
질병관리청
국립보건연구원
국립중앙인체자원은행
NATIONAL BIOBANK OF KOREA
KBN
한국인체자원은행네트워크
Korea Biobank Network
2026년 한국인체자원은행사업
KOREA BIOBANK PROJECT
KBP
심포지엄
2026. 11. 24 화
보코 서울 명동

2026년 한국인체자원은행사업

KBP심포지엄

KOREABIOBANKPROJECT

KBP 5기 특성화 사업 참여 인체자원은행 홍보 영상 YouTube

가톨릭대학교 서울성모병원
THE CATHOLIC UNIV. OF KOREA SEOUL ST. MARY'S HOSPITAL

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KYUNGPOOK NATIONAL UNIVERSITY HOSPITAL

SNUH 서울대학교병원

서울대학교치과병원
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SCH 순천향대학교 부천병원
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2026년 한국인체자원은행사업

KBP심포지엄

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PROGRAM

시간		내용	발표자
11:00~13:00	120분	등록	
Opening (유공자 포상 및 기조강연)			진행 : **신은아** 교수 (용인세브란스병원)
13:00~13:05	5분	개회사	**남재환** 원장 (국립보건연구원)
13:05~13:10	5분	환영사	**임승관** 청장 (질병관리청)
13:10~13:15	5분	축사	**박경신** 회장 (KBP 협의회)
13:15~13:35	20분	KBP 유공자 포상 및 기념촬영	바이오뱅크과
13:35~14:05	30분	기조강연 The Next Chapter of Biobanking in the AI and Precision Medicine Era	**송시영** 교수 (연세대학교)
Session Ⅰ (인체자원 활용성과 우수사례)			좌장 : **김동기** 교수 (서울대병원)
14:05~14:15	10분	인체 유래물을 이용한 중개 연구 사례	**김송철** 교수 (서울아산병원)
14:15~14:25	10분	바이오뱅크 데이터를 활용한 한국인 질병 유전체 연구	**이승근** 교수 (서울대학교)
14:25~14:35	10분	육종 연구를 위한 혁신형 바이오뱅킹 사업	**김준혁** 교수 (국립암센터)
14:35~14:40	5분	질의응답	-
14:40~15:10	30분	**휴식 (포스터 발표 및 네트워킹)**	
Session Ⅱ (인체자원은행 운영 및 성과 사례)			좌장 : **차장규** 교수 (순천향대부천병원)
15:10~15:20	10분	간암·위암·폐암 기반 개인맞춤형의료 및 신약스크리닝을 위한 바이오뱅크 플랫폼 구축	**정재욱** 교수 (충남대병원)
15:20~15:30	10분	정상조직 분야 혁신형 바이오뱅크 컨소시엄 운영	**김민규** 교수 (연세대학교)
15:30~15:40	10분	치매뇌은행 구축운영 및 뇌부검 신경병리 연구	**원재경** 교수 (서울대병원)
15:40~15:45	5분	질의응답	-
Session Ⅲ (패널 토의 (KBN CDM 기반 데이터 활용 확대 전략))			진행 : **김광수** 교수 (서울대병원)
15:45~16:05	20분	KBN CDM 기반 데이터 활용 확대 전략	**최인영** 교수 (가톨릭대학교)
16:05~16:40	35분	**패널 토의 및 질의 응답** 패널 : 최인영 교수 (가톨릭대학교), 김종덕 단장 (한국보건의료정보원), 양광모 교수 (삼성서울병원), 박래웅 교수 (아주대의료원), 정지원 과장 (국립보건연구원)	
Session Ⅳ (국가인체자원은행 거버넌스 현황)			좌장 : **홍승모** 교수 (서울아산병원)
16:40~16:50	10분	바이오뱅크 데이터 활용 활성화를 위한 입법적 쟁점 - 법령 간 정합성 논의를 중심으로 -	**김재선** 교수 (동국대학교)
16:50~17:00	10분	시체해부 및 보존 등에 관한 법률 개정의 이해	**문서현** 주무관 (보건복지부)
17:00~17:10	10분	인체유래물 허가·관리 제도 운영 현황	**이애정** 연구사 (국립보건연구원)
17:10~17:15	5분	질의응답	-
17:15~17:25	10분	**우수 포스터 시상 / 경품추첨**	
17:25~17:30	5분	폐회사	**전재필** 부장 (국립보건연구원)

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2026년 한국인체자원은행사업

KBP심포지엄 KOREABIOBANKPROJECT

Session 1

인체 유래물을 이용한 중개 연구 사례

서울아산병원
김송철 교수

인체 유래물을 이용한 중개 연구 사례

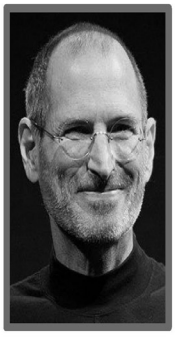
: Facing Pancreatic Cancer

김송철

울산대학교 의과 대학 및 서울아산 병원 간담췌외과

Famous Figures Who Lost Their Lives to Pancreatic Cancer



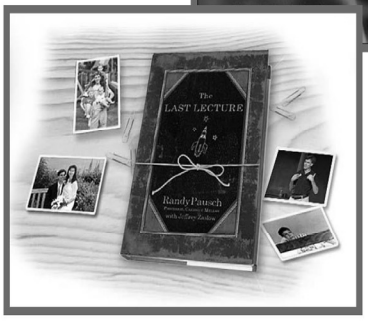












Survival of Pancreatic Cancer patients

OPEN ACCESS Freely available online PLOS MEDICINE

Preoperative/Neoadjuvant Therapy in Pancreatic Cancer: A Systematic Review and Meta-analysis of Response and Resection Percentages

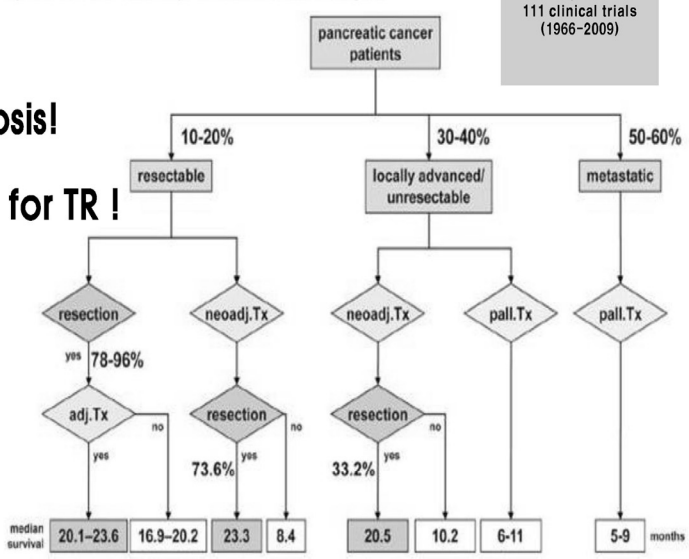
PLoS Med. 2010 Apr 20;7(4):e1000267.

Sonja Gillen[1], Tibor Schuster[2], Christian Meyer zum Büschenfelde[3], Helmut Friess[1], Jörg Kleeff[1,4]

4394 patients
111 clinical trials
(1966–2009)

Early Diagnosis!

Tissue is Issue for TR !



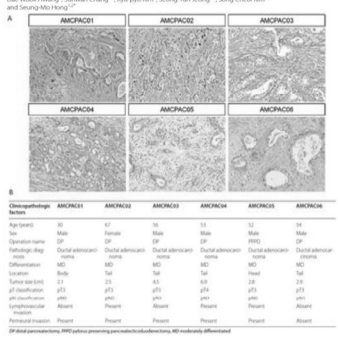
Utilization of a Theragnostic Platform

Patient Derived Cell line (PDC)

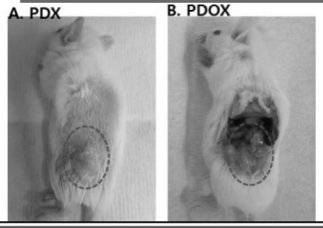
Cancer Cell International

PRIMARY RESEARCH Open Access

Establishment and characterization of 6 novel patient-derived primary pancreatic ductal adenocarcinoma cell lines from Korean pancreatic cancer patients



Patient Derived Xenograft (PDX)



www.impactjournals.com/oncotarget/ Oncotarget, 2018, Vol. 9, (No. 8), pp: 7867-7881

Research Paper

Genetic and metabolic comparison of orthotopic and heterotopic patient-derived pancreatic-cancer xenografts to the original patient tumors

Eunsung Jun[1,2], Seung-Mo Hong[3], Hyun Ju Yoo[4], Moon-Bo Kim[5], Ji Sun Won[1], Soyeon An[1], In Kyong Shim[6], Suhwan Chang[2], Robert M. Hoffman[7,8] and Song Cheol Kim[1]

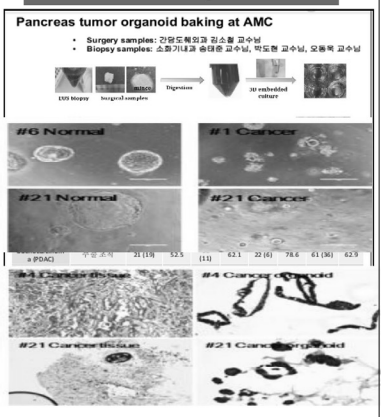
www.impactjournals.com/oncotarget/ Oncotarget, 2017, Vol. 8, (No. 25), pp: 40140-40151

Research Paper

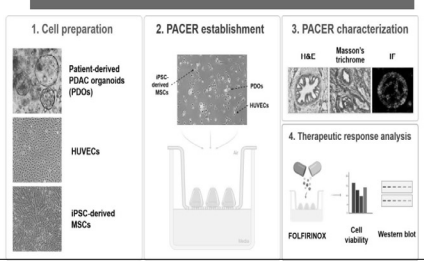
Locally-applied 5-fluorouracil-loaded slow-release patch prevents pancreatic cancer growth in an orthotopic mouse model

In Kyong Shim[1,*], Hye-Jin Yi[1,*], Hee-Gyeong Yi[2], Chan Mi Lee[1], Yu Na Lee[1], Yeong-Jin Choi[3], Seong-Yun Jeong[1], Eunsung Jun[4,5], Robert M. Hoffman[6,7], Dong-Woo Cho[2] and Song Cheol Kim[5]

Patient Derived Organoid (PDO)



Multicellular Platform Reconstituting Patient-derived Organoids and iPSC-derived Stroma

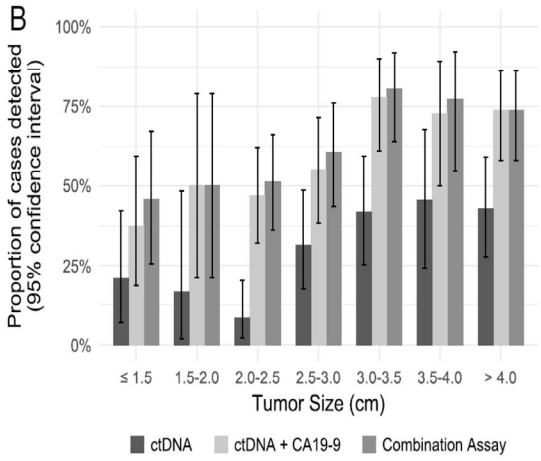


Early Diagnosis

Combined circulating tumor DNA and protein biomarker-based liquid biopsy for the earlier detection of pancreatic cancers

Joshua D. Cohen[a,b,c,d,e], Ammar A. Javed[f], Christopher Thoburn[c], Fay Wong[a,b,c,d], Jeanne Tie[g,h,i], Peter Gibbs[g,h,i], C. Max Schmidt[j,k], Michele T. Yip-Schneider[j], Peter J. Allen[l], Mark Schattner[m], Randall E. Brand[n], Aatur D. Singhi[o], Gloria M. Petersen[p], Seung-Mo Hong[q], Song Cheol Kim[r], Massimo Falconi[s], Claudio Doglioni[s], Matthew J. Weiss[f],

10202–10207 | PNAS | September 19, 2017 | vol. 114 | no. 38



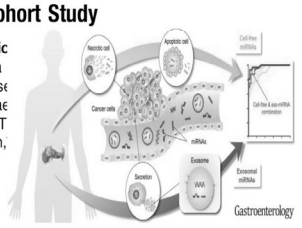
any single marker. The cohort tested included 221 patients with resectable pancreatic ductal adenocarcinomas and 182 control patients without known cancer. *KRAS* mutations were detected in the plasma of 66 patients (30%), and every mutation found in the plasma was identical to that subsequently found in the patient's primary tumor (100% concordance). The use of *KRAS* in conjunction with four thresholded protein biomarkers increased the sensitivity to 64%. Only one of the 182 plasma samples from the control cohort was positive for any of the DNA or protein biomarkers (99.5% specificity). This combinatorial approach may prove useful for the earlier detection of many cancer types.

