

Curriculum Vitae

last update: Jan 10th 2024

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Associate Professor, Graduate School of Medical Sciences and Engineering (GSMSE),
Korea Advanced Institute of Science and Technology (KAIST), Daejeon, 34141, Korea

EXPERIENCE

- 2021.3 – present **Director** Center for Somatic Mutation and Mosaicism (CSM2), KAIST, Korea
- 2015.10 - present **Assistant Professor/Associate Professor**, GSMSE, KAIST, Korea
PI of Laboratory of COSMOS (<http://julab.kaist.ac.kr>)
(Comprehensive Observation of Somatic Mosaicism, its Origins and Significance)
- 2020.1 - present **Genomics co-founder**, GENOME INSIGHT Inc., San Diego, CA, USA.
- 2013.4 - 2015.10 **Postdoctoral Fellow**, Cancer Genome Project, Wellcome Sanger Institute, UK
Advisors: Prof. Mike Stratton & Dr. Peter Campbell
- 2010.3 - 2013.3 **Postdoctoral Research** (Substitution of military service), Macrogen Inc., Korea

EDUCATION AND TRAINING

- 2007.3 - 2010.2 **PhD** Biochemistry, Seoul National University College of Medicine, Korea
- 2001.3 - 2007.2 **MD** Seoul National University College of Medicine, Korea

10 SELECTED PUBLICATIONS

(#, corresponding author; * first author; First four and last authors are shown for papers with >5 authors; JIF from Clarivate 2023; Citation numbers from Google Scholar)

1. Nam CH, Youk J, Kim JY, Lim J, ..., Kwon HW#, Kim MJ#, **Ju YS#**. Widespread somatic L1 retrotransposition in normal colorectal epithelium. *Nature*. 2023; 617(7961):540-547. (JIF=64.8; Citation 8)
Discovery of high jumping (retrotransposition) activity of LINE-1 elements in human normal somatic cells.
2. Park S, Mali NM, Kim R, Choi J-W, ..., Oh JW#, **Ju YS#**. Clonal dynamics in early human embryogenesis inferred from somatic mutation. *Nature*. 2021;597(7876):393-397. (JIF=64.8; Citation 77)
Tracing early human embryonic development by uncovering the lineage relationships between cells that reside in different parts of the body using somatic mutations as endogenous cellular barcodes.
 - ✓ News & Views, *Nature*, "Mutation fingerprints encode cellular histories",
 - ✓ Research Highlight, *Nat Rev Genet*, "A body-wide view of somatic mutations",
 - ✓ News & Views, *Nat Rev Nephrol*, "Mutational fingerprints reconstruct human cell genealogies"
3. Lee JJ-K, Park S, Park H, Kim S, ... , **Ju YS# (Lead Contact)** & Kim YT#. Tracing oncogene rearrangements in the mutational history of lung adenocarcinoma. *Cell*. 2019;177(7):1842-1857. (JIF=64.5; Citation 160)
Tracing tumorigenic processes of fusion-oncogene driven lung adenocarcinomas of non-smokers using 138 cancer whole-genome sequencing.
4. Lee JK, Lee J, Kim S, Kim S, ..., Kim TM# & **Ju YS#**. Clonal history and genetic predictors of transformation into small cell carcinomas from lung adenocarcinomas. *Journal of Clinical Oncology* 2017 10;35(26):3065-3074. (JIF=45.3; Citation 372)
Tracing the cellular trajectory of clonal evolution from lung adenocarcinoma to small cell carcinomas during clinical course using longitudinal whole-genome sequencing.
 - ✓ Editorial, *J Clin Oncol*, "Unlocking the mystery of SCLC transformation in EGFR mutant adenocarcinoma"
5. Youk J, Kim T, Evans KV, Jeong Y-I, ... Choi B-S#, **Ju YS# (Lead Contact)**, Lee JH#. Three-dimensional human alveolar stem cell culture models reveal infection response to SARS-CoV-2. *Cell Stem Cell*. 2020;27(6):905-919. (JIF=23.9; Citation 200)
The first establishment of feeder-free, human alveolar organoid in chemically defined condition and investigating early infection response of human alveolar cells against SARS-CoV-2.
 - ✓ News Feature, *Nature*, "The mini lungs and other organoids helping to beat COVID"

