awards

2010 Best Ph.D. Dissertation Award, Korean Society for Molecular and Cellular Biology
2010 20th Wunsch Medical Award (for young medical scientist), Korean Academy of Medical Sciences
2013-2015 EMBO Long Term Fellowship for Postdoctoral Research
2018-2019 POSCO TJ Park Science Fellowship for Young Assistant Professors
2018- Member of Y-KAST (Young Korean Academy of Science and Technology)
2018- HFSP Young Investigator Grants (with BK Koo at IMBA and Snippert H at UMC Utrecht)
2019- Research Fellow of the Suh Kyungbae Research Foundation (SUHF)
2020 24th Young Scientist Award, Ministry of Science and ICT (Korea President Award)
2020 13th Asan Award in Medicine for Young Medical Scientists
2022 1st Lim Sungki Researcher Award for Young Medical Scientists
2022 International Human Genome Organization (HUGO) Chen Award of Excellence

C. Contributions to Science

1. (2007-2011; genome technologies) I focused on developing key bioinformatics techniques for exploring the personal whole-genome and transcriptome sequences. The techniques are foundations for his subsequent cancer genome and somatic mosaicism studies.

- a. Kim J*, **Ju YS*** et al. Seo J. (2009) A highly annotated whole-genome sequence of a Korean individual. *Nature* 460(7258). PMID:19587683 (co-first author)
- b. Park H*, Kim JI*, **Ju YS*** et al. Seo J. (2010) Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. *Nat Genet.* 42(5). PMID:20364138 (co-first author)
- c. **Ju YS** et al. Seo J. (2011) Extensive genomic and transcriptional diversity identified through massively parallel DNA and RNA sequencing of eighteen Korean individuals. *Nat Genet.* 43(8). PMID:21725310

2. (2012-present; cancer genomics) I applied genome techniques to cancer tissues to identify somatically acquired mutations in human neoplastic cells.

- a. **Ju YS** et al. Seo J. (2012) A transforming KIF5B and RET gene fusion in lung adenocarcinoma revealed from whole-genome and transcriptome sequencing. *Genome Res.* 22(3). PMID:22194472
- b. **Ju YS** et al. Campbell PJ. (2014) Origins and functional consequences of somatic mitochondrial DNA mutations in human cancer. *eLife.* 2014 Oct 1(3). PMID:25271376
- c. **Ju YS** et al. Stratton MR. (2015) Frequent somatic transfer of mitochondrial DNA into the nuclear genome of human cancer cells. *Genome Res.* 25(6). PMID:25963125
- d. Lee JK et al. **Ju YS.** (2017) Complex chromosomal rearrangements by single catastrophic pathogenesis in NUT midline carcinoma. *Ann Oncol.* 28(4). PMID:28203693

- e. Lee JK et al. **Ju YS**. (2017) Clonal history and genetic predictors of transformation into small cell carcinomas from lung adenocarcinomas. *J Clin Oncol*. 35(26). PMID:28498782
 - f. Lee JJ-K et al. **Ju YS#** & Kim YT#. (2019) Tracing oncogene rearrangements in the mutational history of lung adenocarcinoma. *Cell*. 177(7). PMID:31155235 (lead contact, co-corresponding)
 - g. Yuan Y*, **Ju YS*** et al. Liang H. (2020) Comprehensive molecular characterization of mitochondrial genomes in human cancers. *Nat Genet*. 52(3). PMID:32024997 (co-first author)
3. (2015-present; somatic mosaicism) I applied genome techniques to reveal somatic mosaicism in normal tissues, and to reveal clonal dynamics from the human early embryonic development across the aging.
- a. **Ju YS** et al. Stratton MR. (2017) Somatic mutations reveal asymmetric cellular dynamics in the early human embryo. *Nature*. 543(7647). PMID:28329761
 - b. Park S et al. **Ju YS**. (2022) Clonal dynamics in early human embryogenesis inferred from somatic mutation. *Nature*. 597(7876). PMID:34433967
 - c. Nam C et al. **Ju YS**. (2023) Widespread somatic L1 retrotransposition in normal colorectal epithelium. *Nature*. Published online ahead of print. PMID:37165195
 - d. Youk J et al. **Ju YS**. (2021) Mutational impact and signature of ionizing radiation. *bioRxiv*.
4. (2020-present) I applied genome and organoid culture techniques to understand COVID-19 pathophysiology.
- a. Youk J et al. **Ju YS#**, Lee JH#. (2020) Three-dimensional human alveolar stem cell culture models reveal infection response to SARS-CoV-2. *Cell Stem Cell*. 27(6). PMID:33142113 (lead contact, co-corresponding)
 - b. Yi K et al. **Ju YS**. (2021) Mutational spectrum on SARS-CoV-2 during the global pandemic. *Exp Mol Med*. 53(8). PMID:34453107
 - c. Kim T et al. **Ju YS**. (2022) Relative infectivity of the SARS-CoV-2 Omicron variant in human alveolar cells. *iScience*. 25(12). PMID:36406862

Complete List of Published Work:

<https://pubmed.ncbi.nlm.nih.gov/?term=young+seok+ju%5Bauthor%5D&sort=date>