Early Diagnosis

Gastroenterology 2022;163:1252–1266

An Exosome-based Transcriptomic Signature for Noninvasive, Early Detection of Patients With Pancreatic Ductal Adenocarcinoma: A Multicenter Cohort Study

Kota Nakamura,[1,*] **Zhongxu Zhu**,[1,2,3,*] **Souvic** Ruben M. Munoz,[5] Satoshi Nishiwada,[1] Geeta Frederic Zenhausern,[6] Seungchan Kim,[7] Denise Douglas B. Evans,[11] Suguru Yamada,[12] Michae Scott A. Celinski,[13] Erkut Borazanci,[14] Susan T John S. Bolton,[16] Xin Wang,[2] Song Cheol Kim,

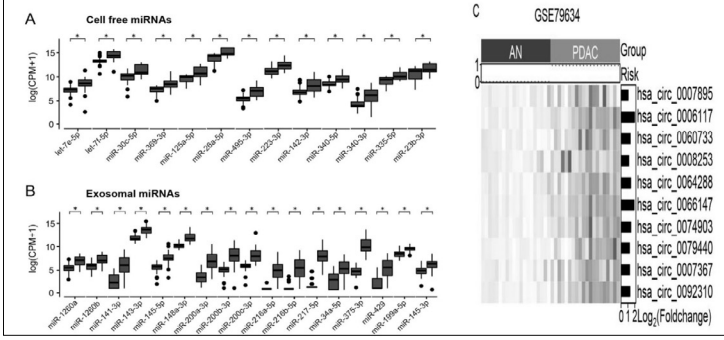


Gastroenterology 2024;166:178–190

PREVENTION AND EARLY DETECTION

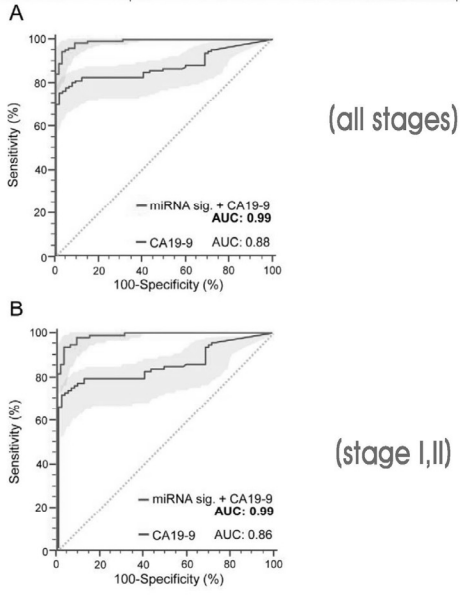
A Circulating Panel of circRNA Biomarkers for the Noninvasive and Early Detection of Pancreatic Ductal Adenocarcinoma

Caiming Xu,[1,2,3] Eunsung Jun,[4] Yoshinaga Okugawa,[5] Yuji Toiyama,[5] Erkut Borazanci,[6] John Bolton,[7] Akinobu Taketomi,[8] Song Cheol Kim,[4] Dong Shang,[2,3] Daniel Von Hoff,[9] Guixin Zhang,[2,3] and Ajay Goel[1,10]



FDP Foreign Fixed Rate Subaward

Federal Awarding Agency:	National Institutes of Health (NIH)
Pass-Through Entity (PTE):	Subrecipient:
Beckman Research Institute of the City of Hope	Asan Medical Center
PTE PI: Ajay Goel	Sub PI: Song Cheol Kim
PTE Federal Award No: U01CA214254-07	Subaward No: 64787.2006995.669311
Project Title: Exosome-based microRNA biomarkers for Non-invasive and Early Detection of Pancreatic Cancer	
Subaward Budget Period: Start: 05/01/2025 End: 07/31/2025	Amount Funded This Action (USD): $ 27,050.00



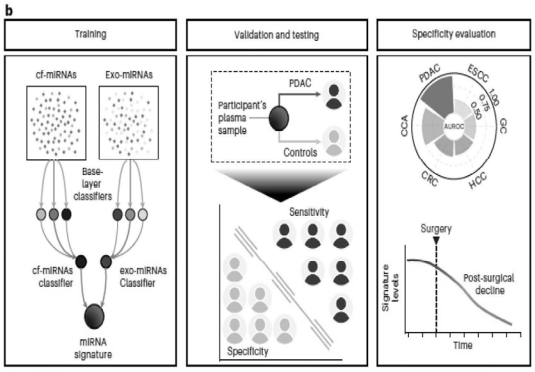
nature medicine

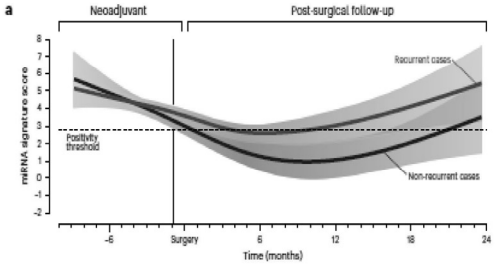
Article

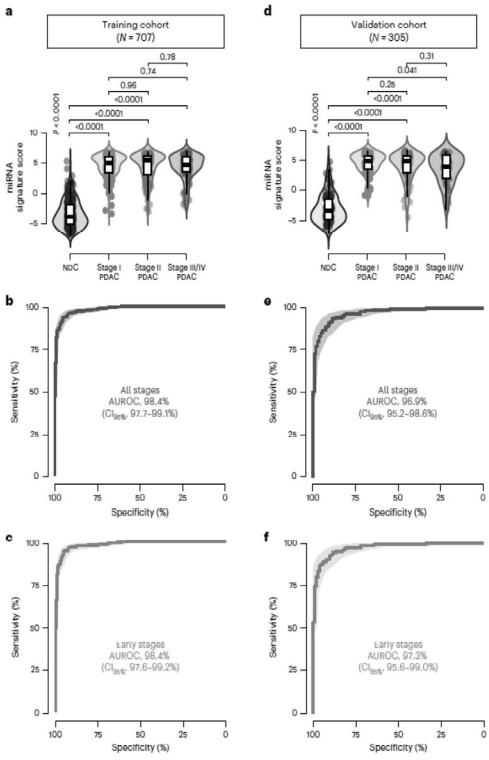
https://doi.org/10.1038/s41591-026-04625-x

Liquid biopsy for early detection of pancreatic ductal adenocarcinoma

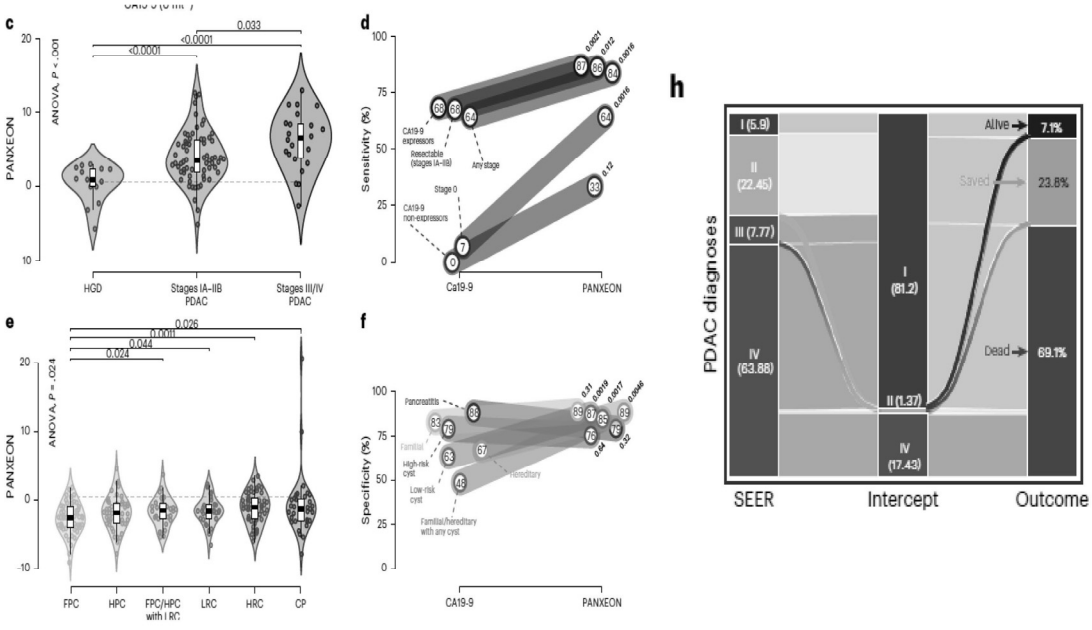
Nature Medicine | Volume 32 | October 2026 | 3781–3791







PANXEON, a combined miRNA signature and CA19-9 score



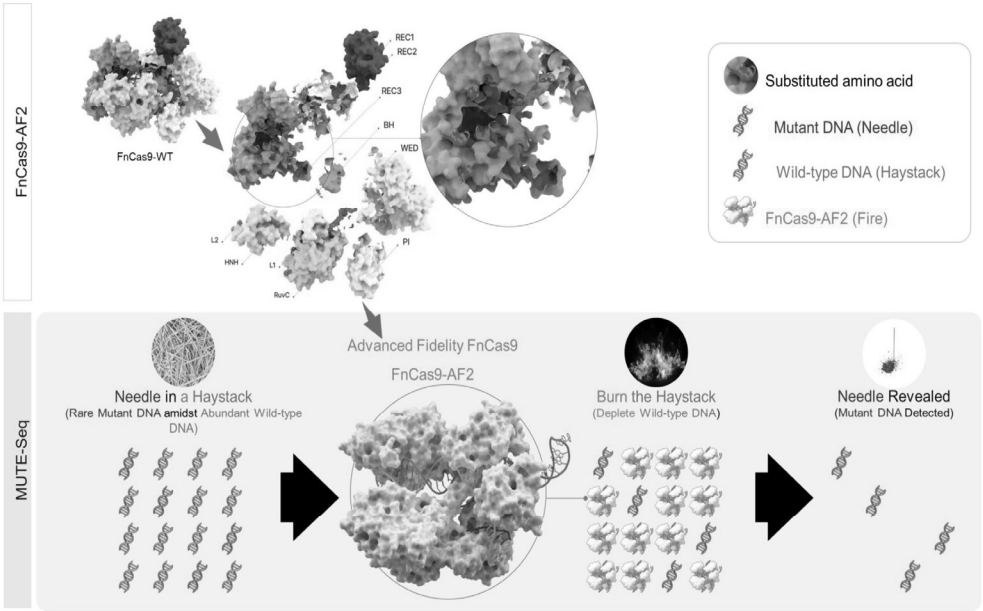
Early Diagnosis

ADVANCED MATERIALS
Research Article | Open Access

MUTE-Seq: An Ultrasensitive Method for Detecting Low-Frequency Mutations in cfDNA With Engineered Advanced-Fidelity FnCas9

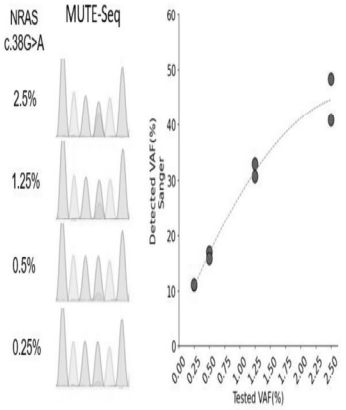
Sunghyeok Ye, Jin-Soo Kim, Myungshin Kim, Ki-Yeon Kim, Yoon-Ho Won, Taegun Park, Sungjae An, Haerin Jeong, Hee-joon Chung, In Seon Lee, Myoung-Hee Kang, Chan Young Kang, Mi Young Kim, Jae Ho Chung, Jeong-An Gim, Woochang Hwang, Yonggoo Kim, Song Cheol Kim, Sungho Lee

- high-precision CRISPR genome editor FnCas9(Francisella novicida Cas9) -AF2 development
- MUTE-Seq : Mutation tagging by CRISPR-based Ultra-precise Targeted Elimination in Sequencing metho

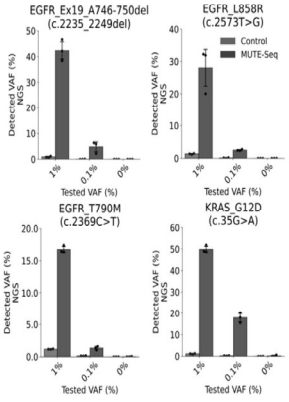


MUTE-Seq performance

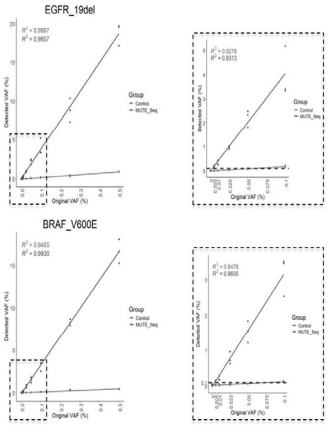
A 40-fold increase in the detection limit of Sanger sequencing (0.25% → 10%)

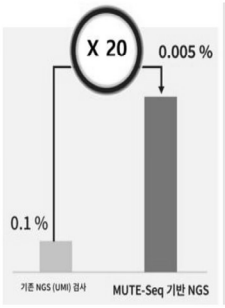


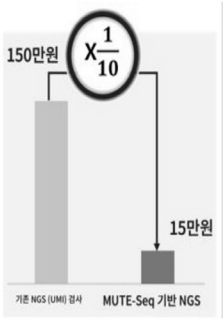
multiplexed amplification



Analytical limit of detection (LOD) : 0.005%

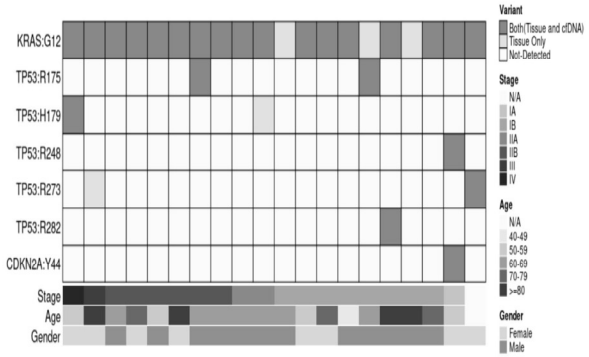




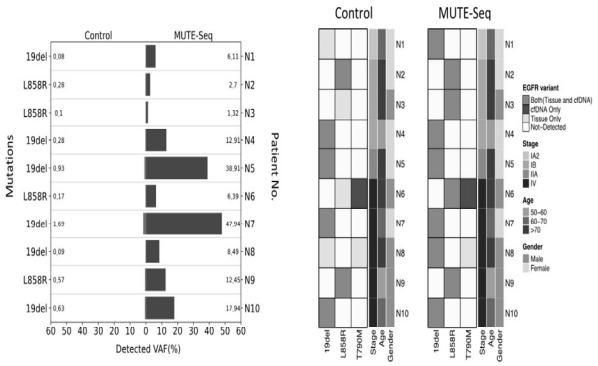


Performance validation for early detection of pancreatic and lung cancers

Stage I/II pancreatic cancer (n=20): accuracy 93%, sensitivity 82.8%, specificity 100%



Non-small cell lung cancer (n=20): concordance 93.3%, sensitivity 90.9%, specificity 94.7%.