6. Lee JK, Louzada S, An Y, Kim SY, ... Kim TM# & **Ju YS#**. Complex chromosomal rearrangements by single catastrophic pathogenesis in NUT midline carcinoma. *Annals of Oncology* 2017; 28(4):890-897. (JIF=50.5; Citation 61)
Exploring whole-genome sequences of NUT carcinomas which are rare but highly aggressive. Identified chromoplexy as a mechanism for the genesis of oncogenic NUT fusion genes.
7. **Ju YS***, Martincorena I, Gerstung M, Petljak M, ... & Stratton MR. Somatic mutations reveal asymmetric cellular dynamics in the early human embryo. *Nature*. 2017;543(7647):714-718. (JIF=64.8; Citation 274)
Exploring 241 whole-genome sequences of normal blood cells to infer mosaic point mutations that are likely to have arisen during early embryogenesis. It provides preliminary insights into somatic mutation rates and processes that occur during early human embryogenesis and how these contribute to adult tissues.
8. Yuan Y*, **Ju YS***, Kim Y*, Li J, ... , & Liang H. Comprehensive molecular characterization of mitochondrial genomes in human cancers. *Nat Genet*. 2020;52(3):342-352. (JIF=30.8; Citation 282)
Exploring mitochondrial DNA alterations in cancers using 2,658 tumor whole-genome sequences.
✓ News & Views, *Nat Metabol*, "Cracking the enigma of mitochondrial-DNA variants and cancer"
- 9 **Ju YS***, Lee WC, Shin JY, Lee S, ... & Seo JS. A transforming KIF5B and RET gene fusion in lung adenocarcinoma revealed from whole-genome and transcriptome sequencing. *Genome Res*. 2012;22(3):436-45. (JIF=9.4; Citation 585)
First identification of KIF5B-RET fusion oncogene in lung adenocarcinoma using transcriptome sequencing.
10. Kim JI*, **Ju YS***, Park H, Kim S, ... & Seo JS. A highly annotated whole-genome sequence of a Korean individual. *Nature*. 2009 Aug 20;460(7258):1011-5. PMID:19587683 (JIF=64.8; Citation 388)
The fourth personal whole-genome sequenced by next-generation sequencing.

MAJOR GRANTS

- 2020-2029 **Leader Research Grant**, Nat'l Research Foundation of Korea
Advanced grants providing an opportunity to well-established and outstanding scientists to pursue innovative, high-risk research (Grant USD 6.6M for nine years)
- 2018-2023 **Suh Kyungbae Research Foundation** (SUHF) (<https://www.suhf.org/>)
Funding from Korean philanthropic foundation for young Korean scientists exploring new research areas in the field of biological science (Grant USD 2.25M for five years)
- 2018-2021 **HFSP Young Investigator Grants**, Human Frontier Science Program (<https://bit.ly/310I7v1>)
Funding for innovative biological research with novel and interdisciplinary approaches (Grant USD 1.05M for a team with BK Koo at IMBA and H Snippert at UMC Utrecht)
- 2017-2021 **MediStar Grants**, KHIDI, supported by Ministry of Health and Welfare of Korea
Consolidator grant for excellent biomedical scientists in Korea (Grant USD 1.1M for five years)

HONORS & AWARDS

- 2023 **The Kyung-Ahm's Prizes**, Kyung-Ahm Education & Culture Foundation
- 2023 **The 6th Hansung Science Awards**, Hansung Sohn Jaehan Foundation
- 2022 **The 1st Lim Sung-Ki Research Award** for Young Investigators, Lim Sung-Ki Foundation
- 2020 **Chen Award of Excellence**, Human Genome Organization (HUGO) (<https://bit.ly/3105GUj>)
- 2020 **Young Scientist Award**, Ministry of Science and ICT and Nat'l Research Foundation of Korea
- 2020 **Asan Award in Medicine** for Young Investigators, Asan Foundation (<https://bit.ly/3pa5Obk>)
- 2018 **Y-KAST** (Young Korean Academy of Science and Technology) (<https://y-kast.or.kr/en/>)
- 2018 **POSCO TJ Park Science Fellowship** for Young Assistant Professors (<https://bit.ly/3CTXaDa>)
- 2013 **EMBO Long Term Fellowship** for Postdoctoral Research (<https://bit.ly/3p8XNnC>)
- 2010 **Wunsch Medical Award** for Young Investigator, Korean Academy of Medical Sciences
- 2010 **Best Dissertation Award**, Korean Society for Molecular and Cellular Biology