CRISPR-Based Integrated Proteogenomic Diagnostic Technology for Pancreas and Lung Cancer Early Detection

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대한민국 의학한림원 대한 암연구 재단 암연구지원사업

초정밀 유전자가위 기반 단백체-유전체 통합 진단기술을 이용한 췌장암 및 폐암의 조기진단 성능 규명 및 임상 유효성 평가

CRISPR-Based Integrated Proteogenomic Diagnostic Technology for Pancreas and Lung Cancer Early Detection

주관연구책임자 | 김송철 | 서울아산병원 간담췌외과

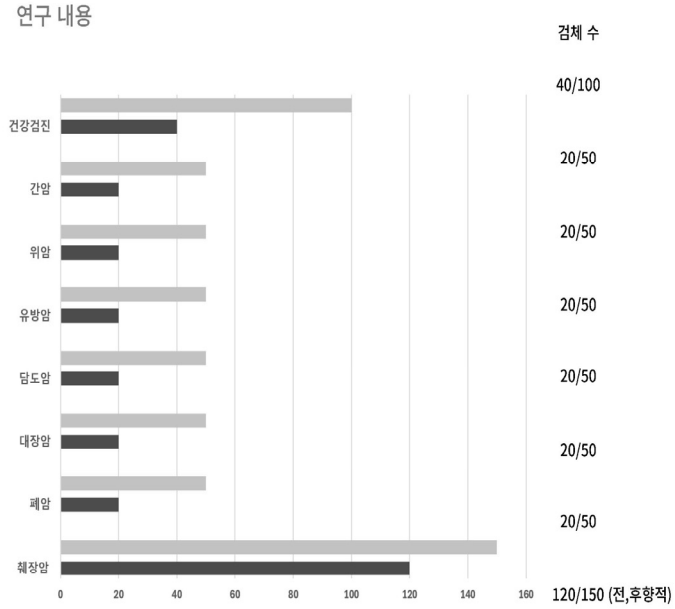
총 연구기간 2025.11.01~2027.10.31 | 1차년도 연구 기간 2025.11.01~2026.10.31

최종목표

췌장암과 폐암을 포함한 고위험 다종 암종의 조기진단 (Multi Cancer Early Detection)을 위해

초정밀 유전자가위 기반 ctDNA 분석과 다중 종양 단백질 분석을 통합한 유전 단백체 통합 진단 플랫폼(IPG-Dx) 을 개발하고 임상적 유효성을 검증

연구 내용



검체 수

40/100

20/50

20/50

20/50

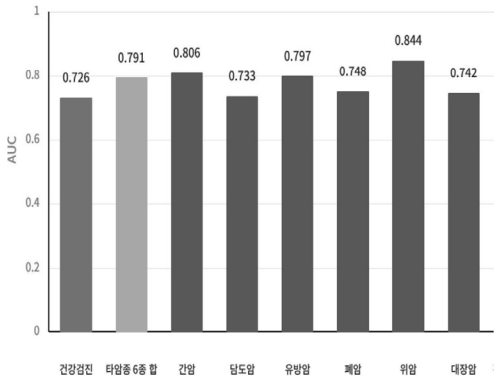
20/50

20/50

20/50

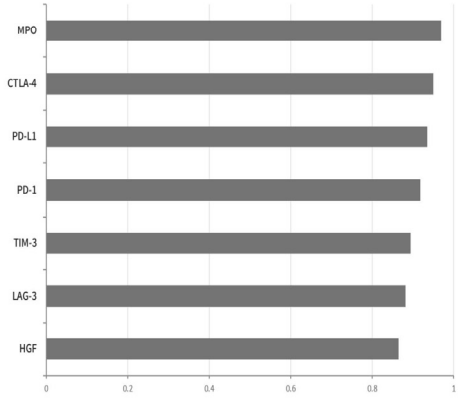
120/150 (전,후향적)

* 유전체 K ras 마커 AUC 비교



단백체 마커

* 전체 암 대비 건강검진 신호 상위 7종 AUC



Marker	AUC	민감도	특이도
MPO	0.969	93%	97%
CTLA-4	0.950	90%	91%
PD-L1	0.935	79%	97%
PD-1	0.918	70%	97%

* 각 암종 특이적 마커별 AUC heatmap

Marker	간암	담도암	대장암	위암	폐암	유방암	췌장암
MPO	0.98	0.99	0.98	0.98	0.92	0.96	0.97
CTLA-4	0.84	0.86	1.00	1.00	0.94	0.94	0.98
PD-L1	0.81	0.87	1.00	1.00	0.89	0.92	0.96
PD-1	0.86	0.90	1.00	1.00	0.88	0.94	0.90
AFP	0.90	0.68	0.58	0.62	0.58	0.60	0.99
HGF	0.99	1.00	0.66	0.57	0.82	0.63	1.00
OPN	0.84	0.96	0.65	0.52	0.67	0.53	0.77
HTIMP-1	0.70	0.93	0.80	0.72	0.52	0.52	0.65
CA125	0.51	0.64	0.55	0.56	0.51	0.72	0.98
IL-6	0.72	0.62	0.59	0.53	0.51	0.51	0.97
CA19-9	0.66	0.85	0.55	0.53	0.54	0.51	0.95
CEA	0.60	0.55	0.76	0.56	0.65	0.54	0.87

➔ MPO와 immune-checkpoint axis가 가장 강한 cancer signal 형성 : pan-cancer
➔ AFP・CA125・IL-6・CA19-9 조합은 특정 암종 축에서 상대적으로 강한 신호
➔ 19종 통합 시 가장 높은 분류 성능을 보이며, 면역/염증군의 기여도가 큰 것으로 확인

Early Diagnosis

울산대학교 글로컬대학 COMPaaS 사업 과제| 신산업 기반 기술 확보형

2024.06.27.(목) 14:00 ~ 14:25

샘플 전처리 및 유전자 증폭이 필요 없는 혈액 엑소좀 기반 췌장암 초고민감도 디지털 분자진단 기술 개발

Ultra-Sensitive Digital Molecular Diagnostics for Pancreatic Cancer Using Blood Exosomes Without Sample Preprocessing and Gene Amplification

- 연구책임자 1. 조윤경 (울산과학기술원, 바이오메디컬공학과)
- 연구책임자 2. 김송철 (울산대의과대학, 서울아산병원 간담도췌외과)

UNIST ULSAN NATIONAL INSTITUTE OF SCIENCE AND TECHNOLOGY 2009

울산대학교 UNIVERSITY OF ULSAN

Genome Wide Association Studies (GWAS) for PDAC; KPC-Gen GWAS Consortium

KPC-Gen update

NCC, October 14th, 2024

Laufey Amundadottir, PhD
Senior Investigator
Laboratory of Translational Genomics
Division of Cancer Epidemiology and Genetics

NIH NATIONAL CANCER INSTITUTE

PDAC GWAS in Asian populations

Finished:

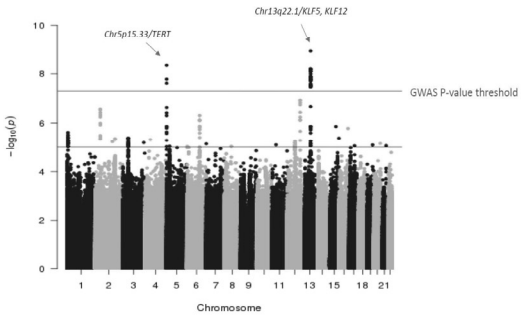
- **YALE-Shanghai**: 736 cases, 726 controls, PI: Dr. Harvey Risch
- **JapanPC**: 2,039 cases, 32,592 controls, PI: Dr. Yingsong Lin
- **Asian in PanScan**: 290 cases, 290 controls, Pis: multiple

Ongoing:

- ★ **Korean PDAC GWAS**: 3,300 cases, 5,600 controls, PI's: multiple / KPC-Gen
- **SMWHS GWAS**: 297 cases, 460 controls, PI's: Drs Xiao-Ou Shu and Wei Zheng

Total: ~7,000 PDAC and 40,000 controls

GWAS results in Korean PDAC and controls



Possible future KPC-Gen analyses

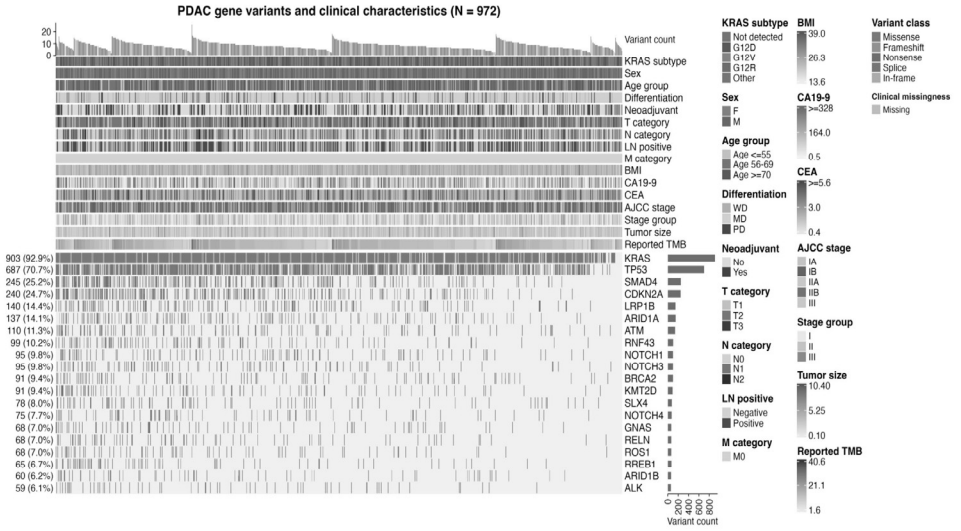
- **Assess high risk coding variants in KPC-Gen**
  - Use Whole Exome Sequencing (WES) – detect coding variants only
  - Use Whole Genome Sequencing (WGS) – coding and noncoding variants
  - Use Long Read Sequencing (Pac Bio etc) – coding/noncoding/structural variants
  - We have leftover DNA from most of the GWAS samples that could be used for this
- **Study epidemiological risk factor for PDAC in Korea**
  - Smoking, diabetes, alcohol use, pancreatitis etc.
  - Compare to European ancestry studies

Multi-omics for precision medicine and targeted therapy

Manuscript in preperation

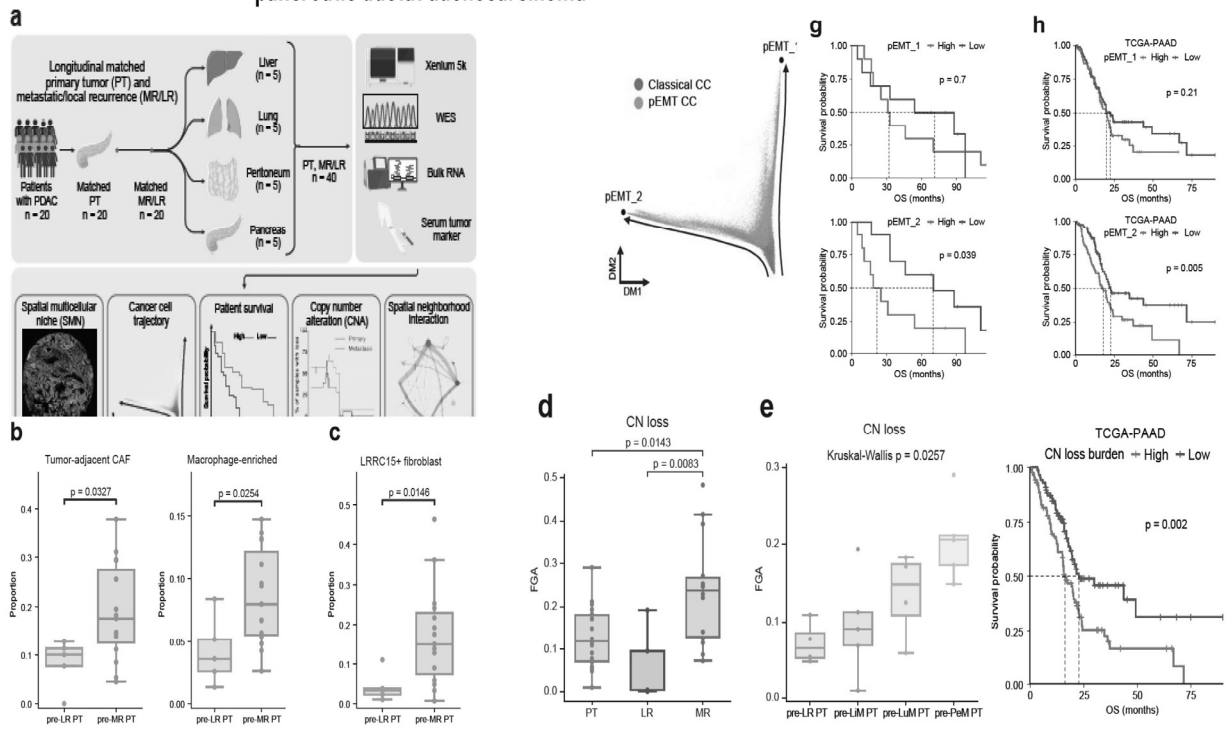
Integrated Clinical–Genomic Platform for Pancreatic Cancer at Asan Medical Center (Oncopanel-Precision)

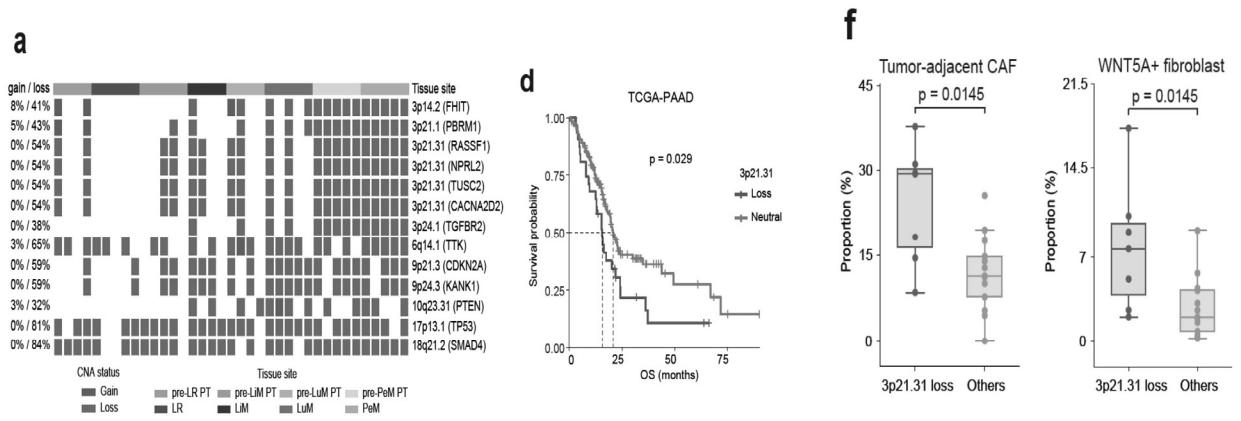
Clinicogenomic Characterization and Prognostic Analysis of Surgically Resected Pancreatic Ductal Adenocarcinoma: A Targeted Next-Generation Sequencing-Based Study



Spatiotemporal multi-omic profiling of primary and recurrent metastatic pancreatic ductal adenocarcinoma

Under revision at Nat Genetics





Summary

1.Recurrence site is already encoded in the primary tumor.

2.Tumors that go on to metastasize have an immunosuppressive environment.

3.The pEMT_2 state is linked to liver and peritoneal recurrence and worse survival..

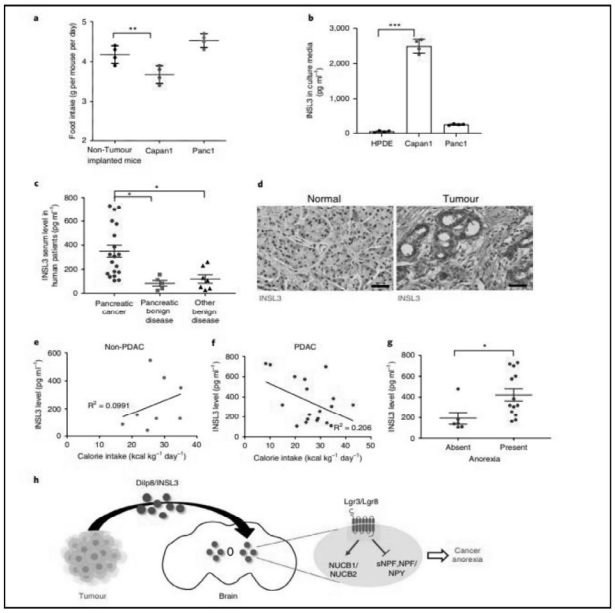
4.3p21.31 loss is linked to peritoneal recurrence and poor survival.

cancer cachexia

ARTICLES
https://doi.org/10.1038/s41556-020-00628-z
nature cell biology
Check for updates

Tumour-derived Dilp8/INSL3 induces cancer anorexia by regulating feeding neuropeptides via Lgr3/8 in the brain

Eunbyul Yeom[1,2,10], Hyemi Shin[3,10], Wonbeak Yoo[4,10], Eunsung Jun[5], Seokho Kim[6], Seung Hyun Hong[1], Dae-Woo Kwon[1,7], Tae Hoon Ryu[1,7], Jae Myoung Suh[3]✉, Song Cheol Kim[8]✉, Kyu-Sun Lee[1,7]✉ and Kweon Yu[1,7,9]✉



AMC Clinical Trial

Neoadjuvant mFOLFIRINOX vs Upfront surgery

- Resectable pancreatic cancer by NCCN resectability criteria
- ECOG PS 0–1
- No prior treatment
- Eligible medical condition for surgery

N=314

R

1:1

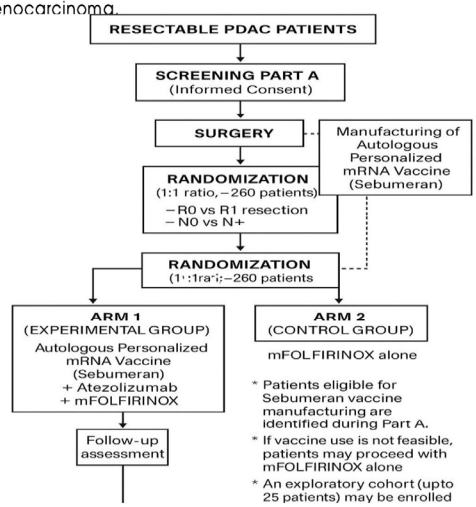
Upfront surgery → Postop adjuvant therapy for 6 months (mFOLFIRINOX is preferred)

Preoperative mFOLFIRINOX x 8 cycles → Surgery → Postop mFOLFOX x 4 cycles

Stratification factors: Tumor site (Head vs Body/Tail)

Primary endpoints : Event-free survival (EFS)
Hypothesis: median EFS 9 months → 13.4 months (HR 0.67)

A Phase II, open-label, multicenter clinical trial to evaluate the efficacy and safety of adjuvant therapy with **mFOLFIRINOX** combined with the **autologous personalized mRNA vaccine** ***Sebumeran*** and **atezolizumab** in patients with resected pancreatic ductal adenocarcinoma.



Raising Awareness and Building Hope for Pancreatic Cancer Patients

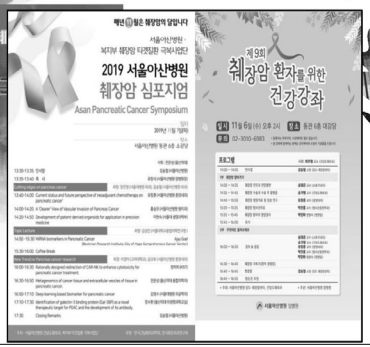




The 1st and 2nd Asan Medical Center

Pancreatic Cancer Hope Walk



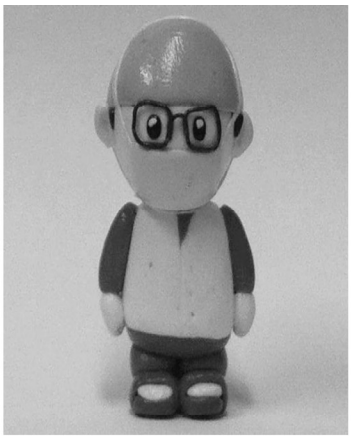






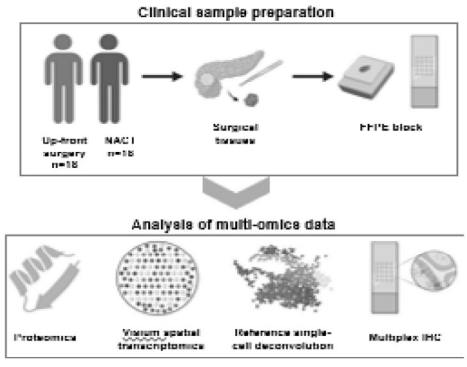
경청해 주셔서 감사합니다!

2026 서울아산 병원 간담췌 외과 "췌-담도암" 심포지엄에 많은 참석 부탁드립니다

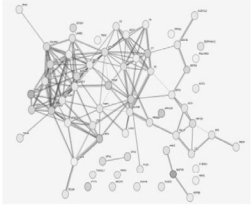


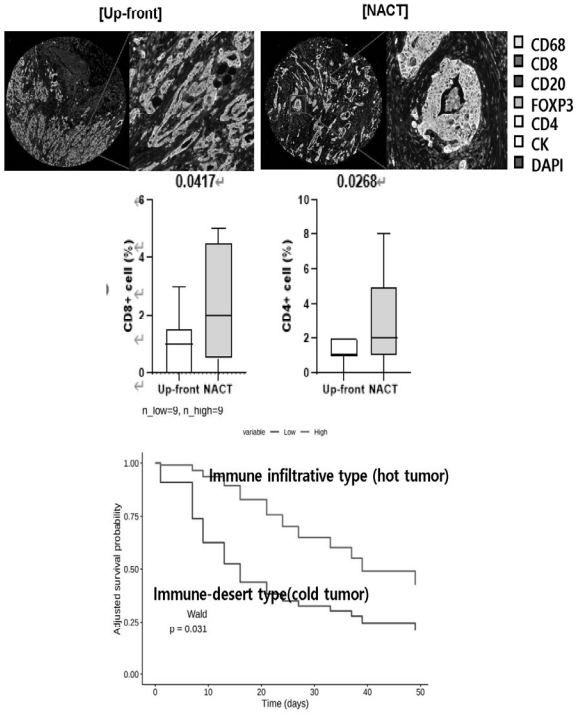
Multi-omics study of genoproteomic change after neoadjuvant -chemotherapy in pancreatic cancer

2024 국가 아젠다 과제



Proteomics





2026년 한국인체자원은행사업

KBP심포지엄 KOREABIOBANKPROJECT

Session 1

바이오뱅크 데이터를 활용한 한국인 질병 유전체 연구

서울대학교
이승근 교수

바이오뱅크 데이터를 활용한
한국인 질병 유전체 연구

이승근
Professor

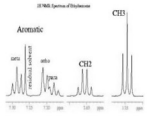
Graduate School of Data Science
Seoul National University

Biobank Data: Genome/OMICs + EHR + Biomarkers





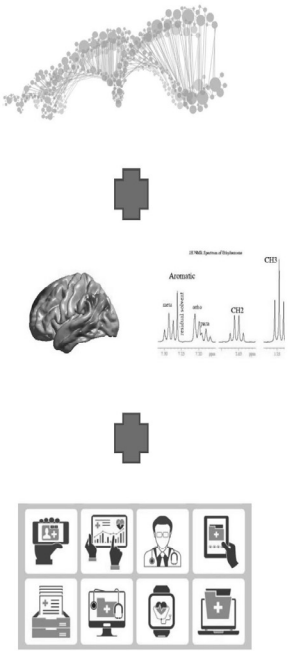


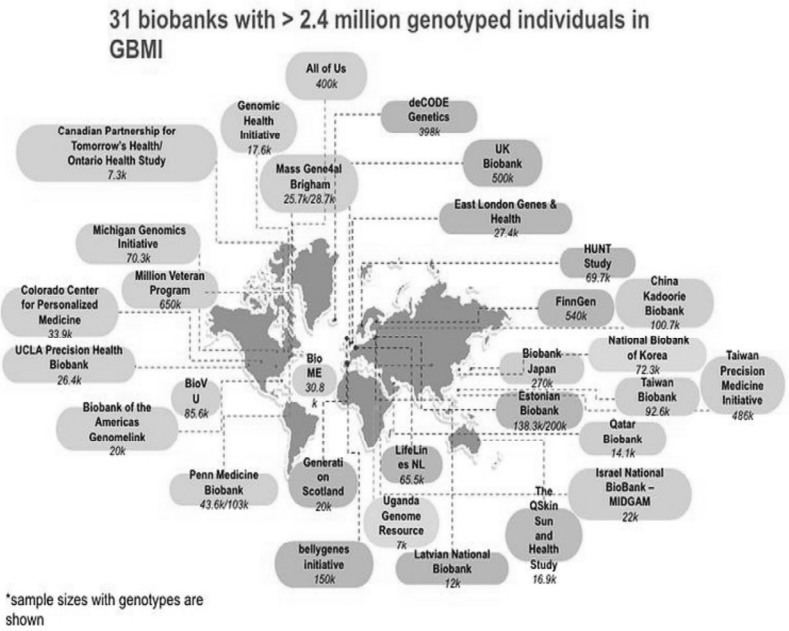






Biobank Data: Genome/OMICs + EHR + Biomarkers





Scalable and Accurate association tests

- Challenges in Big Biobank GWAS

Big data

- Large sample
- Large number of variants+ phenotypes

Statistical Issues

- Sample relatedness
- Unbalanced phenotypes

Efficient mixed model computation + **Saddlepoint Approximation**

Binary traits

- SAIGE: Zou et al. (2018), *Nature Genetics*
- FastSPA: Dey et al. (2017), *AJHG*

Categorical Phenotypes

- POLMM: Bi et al. (2021), *AJHG*

Survival

- SPAGE: Bi et al (2020), *AJHG*

Pathway

- GAUSS: Dutta et al. (2021), *AJHG*

Rare Variant Test



Big data

- Large sample
- Large number of variants+ phenotypes

Statistical Issues

- Sample relatedness
- Unbalanced phenotypes
- Gene-based test

Efficient mixed model computation + **Saddlepoint Approximation**

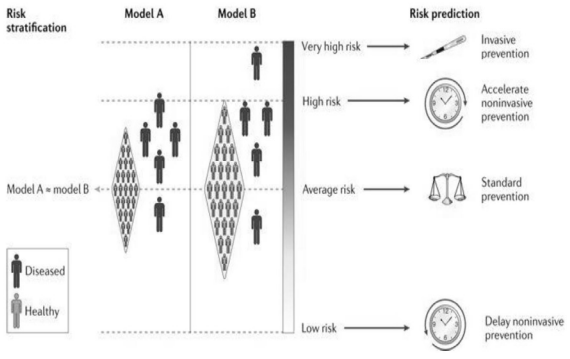
Rare variant test-Binary

- Meta-SAIGE: Park et al. (2025), *Nature Genetics*
- SAIGE-GENE+: Zou et al. (2022), *Nature Genetics*
- SAIGE-GENE: Zou et al. (2020), *Nature Genetics*

Rare variant test-Categorical

- POLMM-Gene: Bi et al. (2023), *AJHG*

Risk Prediction



Rare Variant

- RareEffect: Nam et al. (2026), *Nature Genetics*

Functional Annotation

- DeepUPRS: Lee et al. (2026), *Briefings of Bioinfo*

Cross-Ancestry

- TL-PRS: Zhao et al (2022), *AJHG*
- PTL-PRS: Cho et al (2025), Bioinformatics

Explanation/Bias Adjustment

- XPRS: Kim et al. (2025), Bioinformatics
- OAPRS: Jeong et al (2025), Alzheimer's & Dementia