FULL PUBLICATIONS *In a reverse chronological order*

A. CORRESPONDING AUTHOR ORIGINAL ARTICLES

14. Youk J, Kwon HW, Lim J, Kim E, Kim T, ..., Chang JH #, Kim KS#, Son TG, **Ju YS#**. Quantitative and qualitative mutational impact of ionizing radiation on normal cells. *Cell Genomics*. *In Press*. (preprint @ bioRxiv: <https://www.biorxiv.org/content/10.1101/2021.01.12.426324v1>)
13. Nam CH, Youk J, Kim JY, Lim J, Park JW, ..., Kwon HW#, Kim MJ#, **Ju YS#**. Extensive mosaicism by somatic L1 retrotransposition in normal human cells. *Nature*. 2023 May;617(7961):540-547. PMID:37165195
12. Kim T, Min KI, Yang J, Kim JW, Cho J, ..., Na KJ#, Lee J-Y, **Ju YS#**. Relative infectivity of the SARS-CoV-2 Omicron variant in human alveolar cells. *iScience*. 2022 Dec 22;25(12):105571. PMID:36406862
11. Yoon C, Kim S, Nam CH, Lee JH, Park JW, ..., Griffith OL #, **Ju YS#**. Estimation of intrafamilial DNA contamination in family trio genome sequencing using deviation from Mendelian inheritance. *Genome Res*. 2022 Nov-Dec;32(11-12):2134-2144. PMID:36617634
10. An J, Min KI, **Ju YS#**. Identifying Somatic Mitochondrial DNA Mutations. *Methods Mol Biol*. 2022;2493:153-165. doi: 10.1007/978-1-0716-2293-3_10. PMID:35751814
9. Park S, Mali NM, Kim R, Choi J-W, Lee J, ..., Oh JW#, **Ju YS#**. Clonal dynamics in early human embryogenesis inferred from somatic mutation. *Nature*. 2021 Sep;597(7876):393-397. PMID:34433967
8. Yi K, Kim SY, Bleazard T, Kim T, Youk J, , **Ju YS#**. Mutational spectrum of SARS-CoV-2 during the global pandemic. *Exp Mol Med*. (2020 IF=8.718) 2021 Aug;53(8):1229-1237. PMID:34453107
7. Youk J*, Kim T*, Evans KV*, Jeong Y-I*, Hur Y*, Hong SP*, ..., Kim YT#, Koh GY#, Choi B-S#, **Ju YS#**, Lee JH#. Three-dimensional human alveolar stem cell culture models reveal infection response to SARS-CoV-2. *Cell Stem Cell*. 2020 Dec 3;27(6):905-919. PMID:33142113
6. Youk J, An Y, Park S, Lee JK & **Ju YS#**. The genome-wide landscape of C:G>T:A polymorphism at the CpG contexts in the human population. *BMC Genomics*. 2020 Mar 30;21(1):270. PMID:32228436
5. Lee JS, An Y, Yoon CJ, Kim JY, Kim KH, ... , Lee EY# & **Ju YS#**. Germline gain-of-function mutation of STAT1 rescued by somatic mosaicism in immune dysregulation-polyendocrinopathy-enteropathy-X-linked-like disorder. *J Allergy Clin Immunol*. 2020 Mar;145(3):1017-1021. PMID:31805313
4. Lee JJ-K, Park S, Park H, Kim S, Lee J, ... , **Ju YS#** & Kim YT#. Tracing oncogene rearrangements in the mutational history of lung adenocarcinoma. *Cell*. 2019 Jun 13;177(7):1842-1857. PMID:31155235.
3. Lee J, Lee AJ, Lee JK, Park J, Kwon Y, ..., **Ju YS#** & Hong D#. MUTALISK: a web-based somatic MUTation AnaLysis toolKit for genomic, transcriptional and epigenomic signatures. *Nucleic Acids Res*. 2018 Jul 2;46(W1):W102-W108. PMID:29790943.
2. Lee JK, Lee J, Kim S, Kim S, Youk J, ..., Kim TM# & **Ju YS#**. Clonal history and genetic predictors of transformation into small cell carcinomas from lung adenocarcinomas. *Journal of Clinical Oncology* 2017 Sep 10;35(26):3065-3074. PMID:28498782
1. Lee JK, Louzada S, An Y, Kim SY, ..., Kim TM# & **Ju YS#**. Complex chromosomal rearrangements by single catastrophic pathogenesis in NUT midline carcinoma. *Ann Oncol*. 2017 Feb 14. 28(4):890-897. PMID:28203693

B. CORRESPONDING AUTHOR REVIEW ARTICLES

6. (Review) Youk J, Kwon HW, Kim R, **Ju YS#**. Cdissecting single-cell genomes through the clonal organoid technique. *Exp Mol Med*. (2020 IF=8.718) 2021 Oct;53(10):1503-1511. PMID:34663940
5. (Mini review) Kim T, Lee JS, **Ju YS#**. Experimental Models for SARS-CoV-2 Infection. *Mol Cells*. (2021 IF=5.034) 2021 Jun 30;44(6):377-383. PMID:34187969
4. (Spotlight) **Ju YS#**. A large-scale snapshot of intratumor heterogeneity in human cancer. *Cancer Cell*. 2021 Apr 12;39(4):463-465. PMID:33848477
3. (Review) Yi K and **Ju YS#**. Patterns and mechanisms of structural variations in human cancer. *Exp Mol Med*. (2020 IF=8.718) 2018 Aug 7;50(8):98. PMID:30089796.
2. (Review) **Ju YS#**, The mutational signatures and molecular alterations of bladder cancer. *Transl Cancer*

Res. 2017;6(Suppl 4):S689-S701

1. (Review) **Ju YS#**. Intracellular mitochondrial DNA transfers to the nucleus in human cancer cells. *Curr Opin Genet Dev.* 2016 Jun;38:23-30. PMID:27010587

C. FIRST AUTHOR PAPERS

12. Yuan Y*, **Ju YS***, Kim Y*, Li J, Wang J, ... , & Liang H. Comprehensive molecular characterization of mitochondrial genomes in human cancers. *Nat Genet.* 2020 Mar;52(3):342-352. PMID:32024997.
11. **Ju YS***, Martincorena I, Gerstung M, Petljak M, Alexandrov LB, ... & Stratton MR. Somatic mutations reveal asymmetric cellular dynamics in the early human embryo. *Nature.* 2017 Mar 30;543(7647):714-718. PMID:28329761
10. **Ju YS***, Tubio JM, Mifsud W, Fu B, Davies HR, ... & Stratton MR. Frequent somatic transfer of mitochondrial DNA into the nuclear genome of human cancer cells. *Genome Res.* 2015 Jun;25(6):814-24. PMID:25963125
9. **Ju YS***, Alexandrov LB, Gerstung M, Martincorena I, Nik-Zainal S, ... & Campbell PJ. Origins and functional consequences of somatic mitochondrial DNA mutations in human cancer. *eLife.* 2014 Oct 1;3. PMID:25271376
8. Seo JS*, **Ju YS***, Lee WC*, Shin JY, Lee JK, ..., Kim YT. The transcriptional landscape and mutational profile of lung adenocarcinoma. *Genome Res.* 2012 Nov;22(11):2109-19. PMID:22975805
7. **Ju YS***, Lee WC, Shin JY, Lee S, Bleazard T, ... & Seo JS. A transforming KIF5B and RET gene fusion in lung adenocarcinoma revealed from whole-genome and transcriptome sequencing. *Genome Res.* 2012 Mar;22(3):436-45. PMID:22194472
6. **Ju YS***, Kim JI*, Kim S*, Hong D, Park H, ... & Seo JS. Extensive genomic and transcriptional diversity identified through massively parallel DNA and RNA sequencing of eighteen Korean individuals. *Nat Genet.* 2011 Jul 3;43(8):745-52. PMID:21725310
5. Hong D*, Park SS*, **Ju YS***, Kim S, Shin JY, ... & Seo JS. TIARA: a database for accurate analysis of multiple personal genomes based on cross-technology. *Nucleic Acids Res.* 2011 Jan;39(Database issue):D883-8. PMID:21051338
4. **Ju YS***, Hong D*, Kim S*, Park SS, Kim S, ... & Seo JS. Reference-unbiased copy number variant analysis using CGH microarrays. *Nucleic Acids Res.* 2010 Nov;38(20):e190. PMID:20802225
3. Park H*, Kim JI*, **Ju YS***, Gokcumen O, Mills RE, ... & Seo JS. Discovery of common Asian copy number variants using integrated high-resolution array CGH and massively parallel DNA sequencing. *Nat Genet.* 2010 May;42(5):400-5. PMID:20364138
2. Kim JI*, **Ju YS***, Park H, Kim S, Lee S, ... & Seo JS. A highly annotated whole-genome sequence of a Korean individual. *Nature.* 2009 Aug 20;460(7258):1011-5. PMID:19587683
1. **Ju YS***, Park H, Lee MK, Kim JI, Sung J, ... & Seo JS. A genome-wide Asian genetic map and ethnic comparison: the GENDISCAN study. *BMC Genomics.* 2008 Nov 25;9:554. PMID:19025666