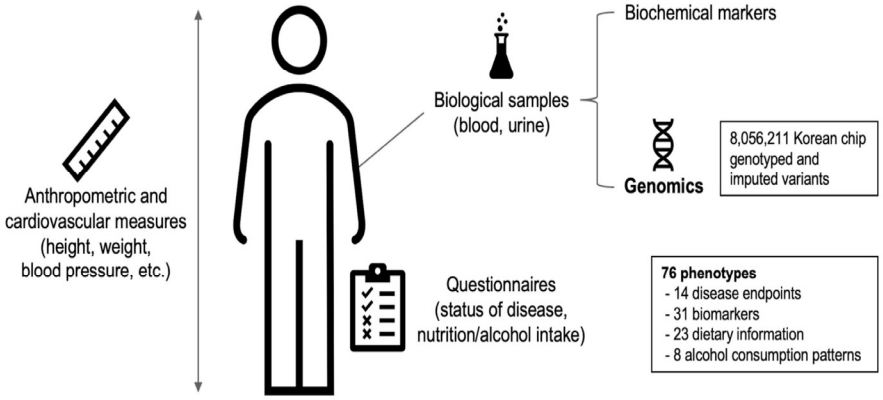
KoGES Data Analysis


Kisung Nam


Jangho Kim

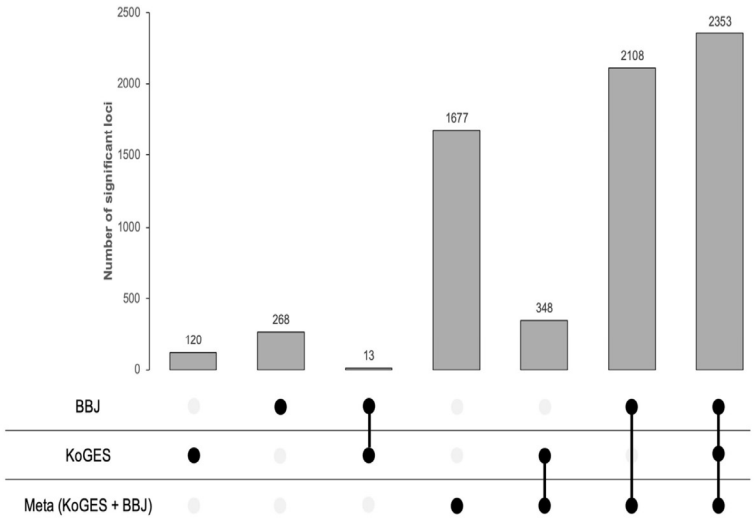
KoGES DATA

Cohort	Male	Female	Total	Mean age (SD)
Community	2,616	2,877	5,493	51.55 (8.51)
Urban	20,293	38,407	58,700	53.80 (8.02)
Rural	3,059	5,046	8,105	58.44 (8.84)
Total	25,968	46,330	72,298	54.15 (8.31)



Nam et al (2022) *Cell Genomics*

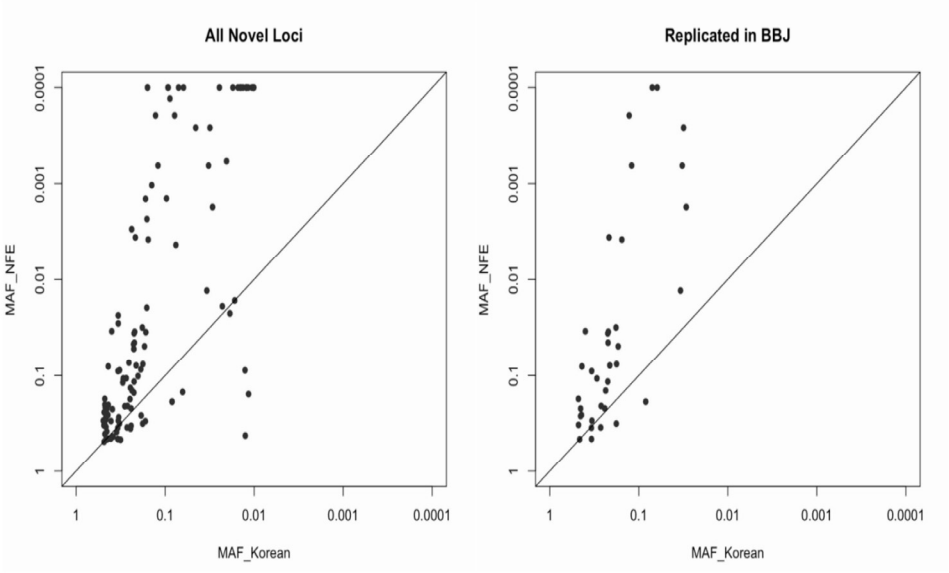
KoGES Data Analysis: Number of Associated Variants



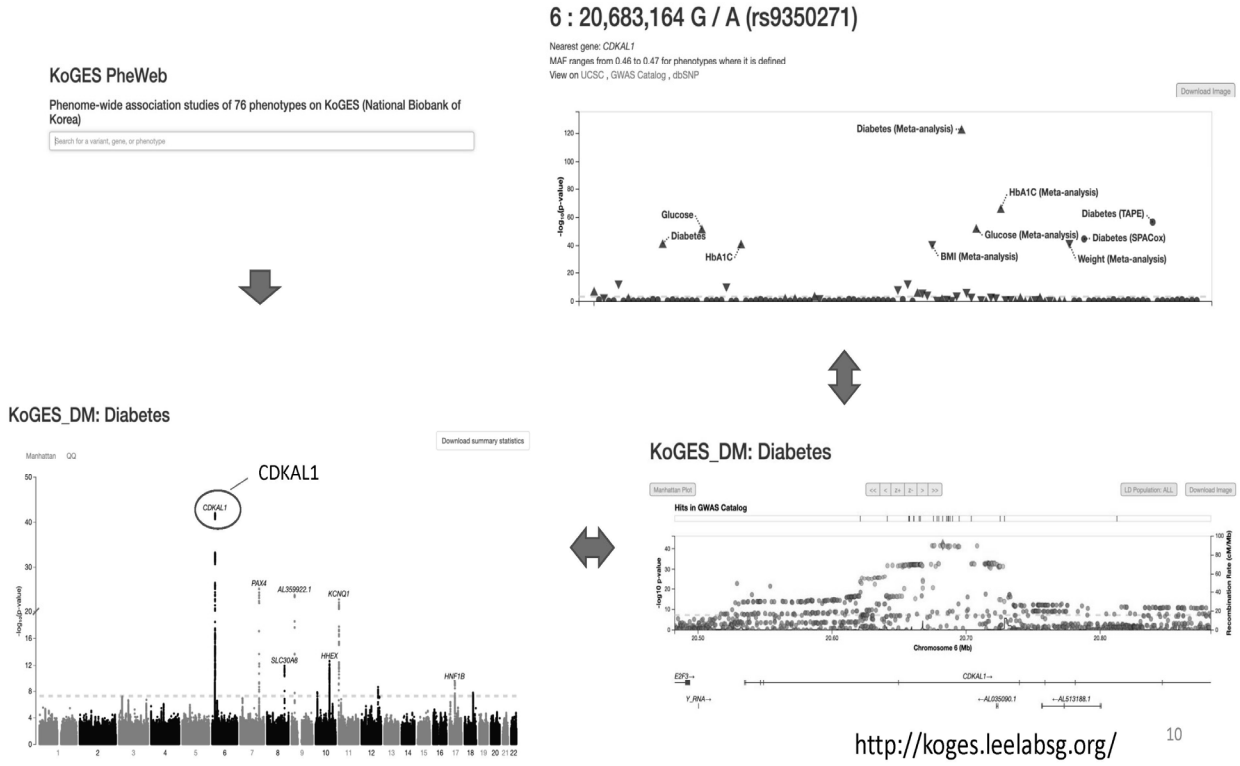
- > 2400 Genome-wide significant associations (p-value < 5E-8) in KOGES
  - Most of them present in BBJ
- Meta-analysis identifies more associations

KoGES Data Analysis: Novel Associations

- Novel loci tends to have low MAFs in European



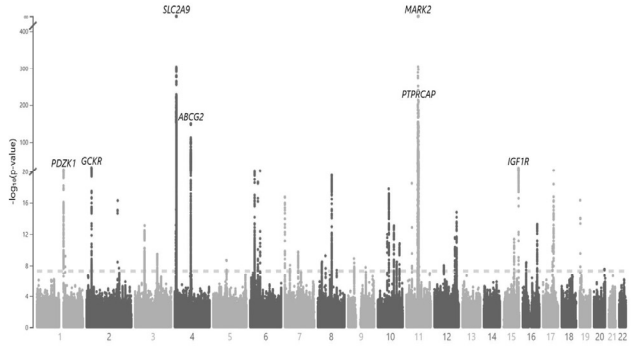
KoGES Data Analysis: PheWEB



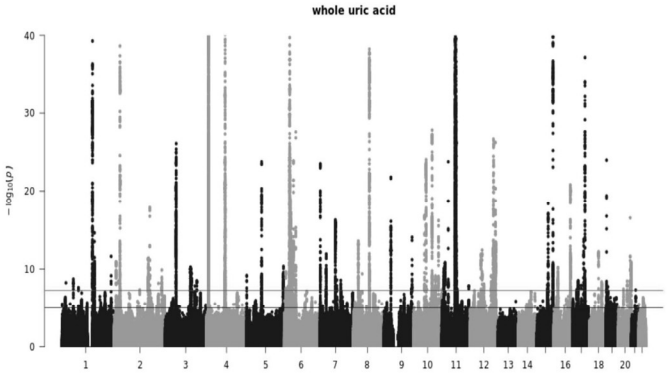
http://koges.leelabsg.org/

OpenKoGES: More samples

prev KoGES
samples: 66,693



OPEN KoGES
samples: 149,640

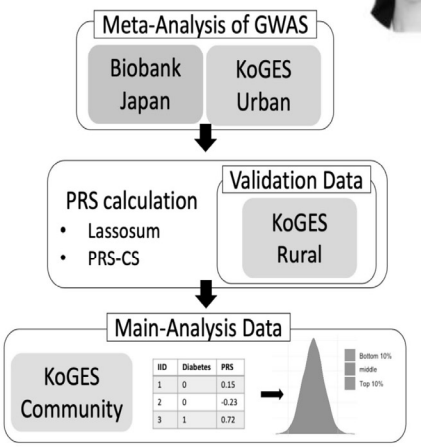


Korean T2D PRS





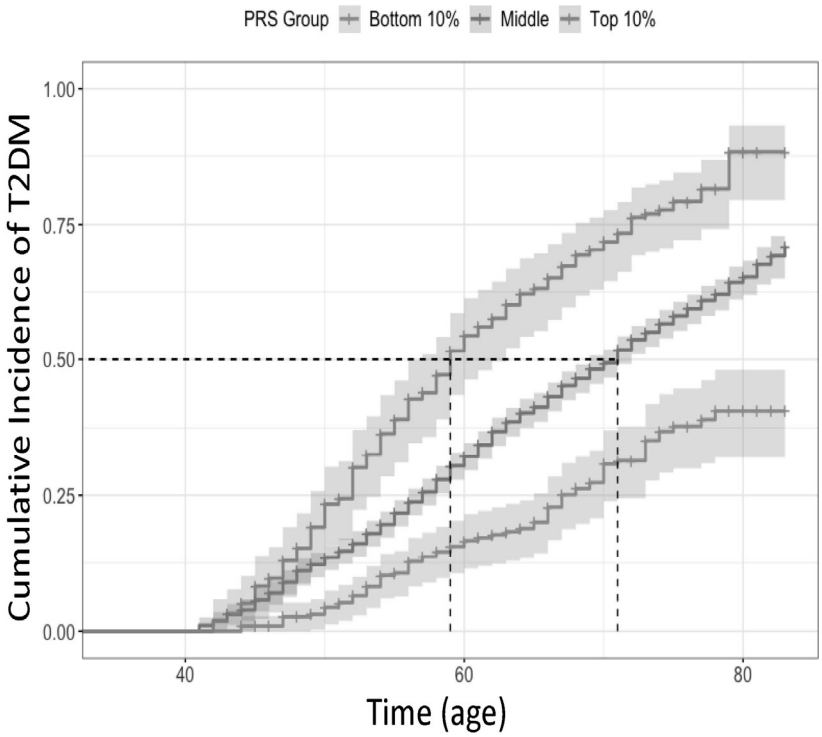
Cohort	Purpose	Sample Size
KoGES Urban	GWAS	58,698
KoGES Rural	Validation Set	8,105
KoGES Community (Ansan_Ansung)	Main-Analysis Data	5,493
BBJ	GWAS	210,865



1. T2DM progression
2. T2DM severity
3. Association with biomarker
4. Evaluation of prediction model with PRS and traditional risk factors

Kim et al (2024) *Sci Report*

Korean T2D PRS



Korean T2D PRS

(% ↑) increase in C-index (%)	Without PRS	With Standardized PRS	Standardized PRS vs. without PRS	
	Harrell C (96% CI)	Harrell C (96% CI)	C-Index Difference	p-value
Model 1 (sex + age)	0.567 (0.550 – 0.584)	0.623 (0.607 – 0.639)	0.056 (10% ↑)	< 0.001
Model 2 (+family history)	0.586 (0.569 – 0.603)	0.631 (0.615 – 0.647)	0.045 (7.7% ↑)	< 0.001
Model 3 (+ traditional risk factors)	0.656 (0.640 – 0.672)	0.684 (0.668 – 0.699)	0.027 (4.1% ↑)	< 0.001

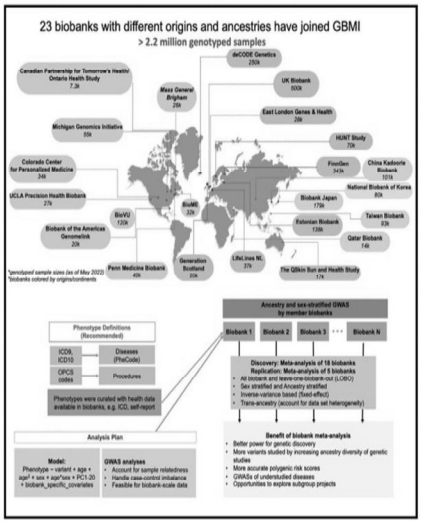
Global Collaboration

Cell Genomics

Article

Global Biobank Meta-analysis Initiative: Powering genetic discovery across human disease

Graphical abstract



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In brief

Zhou et al. present the flagship project of the Global Biobank Meta-analysis Initiative (GBMI). They demonstrate the substantial benefits of the collaborative efforts of 23 biobanks worldwide to advance genetic discoveries for human diseases with larger sample sizes and increased ancestry diversity and highlight issues and challenges in biobank meta-analyses.