D. CONTRIBUTING AUTHOR PAPERS

55. Bruzos AL, Santamarina M, García-Souto D, Díaz S, Rocha S, ..., Tubio JMC. Somatic evolution of marine transmissible leukemias in the common cockle, *Cerastoderma edule*. *Nature Cancer.* 2023. Online ahead of Print (Oct 02 2023)
54. Cho S, Park S, Lee JS, **Ju YS**, Choi YJ, Lee S. Adenosine Deaminase 2 Deficiency Caused by Biallele Variants Including Splicing Variant: The First Case in Korea. *J Rheum Dis.* 2022 Oct 1;29(4):254-260. PMID:37476427
53. Jikuya R, Johnson TA, Maejima K, An J, **Ju YS**, ... , Hasumi H. Comparative analyses define differences between BHD-associated renal tumour and sporadic chromophobe renal cell carcinoma. *EBioMedicine.* 2023 Jun;92:104596. PMID:37182269
52. Hong Y, Kim HJ, Park S, Yi S, Lim MA, ..., Kang YE. Single Cell Analysis of Human Thyroid Reveals the Transcriptional Signatures of Aging. *Endocrinology.* 2023 Feb 11;164(4):bqad029. PMID: 36791033
51. Kim JH, Park JH, Lee J, Park JW, Kim HJ, ..., Lee JH. Ultra-Low Level Somatic Mutations and Structural Variations in Focal Cortical Dysplasia Type II. *Ann Neurol.* 2023 Jun;93(6):1082-1093. PMID:36700525

50. Sohn JI, Choi MH, Yi D, Menon VA, Kim YJ, ..., Nam JW. Ultrafast prediction of somatic structural variations by filtering out reads matched to pan-genome k-mer sets. *Nat Biomed Eng.* 2023 Jul;7(7):853-866. PMID:36536253
49. Lomakin A, Svedlund J, Strell C, Gataric M, Shmatko A, ..., Yates LR. Spatial genomics maps the structure, nature and evolution of cancer clones. *Nature.* 2022 Nov;611(7936):594-602. PMID:36352222
48. Park CS, Kim SH, Yang HY, Kim JH, Schermuly RT, ..., Kim I. Sox17 Deficiency Promotes Pulmonary Arterial Hypertension via HGF/c-Met Signaling. *Circ Res.* 2022 Oct 28;131(10):792-806. PMID:36205124
47. Johnson TA, Maekawa S, Fujita M, An J, **Ju YS**, ..., Nakagawa H. Genomic features of renal cell carcinoma developed during end-stage renal disease and dialysis. *Hum Mol Genet.* 2023 Jan 6;32(2):290-303. PMID:35981075
46. Kim IB, Lee T, Lee J, Kim J, Lee S, ..., Lee JH. Non-coding de novo mutations in chromatin interactions are implicated in autism spectrum disorder. *Mol Psychiatry.* 2022 Nov;27(11):4680-4694. PMID:35840799
45. Jeong D, Kim SM, Min BJ, Kim JH, **Ju YS**, ..., Lee DS. Heterogeneous genetic landscape of congenital neutropenia in Korean patients revealed by whole exome sequencing: genetic, phenotypic and histologic correlations. *Sci Rep.* 2022 May 7;12(1):7515. PMID:35525891
44. Lee JH, Kim S, Han S, Min J, Caldwell B, ..., Kim JK. p57Kip2 imposes the reserve stem cell state of gastric chief cells. *Cell Stem Cell.* 2022 May 5;29(5):826-839.e9. PMID:35523142
43. Seo J, Kim H, Min KI, Kim C, Kwon Y, ..., Kim J. Weight-bearing activity impairs nuclear membrane and genome integrity via YAP activation in plantar melanoma. *Nat Commun.* 2022 Apr 25;13(1):2214. PMID:35468978
42. Kwon SG, Bae GH, Choi JH, Mali NM, Jun MS, ..., Oh JW. Asymmetric Contribution of Blastomere Lineages of First Division of the Zygote to Entire Human Body Using Post-Zygotic Variants. *Tissue Eng Regen Med.* 2022 Aug;19(4):809-821. PMID:35438457
41. Cho YW, Min DW, Kim HP, An Y, Kim S, ..., Kim TY. Patient-derived organoids as a preclinical platform for precision medicine in colorectal cancer. *Mol Oncol.* 2022 Jun;16(12):2396-2412. PMID:34850547
40. Song U, Ryu YH, Hong K, Shim SY, Park S, ..., Lee S. Severe protein C deficiency in a newborn caused by a homozygous pathogenic variant in the PROC gene: a case report. *BMC Pediatr.* 2021 Oct 16;21(1):453. PMID:34654403
39. Lee JS, Koh JY, Yi K, Kim YI, Park SJ, ..., Park SH. Single-cell transcriptome of bronchoalveolar lavage fluid reveals sequential change of macrophages during SARS-CoV-2 infection in ferrets. *Nat Commun* 2021 Jul 27;12(1):4567. doi: 10.1038/s41467-021-24807-0. PMID:34315893
38. Park HR, Kim TM, Lee Y, Kim S, Park S, ..., Heo DS. Acquired resistance to third-generation EGFR tyrosine kinase inhibitors in patients with de novo EGFR790M-mutant non-small cell lung cancer. *J Thorac Oncol.* 2021 Jul 6:S1556-0864(21)02254-1. Online ahead of print. PMID:34242789
37. Ahn JH, Kim J, Hong SP, Choi SY, Yang MJ, ..., Koh GY. Nasal ciliated cells are primary targets for SARS-CoV-2 replication in the early stage of COVID-19. *J Clin Invest.* 2021 Jul 1;131(13):e148517. PMID:34003804
36. Park S, Lee HY, Kim J, Park H, **Ju YS**, Kim EG, Kim J. Cerebral Cavernous Malformation 1 Determines YAP/TAZ Signaling-Dependent Metastatic Hallmarks of Prostate Cancer Cells. *Cancers (Basel).* 2021 Mar 5;13(5):1125. PMID:33807895
35. Park JH, Kim HJ, Kim CW, Kim HC, Jung Y, ..., Lee HK. Tumor hypoxia represses $\gamma\delta$ T cell-mediated antitumor immunity against brain tumors. *Nat Immunol.* 2021 Mar;22(3):336-346. PMID:33574616
34. Kim HD, Jeong S, Park S, Lee YJ, **Ju YS**, ..., Park SH. Implication of CD69⁺ CD103⁺ tissue-resident-like CD8⁺ T cells as a potential immunotherapeutic target for cholangiocarcinoma. *Liver Int.* 2021 Apr;41(4):764-776. PMID:33548061
33. Leem G, Park J, Jeon M, Kim ES, Kim SW, ..., Park SH. 4-1BB co-stimulation further enhances anti-PD-1-mediated reinvigoration of exhausted CD39⁺ CD8 T cells from primary and metastatic sites of epithelial ovarian cancers. *J Immunother Cancer.* 2020 Dec;8(2):e001650. PMID:33335029
32. Bae K, Kim JH, Jung H, Kong SY, ..., Yoon KA. A fusion of CD63-BCAR4 identified in lung adenocarcinoma promotes tumorigenicity and metastasis. *Br J Cancer.* 2021 Jan;124(1):290-298. PMID:33204025
31. Lee JS, Yi K, **Ju YS**, Shin EC. Effects of Cryopreservation and Thawing on Single-Cell Transcriptomes of

- Human T Cells. *Immune Netw.* 2020 Jul 15;20(4):e34. PMID:32895621
30. Yang JM, Park CS, Kim SH, Noh TW, Kim JH, ... , **Ju YS** & Kim I. Dll4 suppresses transcytosis for arterial blood-retinal barrier homeostasis. *Circ Res.* 2020 Mar 13;126(6):767-783. PMID:32078435.
 29. Rodriguez-Martin B, Alvarez EG, Baez-Ortega A, Zamora J, Supek F, ... , Tubio JMC. Pan-cancer analysis of whole genomes identifies driver rearrangements promoted by LINE-1 retrotransposition. *Nat Genet.* 2020 Mar;52(3):306-319. PMID:32024998.
 28. Kim H, Yoon BH, Oh CM, Lee J, Lee K, ... , & Kim H. PRMT1 is required for the maintenance of mature beta cell identity. *Diabetes.* 2020 Mar;69(3):355-368. PMID:31848151.
 27. Kim H, Lee AJ, Lee J, Chun H, **Ju YS** & Hong D. FIREVAT: finding reliable variants without artifacts in human cancer samples using etiologically relevant mutational signatures. *Genome Med.* 2019 Dec 17;11(1):81. PMID:31847917.
 26. Kim HD, Park S, Jeong S, ... , & Park SH. 4-1BB delineates distinct activation status of exhausted tumor-infiltrating CD8⁺ T cells in hepatocellular carcinoma. *Hepatology.* 2020 Mar;71(3):955-971. PMID:31353502.
 25. Petljak M, Alexandrov LB, Brammeld JS, Price S, Wedge DC, ... , & Stratton MR. Characterizing mutational signatures in human cancer cell lines reveals episodic APOBEC mutagenesis. *Cell.* 2019 Mar 7;176(6):1282-1294. PMID:30849372.
 24. Park ES, Lee AR, Kim DH, Lee JH, Yoo JJ, & Kim KH. Identification of a quadruple mutation that confers tenofovir resistance in chronic hepatitis B patients. *J Hepatol.* 2019 Jun;70(6):1093-1102. PMID:30794889.
 23. Choi W, Namkung J, Hwang I, Kim H, Lim A, ... & Kim H. Serotonin signals through a gut-liver axis to regulate hepatic steatosis. *Nat Commun.* 2018 Nov 16;9(1):4824. PMID:30446669.
 22. Kim HD, Song GW, Park S, Jung MK, Kim MH, ... & Park SH. Association between expression level of PD1 by tumor-infiltrating CD8⁺ T cells and features of hepatocellular carcinoma. *Gastroenterology.* 2018 Dec;155(6):1936-1950. PMID:30145359.
 21. Lee JH, Lee JE, Kahng JY, Kim SH, Park JS, ... & Lee JH. Human glioblastoma arises from subventricular zone cells with low-level driver mutations. *Nature.* 2018 Aug;560(7717):243-247. PMID:30069053.
 20. Cho SY, Sung CO, Chae J, Lee J, Na D, ..., & Lee C. Alterations in the Rho pathway contribute to Epstein-Barr virus-induced lymphomagenesis in immunosuppressed environments. *Blood.* 2018 Apr 26;131(17):1931-1941. PMID:29475961.
 19. Kim IK, KIM K, Lee E, Oh DS, Park CS, .., & Kim I. Sox7 promotes high-grade glioma by increasing VEGFR2-mediated vascular abnormality. *J Exp Med.* 2018. Mar 5;215(3):963-983. PMID:29444818.
 18. Alexandrov LB, **Ju YS**, Haase K, Van Loo P, Martincorena I, ... & Stratton MR. Mutational signatures associated with tobacco smoking in human cancer. *Science.* 2016 Nov 4;354(6312):618-622. PMID:27811275
 17. Shlien A, Raine K, Fuligni F, Arnold R, Nik-Zainal S, ... & Campbell PJ. Direct transcriptional consequences of somatic mutation in breast cancer. *Cell Rep.* 2016 Aug 16;16(7):2032-46. PMID:27498871
 16. Kemper K, Krijgsman O, Kong X, Cornelissen-Steijger P, Shahrabi A, ... & Peeper DS. BRAF(V600E) Kinase domain duplication identified in therapy-refractory melanoma patient-derived xenografts. *Cell Rep.* 2016 Jun 28;16(1):263-77. PMID:27320919
 15. Nik-Zainal S, Davies H, Staaf J, Ramakrishna M, Glodzik D, ... & Stratton MR. Landscape of somatic mutations in 560 breast cancer whole-genome sequences. *Nature.* 2016 Jun 2;534(7605):47-54. PMID:27135926
 14. Kim J, Kim S, Ko S, In YH, Moon HG, ... & Han W. Recurrent fusion transcripts detected by whole-transcriptome sequencing of 120 primary breast cancer samples. *Genes Chromosomes Cancer.* 2015 Nov;54(11):681-91. PMID:26227178
 13. Yates LR, Gerstung M, Knappskog S, Desmedt C, Gundem G, ... & Campbell PJ . Subclonal diversification of primary breast cancer revealed by multiregion sequencing. *Nat Med.* 2015 Jul;21(7):751-9. PMID:26099045
 12. Tubio JM, Li Y, **Ju YS**, Martincorena I, Cooke SL, ... & Campbell PJ. Extensive transduction of nonrepetitive DNA mediated by L1 retrotransposition in cancer genomes. *Science.* 2014 Aug 1;345(6196):1251343. PMID:25082706
 11. Go H, Jung YJ, Kang HW, Park IK, Kang CH, ... & Kim YT. Diagnostic method for the detection of KIF5B-RET transformation in lung adenocarcinoma. *Lung Cancer.* 2013 Oct;82(1):44-50. PMID:23932363