Global Collaboration

nature genetics

Technical Report

https://doi.org/10.1038/s41588-023-01500-0

Boosting the power of genome-wide association studies within and across ancestries by using polygenic scores

Received: 15 December 2022

Accepted: 14 August 2023

Published online: 18 September 2023

Check for updates

Adrian I. Campos[1], Shinichi Namba[2], Shu-Chin Lin[3], Kisung Nam[4], Julia Sidorenko[1], Huanwei Wang[1], Yoichiro Kamatani[5], The Biobank Japan Project*, Ling-Hua Wang[6], Seunggeun Lee[4], Yen-Feng Lin[3], Yen-Chen Anne Feng[6,7], Yukinori Okada[2,8,9,10,11], Peter M. Visscher[1] & Loic Yengo[1]

www.nature.com/mp

Molecular Psychiatry

ARTICLE

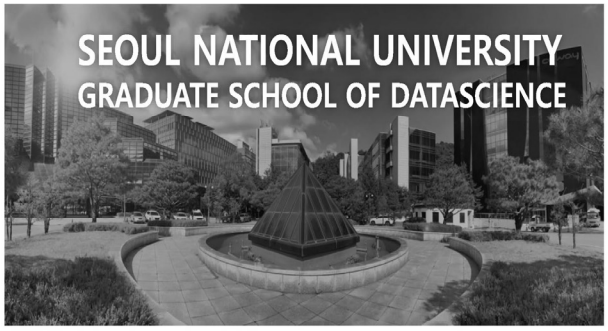
Check for updates

A large-scale genome-wide study of gene-sleep duration interactions for blood pressure in 811,405 individuals from diverse populations

Acknowledgement

국립중앙인체자원은행 질병관리청 국립보건연구원

보건의료연구자원정보센터 CODA



- Current lab members



- Supported by National Research Foundation and Ministry of Food and Drug Safety

2026년 한국인체자원은행사업

KBP심포지엄 KOREABIOBANKPROJECT

Session 1

육종 연구를 위한 혁신형 바이오뱅킹 사업

국립암센터
김준혁 교수

BICSWAN

2026 KBP SYMPOSIUM | NOVEMBER 24, 2026

육종 연구를 위한
혁신형 바이오뱅킹 사업

June Hyuk Kim, MD

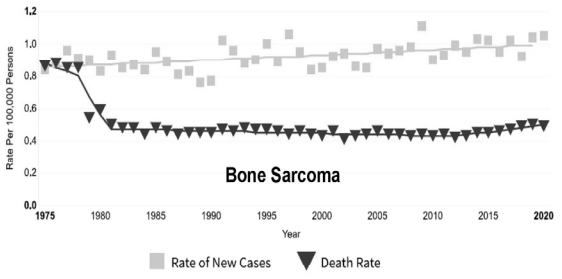
Center for Sarcoma

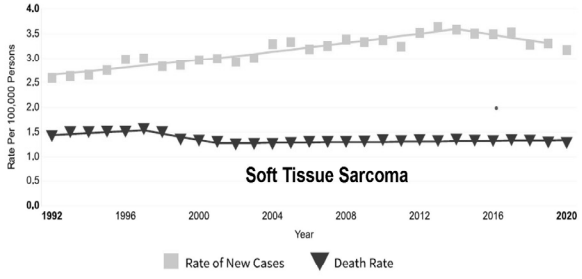
National Cancer Center, Korea

국립암센터
NATIONAL CANCER CENTER

Unmet Needs in Musculoskeletal Oncology

- Molecular Re-classification
- New Drug Development (Re-targeting)
- Precision Medicine in Sarcoma Patients





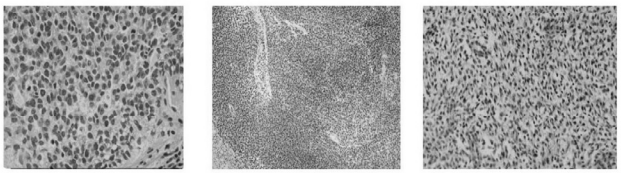
SEER database

Molecular Subtyping of Soft Tissue Sarcoma

TUMOR	ABERRATION	GENE(S) INVOLVED
Malignant Round Cell Tumors		
Alveolar RMS	t(2;13)(q35;q14) t(1;13)(p36;q14) t(X;2)(q13;q35)	*PAX3-FOXO1* *PAX7-FOXO1* *PAX3-AFX*
Desmoplastic small round cell tumor	t(11;22)(p13;q12)	*EWSR1-WT1*
Embryonal RMS	Complex alterations	Multiple, *MYOD1, KRAS, HRAS, TP53, NF1, NRAS, PIK3CA, FBXW7, FGFR4, BCOR*
Ewing sarcoma/peripheral neuroectodermal tumor	t(11;22)(q24;q12) t(21;22)(q22;q12) t(2;22)(q33;q12) t(7;22)(p22;q12) t(17;22)(q12;q12) inv(22)(q12q;12) t(16;21)(p11;q22)	*EWSR1-FLI1* *EWSR1-ERG* *EWSR1-FEV* *EWSR1-ETV1* *EWSR1-E1AF* *EWSR1-ZSG* *FUS-ERG*
Undifferentiated round cell sarcoma	t(4;19)(q35;q13) or t(10;19)(q26;q13) inv(X)(p11.4p11.22)	*CIC-DUX4*[3] *BCOR-CCNB3*[4]
Lipomatous Tumors		
Atypical lipomatous tumor/well-differentiated liposarcoma (ALT/WDLS)	Supernumerary ring chromosomes; giant marker chromosomes	Amplification of region 12q14-15, including *MDM2, CDK4, HMGA2, SAS, GLI*
Dedifferentiated liposarcoma	Same as for ALT/WDLS	Same as for ALT/WDLS
Myxoid/round cell liposarcoma	t(12;16)(q13;p11) t(12;22)(q13;q12)	*FUS-DDIT3* *EWSR1-DDIT3*
Pleomorphic liposarcoma	Complex alterations	Unknown

TUMOR	ABERRATION	GENE(S) INVOLVED
Epithelioid hemangioendothelioma	t(1;13)(p36;q25) t(X;11)(q22;p11.23)	*WWTR1-CAMTA1* *YAP1 - TFE3*
Other Sarcomas - continued		
Epithelioid sarcoma	Inactivation, deletion, or mutation of *INI1 (SMARCB-1)*	*INI1 (SMARCB-1)*
Extrarenal rhabdoid tumor	Inactivation of *INI1 (SMARCB-1)*	*INI1 (SMARCB-1)*
Extraskeletal myxoid chondrosarcoma	t(9;22)(q22;q12) t(9;17)(q22;q11) t(9;15)(q22;q21) t(3;9)(q11;q22)	*EWSR1-NR4A3* *TAF2N-NR4A3* *TCF12-NR4A3* *TFG-NR4A3*
Sporadic and familial GIST Carney-Stratakis syndrome (gastric GIST and paraganglioma)	Activating kinase mutations Krebs cycle mutation	*KIT* or *PDGFRA* *Germline SDH* subunit mutations
Inflammatory myofibroblastic tumor (IMT)	t(1;2)(q22;p23) t(2;19)(p23;p13) t(2;17)(p23;q23) t(2;2)(p23;q13) t(2;11)(p23;p15) inv(2)(p23;q35)	*TPM3-ALK*[5] *TPM4-ALK*[5] *CLTC-ALK*[5] *RANBP2-ALK*[5] *CARS-ALK*[5] *ATIC-ALK*[5]
Leiomyosarcoma	Complex alterations	Unknown
Low-grade fibromyxoid sarcoma	t(7;16)(q33;p11) t(11;16)(p11;p11)	*FUS-CREB3L2* *FUS-CREB3L1*
Malignant peripheral nerve sheath tumor		*NF1, CDKN2A* and *EED* or *SUZ12*
Mesenchymal chondrosarcoma	t(8;8)(q13;q21)	*HEY1 - NCOA2*
Solitary fibrous tumor	inv(12)(q13q13)	*NAB2 - STAT6*
Synovial sarcoma	t(X;18)(p11;q11) t(X;18)(p11;q11) t(X;18)(p11;q11)	*SS18-SSX1* *SS18-SSX2* *SS18-SSX4*
Tenosynovial giant cell tumor/pigmented villonodular synovitis (TGCT/PVNS)	t(1;2)(p13;q35)	*CSF1*

Ewing sarcoma **CIC sarcoma** **BCOR sarcoma**



- NTRK **spindle cell tumor**
- PRRX.:NCOA1/2 **distinctive fibroblastic neoplasm**
- EWSR1::SMAD3 **positive fibroblastic tumor**
- MGA::NUTM1 **new subtype of high-grade spindle cell sarcoma**
- CRTC1 ::SS18 **novel sarcoma subtype**
- GLI **sarcoma**

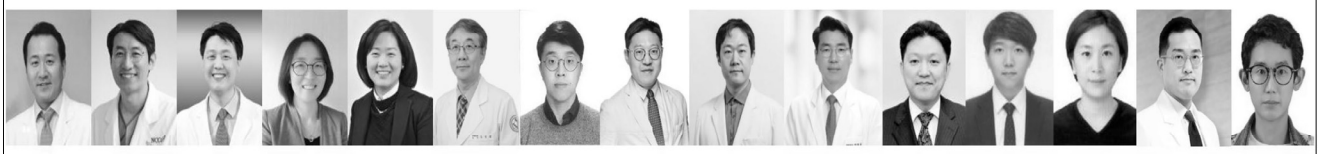
Korea Biobank Project Milestones

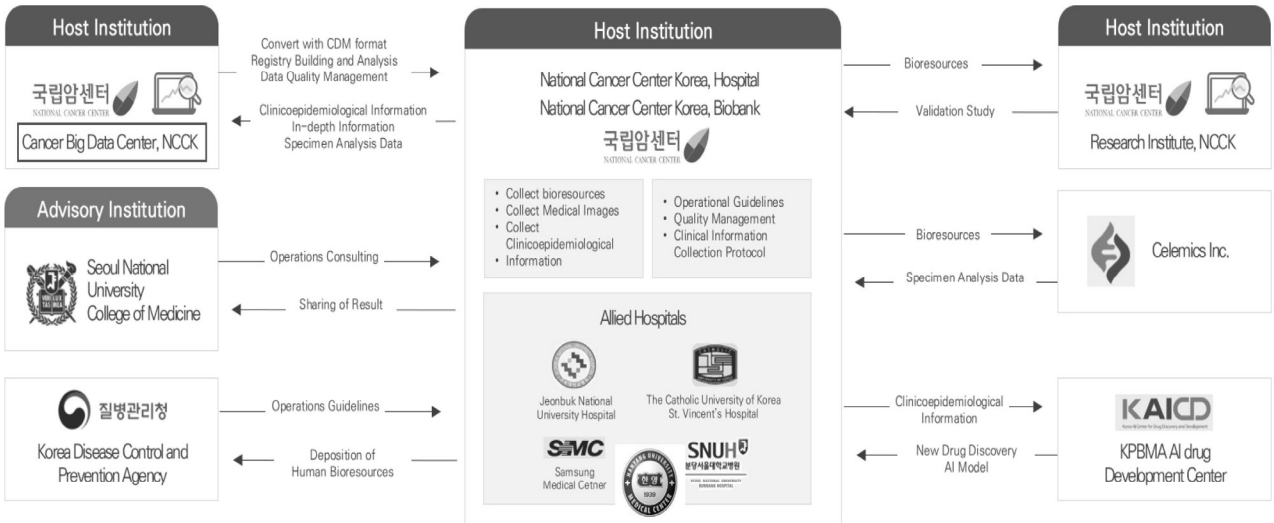
	Pre-KBP	Phase 1	Phase 2	Phase 3	Phase 4
Period	2001–2007	2008–2012	2013–2015	2016–2020	2021–2025
Objectives	Established the biospecimen management foundation and Division of Biobank	Established a national network and expanded biospecimen collection	Expanded the network and promoted use of distributed biospecimens	Expanded the network and facilitated biospecimen access for industry	Restructured the network and achieved ISO 20387 accreditation
Collection	190,000	520,000	710,000	990,000	1.23 million
Distribution	48 projects	700 projects	1,500 projects	3,600 projects	5,500 projects
Institutions	NBK	NBK and 17 affiliated biobanks	NBK, 17 affiliated biobank and 2 KBN partners	NBK, 17 affiliated biobanks and 2 KBN partners	NBK, ~30 KBN biobanks, 2 KBN partners and **4 innovative consortia (BICSWAN)**
Budget	—	KRW 45.1B	KRW 27.6B	KRW 44.5B	KRW 59.5B

NBK, National Biobank of Korea. KBN, Korea Biobank Network.

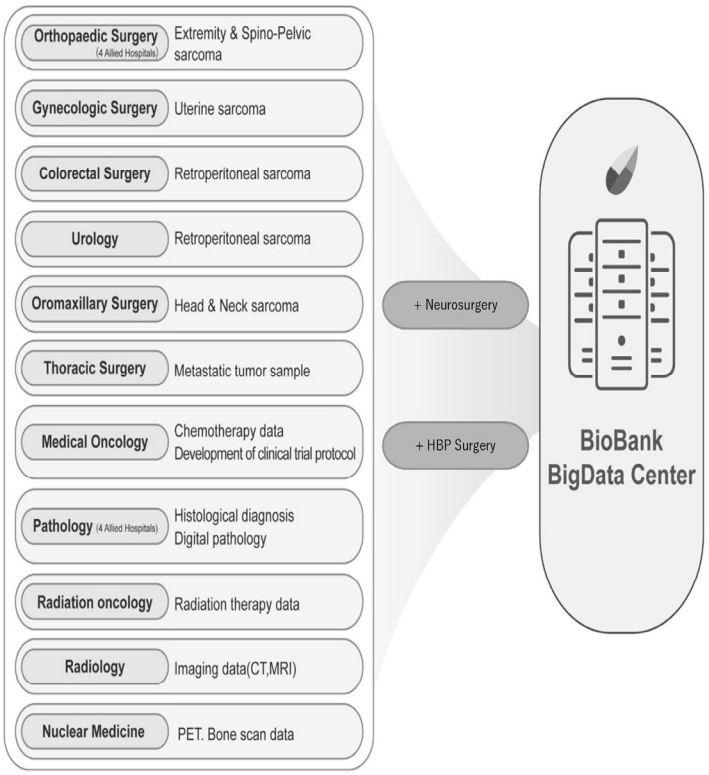
The BICSWAN Consortium

Biobank Innovative Consortium for Sarcoma With Advancements and Networking (2021–)





Multidisciplinary Biospecimen Collection



Collection priorities

- Distribute study guidance to surgical and medical teams.
- Expand snap-freezing to reduce cold-ischemic time
- Collect additional plasma at postoperative visits(>1yr).
- Research nurses track surgical procedures (every week).
- Target normal tissue collection in ≥50% of cases.