10. Kim HJ, Yoo YJ, **Ju YS**, Lee S, Cho SI, ... & Seo JS. Combined linkage and association analyses identify a novel locus for obesity near PROX1 in Asians. *Obesity*. 2013 Nov;21(11):2405-12. PMID:23818313
9. Hong D, Lee J, Bleazard T, Jung H, **Ju YS**, ... & Seo JS. TIARA genome database: update 2013. *Database (Oxford)*. 2013 Mar 20;2013:bat003. PMID:23515433
8. Bleazard T, **Ju YS**, Sung J & Seo JS. Fine-scale mapping of meiotic recombination in Asians. *BMC Genet*. 2013 Mar 8;14:19. PMID:23510153
7. Park H, Kim HJ, Lee S, Yoo YJ, **Ju YS**, ... & Seo JS. A family-based association study after genome-wide linkage analysis identified two genetic loci for renal function in a Mongolian population. *Kidney Int*. 2013 Feb;83(2):285-92. PMID:23254893
6. Park H, Lee S, Kim HJ, **Ju YS**, Shin JY, ... & Seo JS. Comprehensive genomic analyses associate UGT8 variants with musical ability in a Mongolian population. *J Med Genet*. 2012 Dec;49(12):747-52. PMID:23118445
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INVITED TALKS *In a chronological order*

1. Feb.6.2017. **HGM2017, Human Genome Organization (HUGO)**, Barcelona, Spain
Title: Somatic mutations reveal asymmetric cellular dynamics in the early human embryo
2. Jun.14.2017. **The 13th International Cancer Genome Consortium Workshop, ICGC**, Seoul, Korea
Title: First somatic mutations reveal asymmetric cellular dynamics in the early human embryo
3. Aug.10.2017. **US-Korea Conference 2017, Korean American Scientists and Engineers Association**, Washington DC, USA
Title: First somatic mutations reveal asymmetric cellular dynamics in the early human embryo
4. Sep.30.2017. **The 76th Annual Meeting of the Japanese Cancer Association, Japanese Cancer Association**, Yokohama, Japan
Title: Clonal history & genetic predictors of transformation into small cell carcinomas from lung adenocarcinoma
5. Dec.1.2017. **The 1st Human Cell Atlas Asia Meeting, Human Cell Atlas Asian Consortium**, Okinawa, Japan
Title: Somatic mutations in single cells
6. May.17.2018. **The 14th International Cancer Genome Consortium Scientific Workshop, ICGC**, Paris, France
Title: Architecture of genomic rearrangements molding fusion oncogenes in lung adenocarcinomas
7. Feb.26.2019. **Cell Symposia; Single Cells: Technology to Biology, A*STAR**, Singapore
Title: Single genome sequencing of normal cells reveals developmental lineage trees of early human embryogenesis
8. Apr.4.2019. **IASLC Small Cell Lung Cancer Workshop, International Association for the Study of Lung Cancer**. MSKCC, New York, USA
Title: Transformation of lung adenocarcinoma to SCLC
9. Apr.30.2019. **International Precision Medicine Symposium, Xian Jiaotong Univ.**, Xian, China
Title: Tracing genomic rearrangements in the mutational history of lung adenocarcinomas

10. Jul.18.2019 **Europe-Korea Conference on Science and Technology, Vienna University of Economics and Business**, Vienna, Austria
Title: Tracing genomic rearrangements in the mutational history of lung adenocarcinomas
11. Aug.8.2019. **Jackson Laboratory Seminar, Jackson Lab for Genomic Medicine**, Farmington, CT, USA
Title: Tracing genomic rearrangements in the mutational history of lung adenocarcinomas
12. Sep.26.2019. **The 78th Annual Meeting of the Japanese Cancer Association, Japanese Cancer Association**, Kyoto, Japan.
Title: Tracing genomic rearrangements in the mutational history of lung adenocarcinomas
13. Sep.30.2022. **The 81st Annual Meeting of the Japanese Cancer Association, Japanese Cancer Association**, Yokohama, Japan.
Title: Extensive mosaicism by somatic L1 retrotransposition in normal human cells
14. Oct.26.2022. **American Society of Human Genetics Annual Meeting 2022, ASHG, Los Angeles, USA**
Title: Extensive mosaicism by somatic L1 retrotransposition in normal human cells
15. Sep.21.2023. **The 82nd Annual Meeting of the Japanese Cancer Association, Japanese Cancer Association**, Yokohama, Japan.
Title: Extensive mosaicism by somatic L1 retrotransposition in normal human cells
16. Oct.28.2023. **The 127th Titisee Conference, Boehringer Ingelheim Fonds**, Titisee, Germany.
Title: Extensive mosaicism by somatic L1 retrotransposition in normal human cells
17. Nov.17.2023. **EMBL Conference: Cancer Genomics**, Heidelberg, Germany.
Title: Extensive mosaicism by somatic L1 retrotransposition in normal human cells