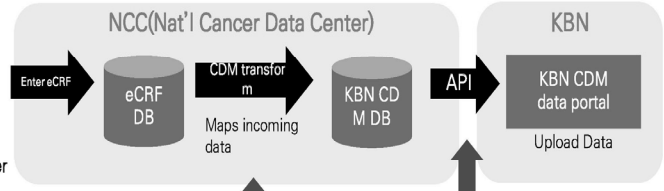
Multicenter Clinical Research Data Platform

KBN CDM: 150 columns and 300 clinical items (88 common, 212 disease-specific)

Follow-up includes recurrence and survival updates for previously enrolled patients



- NCCK
- Jeonbuk Natl Unv. H
- Samsung Medical Center
- SNU Bundang H
- Hanyang Univ. H





Enter and upload data through a common eCRF

The NCC Cancer Data Center maps and transforms incoming data



KBN CDM version 4.0

- Collect, transform and load new records into the KBN CDM
- Update ETL and the KBN database to support versions 4.0



API management

- Adapt and maintain the API modules for KBN CDM 4.0

국립암센터

Sarcoma Biobanking, June 2021–Sep. 2026

Year	No.(Pt.)	FF(T)		FF(N)		FFPE(T)		FFPE(N)		plasma		Serum		Buffy coat		whole blood	
		No.	vials	No.	vials	No.	blocks	No.	blocks	No.	vials	No.	vials	No.	vials	No.	vials
2021	46	28	106	13	34	20	20	4	4	43	205	44	211	41	41	45	89
2022	171	103	336	60	186	43	57	18	33	141	698	142	702	122	122	138	274
2023	233	144	555	83	267	57	80	24	57	204	1,043	204	1,048	184	187	204	526
2024	188	129	400	73	267	75	123	62	119	168	840	168	836	167	167	167	334
2025	183	70	253	75	510	34	49	46	54	147	291	147	707	147	291	147	147
2026	244	117	461	176	806	45	45	55	63	203	1,003	203	1,010	202	207	202	461
Total	1,065	591	2,111	480	2,070	274	374	209	330	906	4,080	908	4,514	863	1,015	903	1,831

* Almost all: Treatment-naïve Samples *Additional Resources: TMA, Liquid biopsy, Digital pathology, Imaging(chest CT, MRI, PET-CT) * FFPE curls available

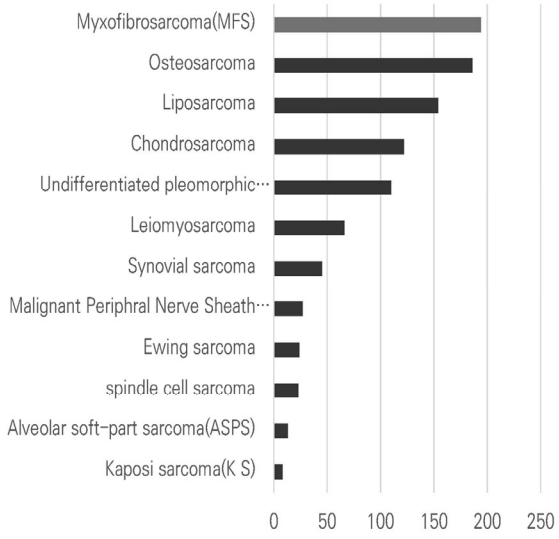
Recurrence and Metastasis Samples

Year	No.(Pt.)			Blood(Vials)					FF(Vials)			FFPE(Blocks)		
	Total	recur	mets	No.	whole	serum	plasma	buffy	No.	normal	tumor	No.	normal	tumor
2022	11	7	5	10	20	50	50	9	8	-	15	1	-	1
2023	23	14	11	22	52	127	130	26	22	3	74	6	-	8
2024	32	25	10	31	70	172	180	36	23	30	59	12	7	9
2025	54	39	15	37	84	195	201	40	16	39	74	8	9	12
2026	45	32	13	38	196	197	39	81	21	44	91	9	6	9
Total	165	117	54	138	422	741	600	192	90	116	313	36	22	39

MALIGNANCY vs BORDERLINE TUMOR

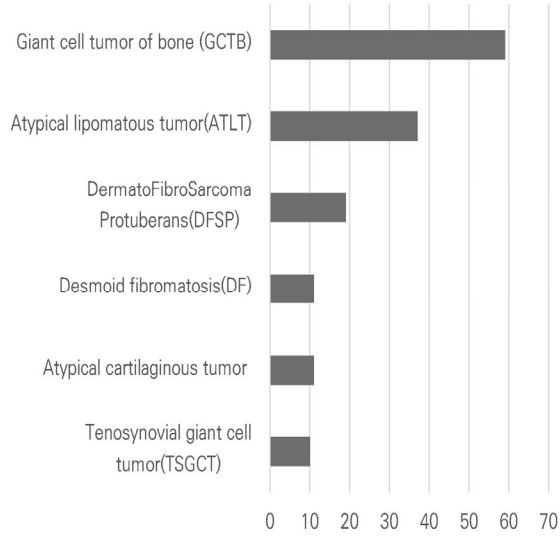
육종암 (악성종양 vs 경계성 종양)

악성 (MALIGNANT) · 1,009건 · 87.3%



악성 아형 중 Myxofibrosarcoma(186건)와 Osteosarcoma(179건)가 가장 많습니다.

경계성 (BORDERLINE / INTERMEDIATE) · 137건 · 11.8%

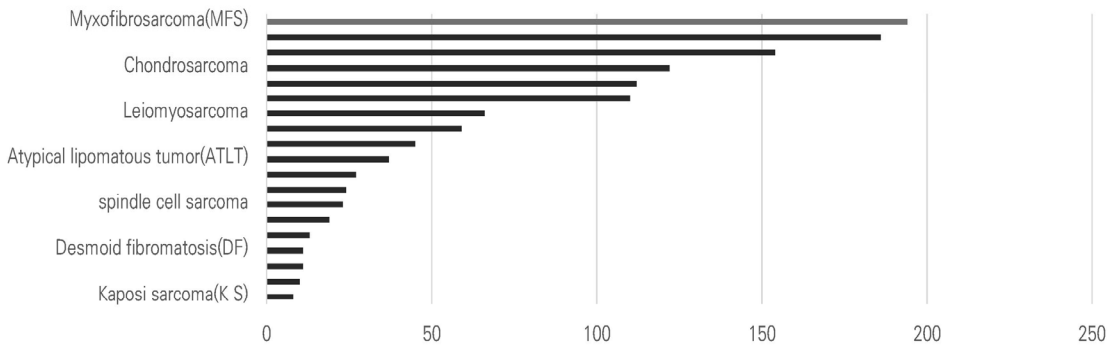


경계성 아형 중 GCTB(59건)가 절반 가까이를 차지합니다.

SUBTYPE PREVALENCE

육종암 전체 아형별 건수

육종 아형별 건수 (상위 10개 및 기타)



Myxofibrosarcoma(182건)와 Osteosarcoma(179건) 두 질환이 총 361건으로 전체의 31.2%를 점유합니다.

Myxofibrosarcoma (182건)

단일 아형 중 최다 빈도, 연조직 육종의 핵심군

Osteosarcoma (179건)

골 유래 육종 중 최다 빈도로 1위 점유

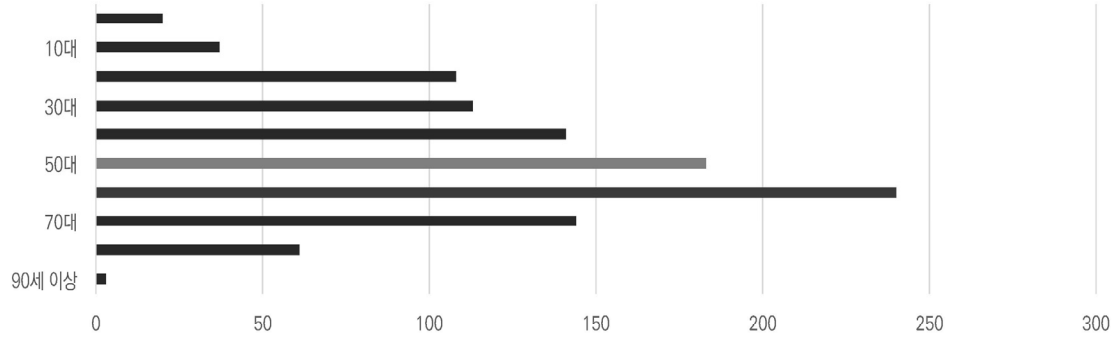
기타 아형 (112건)

- Endometrial stromal sarcoma(ESS)
- Epithelioid sarcoma
- Rhabdomyosarcoma
- Undifferentiated spindle cell sarcoma
- Undifferentiated sarcoma
- Uterine sarcoma

AGE DEMOGRAPHICS

연령대별 육종암 발생 건수

연령대별 건수 (10세 단위, N = 1,155)



60대가 240건으로 단일 연령군 정점을 형성하며, 50대(182건)와 70대(143건) 등 50~70대 고령층에 다수 분포합니다.

60대 최다 발생 (240건)

전체 기재 환자의 21.6% 점유

50~70대 집중 (567건)

중장년 및 고령 환자가 과반 이상(51.1%)

10~20대 청소년·청년 (165건)

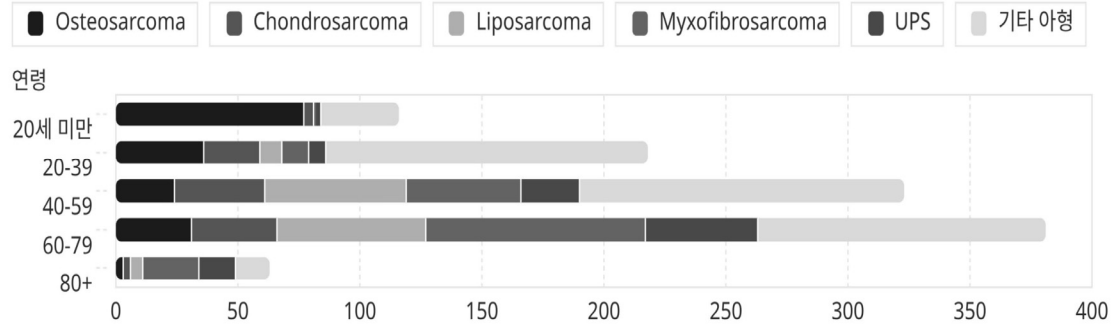
골육종 호발 양상

AGE-SUBTYPE CORRELATION

연령에 따라 주된 육종 아형

연령 구간별 상위 5대 아형 누적 분포

20세 미만은 골육종(77건)이 압도적 우세를 보이며, 60대 이후에는 점액섬유육종과 UPS 비율이 급증



소아·청소년기: Osteosarcoma 지배

20세 미만군 전체 116건 중 Osteosarcoma가 77건(66.4%)으로 독보적인 비중을 차지합니다.

고령기: Myxofibrosarcoma & UPS 급증

60~79세군에서 Myxofibrosarcoma(90건)와 UPS(46건)가 동반 상승하며, 80세 이상에서도 동일한 고령형 아형 패턴이 지속됩니다.

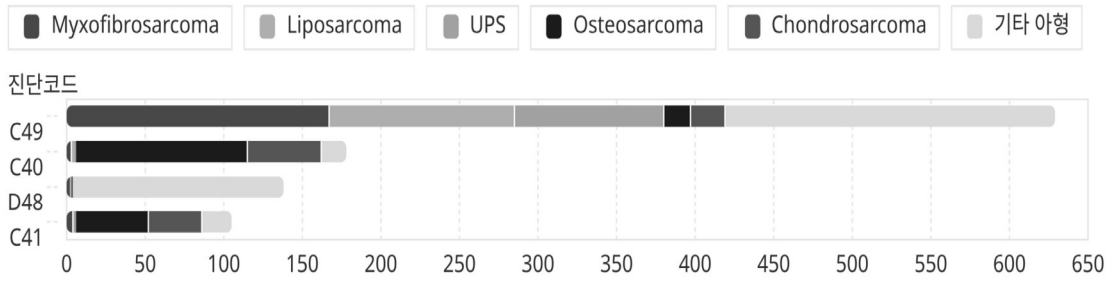
Made with GAMMA

CODE-SUBTYPE CORRESPONDENCE

진단코드별 대표 아형

주요 진단코드별 상위 5대 아형 누적 분포

C49는 점액섬유육종과 지방육종이 주도하고, C40과 C41은 골육종 및 연골육종이 집중되며, D48은 기타 희귀 아형이 대부분입니다.



C49 연조직 코드

Myxofibrosarcoma 167건, Liposarcoma 118건, UPS 95건 등 연조직 아형 중심 구성

C40 & C41 골 코드

C40은 Osteosarcoma 109건, C41은 Chondrosarcoma 34건 및 Osteosarcoma 46건 집중

D48 경계성 종양

GCTB 등 5대 아형 외의 기타 병변이 134건(97.1%)으로 절대 다수를 형성

Derived Research Resources, June 2021–September 2026

Resource	Count (patients)
Cell line development	
■ Osteosarcoma cell lines	29
■ Adult soft tissue sarcoma cell lines	11
Genomic analysis	
■ Sarcoma fusion gene data	24
■ Whole Genome Sequencing	115
■ Whole Exome Sequencing	70
■ Whole Transcriptome sequencing	95
■ Target DNA Data	404
■ Target RNA Data	113
■ Proteomics	11
■ Multi-omics embedding model development and training	45
■ Digital pathology	203/564 slides
■ TMA	14blocks/159cores
■ Single cell/Spatial Transcriptome Sequencing	37

Longitudinal Samples, Digital Pathology and TMAs

Longitudinal resources

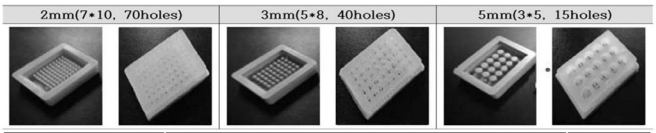
Compare primary and recurrent tumors to investigate recurrence mechanisms.

Compare pretreatment liquid biopsies with samples collected ≥1 year after curative resection while disease-free (IRB amendment).

Digital Pathology

De-identify and convert whole-slide images to TIFF. Upload via secure transfer, then link image metadata through the KBN API.

TMAs



Group	Histology	Core
BS-1	Osteosarcoma	3 mm
BS-2	Chondrosarcoma	3 mm
BS-3	Ewing sarcoma	5 mm
BS-other	Chordoma and other bone sarcomas	5 mm
STS-1	Myxofibrosarcoma	3 mm
STS-2	Undifferentiated sarcoma	3 mm
STS-3	Liposarcoma	3 mm
STS-4	Leiomyosarcoma	5 mm
STS-5	Synovial sarcoma	5 mm
STS-6	Malignant peripheral nerve sheath tumor	5 mm
STS-other	Other soft tissue sarcomas	5 mm
Intermediate 1	Giant cell tumor of bone	3 mm
Intermediate 2	Atypical lipomatous tumor	3 mm
Intermediate 3	Desmoid-type fibromatosis	3 mm
Intermediate 4	Atypical cartilaginous tumor	5 mm
Intermediate 5	Dermatofibrosarcoma protuberans	5 mm

Publication utilizing TMAs

SPANDIDOS PUBLICATIONS

ONCOLOGY LETTERS 30: 552, 2025

Impact of SAMHD1 and phosphorylated ATM levels on the progression and prognosis of patients with soft tissue sarcoma

YUN JAE KIM[1*], YOUNG JAE MOON[2,3*], AE RI AHN[1], HO SUNG PARK[1], MYOUNG JA CHUNG[1], WOO SUNG MOON[1], KYU YUN JANG[1,4,5] and KYOUNG MIN KIM[1,4,5]

- 133 STS patients
- FFPE tissue blocks for TMA construction
- SAMHD1, p-ATM IHC staining & scoring
- co-expression of SAMHD1& p-ATM : the poorest outcomes

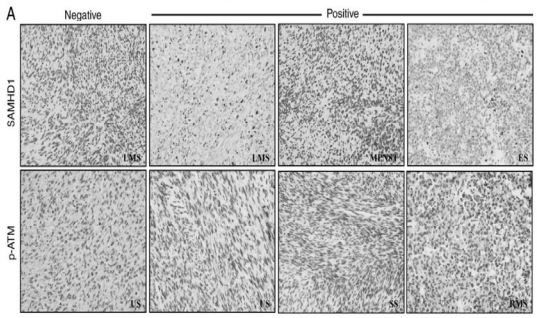
∴ DNA damage repair(DDR) pathways serve a crucial role in sarcoma progression

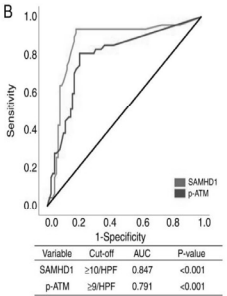
Young Jae Moon. MD.



Kyoung Min Kim. MD.







Variable	Cut-off	AUC	P-value
SAMHD1	≥10/HPF	0.847	<0.001
p-ATM	≥9/HPF	0.791	<0.001

Kim et al. Oncology Letters, 2025

Sarcoma Cell Lines and Experimental Models

Established resources

Cell lines established across at least 7 sarcoma types.

Examples include leiomyosarcoma, osteosarcoma, alveolar sarcoma, undifferentiated sarcoma, synovial sarcoma and liposarcoma.

Experimental models

In vitro

Sarcoma cell lines support 2D and 3D cultures.

In vivo

Patient tumor tissue could be used to develop mouse xenograft models.

Sarcoma NGS Panel Development with Celemics

CELEMICS

Panel design

Select genes from COSMIC, ClinVar and TCGA.

Define target regions, exons and hotspots for target enrichment.

Validate design in silico and manage panel versions.

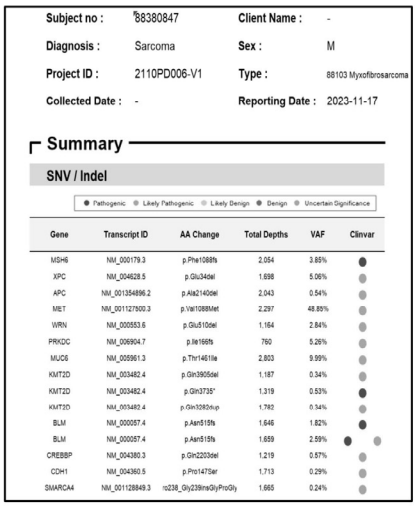
Specimen testing

Perform clinical specimen QC, DNA/RNA extraction and library preparation.

Pilot platform-specific protocols and test liquid biopsies.

Diagnostic reporting

Apply ACMG and AMP/ASCO/CAP guidance. Include process and data QC, clinical interpretation and final review.



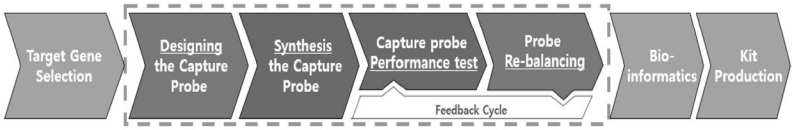
Subject no :	88380847	Client Name :	-
Diagnosis :	Sarcoma	Sex :	M
Project ID :	2110PD006-V1	Type :	88103 Myxofibrosarcoma
Collected Date :	-	Reporting Date :	2023-11-17

Summary

SNV / Indel

● Pathogenic ● Likely Pathogenic ● Likely Benign ● Benign ● Uncertain Significance

Gene	Transcript ID	AA Change	Total Depths	VAF	Clinvar
MSH6	NM_000179.3	p.Phe1088fs	2,054	3.85%	
XPC	NM_004628.5	p.Glu340del	1,698	5.06%	
APC	NM_001354896.2	p.Ala2140del	2,043	0.54%	
MET	NM_001127500.3	p.Val1088Met	2,297	48.89%	
WRN	NM_000553.6	p.Glu510del	1,164	2.84%	
PRKDC	NM_006904.7	p.Ile166fs	760	5.26%	
MUC6	NM_005961.3	p.Thr14611le	2,803	9.99%	
KMT2D	NM_003482.4	p.Gln3935del	1,187	0.34%	
KMT2D	NM_003482.4	p.Gln3735*	1,319	0.53%	
KMT2D	NM_003482.4	p.Gln1092dup	1,792	0.34%	
BLM	NM_000057.4	p.Asn515fs	1,646	1.82%	
BLM	NM_000057.4	p.Asn515fs	1,659	2.59%	
CREBBP	NM_004380.3	p.Gln2203del	1,219	0.57%	
CDH1	NM_004360.5	p.Pro147Ser	1,713	0.29%	
SMARCA4	NM_001128849.3	rs238_Gly239insGlyProGly	1,665	0.24%	



Sarcoma NGS Panel Development with Celemics

CELEMICS

Target-enrichment capturing Next-Generation Sequencing panel for screening sarcoma associated driver mutations

JW Ahn[1]*, JH Kim[2]*, BR Kim[1], Min[1], HG Kang[2], SJ Kim[2], T Yun[2], WY Choi[2], YG Suh[2], SY Park[2], HJ You[2], EY Lee[2], YN Kim[2], JY Sung[2] and KW Kim[2]

Gene	Position	Mutation	Mut1	Mut2	Mut3	Mut4	Mut5	Mut6	Mut7	Mut8	Mut9	Mut10	Mut11	Mut12
MTOR	chr1:11174511	7165-1G>T	31.14	16.04	8.68	2.96	1.21	1.17	0.62	0.41	0.18	0.10	0.10	0.09
MTOR	chr1:11181327	Leu2303Leu	15.55	8.69	5.00	1.54	0.66	0.55	0.30	0.16	0.07	0.06	0.03	NA
MTOR	chr1:11190646	Ser1851Ser	17.07	8.29	4.91	1.90	1.15	0.86	0.55	0.43	0.32	0.27	0.27	0.23
JAK1	chr1:65310489	Pro733Pro	38.73	20.89	11.59	3.48	1.77	1.24	0.51	0.27	0.21	0.09	0.06	0.03
JAK1	chr1:65312342	Arg659Arg	29.12	16.11	8.88	3.46	1.24	1.03	0.56	0.31	0.31	0.19	0.17	0.15
JAK1	chr1:65332696	Tyr281Tyr	11.40	4.71	2.96	0.94	0.44	0.43	0.22	0.21	0.12	0.12	0.10	0.08
MSH6	chr2:48018236	Ser144Ile	30.07	15.68	8.45	2.48	1.44	0.92	0.56	0.31	0.23	0.18	0.11	0.10
MSH6	chr2:48027921	Phe933Phe	26.92	13.06	7.38	1.78	0.95	0.58	0.37	0.24	0.19	0.12	0.06	0.04
RAF1	chr3:12645007	Ala288Ala	27.30	15.29	8.15	2.33	1.08	1.03	0.52	0.34	0.27	0.19	0.11	0.09
PIK3CA	chr3:178936091	p.E545K	7.39	4.43	2.58	0.91	0.47	0.25	0.14	0.10	0.05	NA	NA	NA
PIK3CA	chr3:178947865	Gly914Arg	31.57	16.21	8.68	2.83	1.45	0.85	0.56	0.24	0.18	0.07	0.06	0.05
PIK3CA	chr3:178952085	His1047Arg	16.95	8.07	4.74	1.38	0.63	0.42	0.16	0.15	0.12	0.10	0.06	NA
PDGFRA	chr4:55127413	Ser92Ser	9.72	4.95	2.85	0.97	0.44	0.36	0.18	0.16	0.07	0.06	0.05	0.03
PDGFRA	chr4:55138600	Gly451Asp	32.26	14.91	9.12	3.10	1.48	1.05	0.45	0.37	0.16	0.12	0.06	0.06

100% >
20% >
5% >
1% >
0.5% >
0.2% >

Detection sensitivity test result in 80 mutations from reference sample

Genetics and Molecular Research, 2024

NCC Korea Sarcoma-Specific CGP Panel

CELEMICS

CDS gene (129)

ATRX, CIC, ERG, ETV1, ETV4, EWSR1, FLT4, JUN, MDM2, NF1, PTEN, RB1, STAG2, TP53, SAGE1, FUS, FES, SS18, KRAS, CRKL, MOS , FGR, BRAF, ARAF, FAU, CRK, YES1, SKI, HRAS, BRAFP1, ASPSCR1, KRASP1, SS18L2, SS18L1, KIT, SRC, SS18L2P1, SS18L2P2, SS X1, SSX8, SSX4B, SSX3, SSX6, SSX5, SSX9, SSX7, SSX4, SSX2, SSX2B, SSX2IP, WRN, REQL4, BLM, RPS19, RPL5, RPL11, RPL35A, RPS24, RPS17, RPS7, PDGFRA, ROS1, KMT2D, SETD2, ERBB4, MAX, SMO, ARID1B, CREBBP, FANCA, KDR, KMT2A, MTOR, PIK3CA, SDHB, TET2, TSC2, APC, ASXL1, ATM, BRD4, CARD11, DICER1, GLI1, GSTM5, TCF7L2, ABL1, BRCA1, CDH1, CHEK2, CRTC1, CYLD, E P300, EPHA7, FBXW7, FGFR2, FLT3, JAK1, MET, MSH6, PARK2, PIK3C2B, PIK3R2, PRKAR1A, RAD21, RAF1, RARA, RECQL4, SH2B3, SMAD2, SMARCA4, SUZ12, TCF3, TRAF7, TSC1, XPC, ZNF217, ZNF708, MYOCD, IDH1, IDH2, LYN, PRKDC, CDK4, CDKN2A, CDKN2B, MUC16, OTOP1, MUC6, LRP4, MUC3A, SALL3, WFIKKN1, ANKRD30B, ATG2A, DENND4B

Fusion gene (147)

ACTB, AHRR, ALK, ASPSCR1, ATF1, ATIC, BCOR, BRD3, BRD4, CAMTA1, CARS, CCNB3, CDH11, CDX1, CD63, CEP128, CIC, CITED2, CLTC, CNBP, COL1A1, COL1A2, COL3A1, COL6A3, CREB1, CREB3L1, CREB3L2, CSF1, CXorf67, C11orf95, DDIT3, DUX4, DVL2, EML4, EPC1, EP400, ERG, ETV1, ETV4, ETV6, EWSR1, FEV, FGFR1, FLI1, FN1, FOSB, FOXO1, FOXO4, FUS, GLI1, HAS2, HEY1, HMGA2, IRF 2BP2, JAZF1, KIRREL, KLF17, LAMTOR1, LPP, MAML3, MBTD, MEAF6, MED12, MIR143HG, MKL2, MYH9, NAB2, NCOA1, NCOA2, NFAT C2, NFIB, NOTCH1, NOTCH2, NR4A3, NTRK1, NTRK3, NUMA1, NUTM1, NUTM2B, OMD, OPHN1, PATZ1, PAX3, PAX7, PBX1, PBX3, PD GFB, PDPN, PHF1, PLAG1, PLPP3, POU5F1, PPFIBP1, PRDM10, PRKCA, PRKCB, PRKCD, RAB2A,RAD51B, RANBP2, RNF213, RRAGB , SEC31A, SERPINE1, SETBP1, SFMBT1, SMARCA5, SP3, SQSTM1, SRF, SRSF3, SSX1, SSX2, SSX4, SS18, SS18L1, STAT6, SUZ12, S 100A10, TAF15, TCF12, TEAD1, TFE3, TFG, THRAP3, TPM3, TPM4, TPR, USP6, VCL, VGLL2, WT1, WWTR1, YAP1, YWHAE, ZC3H7B, Z FP36, ZNF444, ASPL

Selected based on literature review and molecular tumor board consensus

Sarcoma Gene Assay Results

352 cases: 67 bone and 285 soft tissue sarcomas

Diagnostic refinement in 14 cases (3.6%)

Indels and CNVs: TP53 (36%), MDM2 (12%), RB1 (14%) and CDK4 (10.5%)

Potentially actionable kinase fusions (3%): ALK (3), NTRK1–3 (2), RET (2), ROS1 (1) and BRAF (1)

Germline findings pending



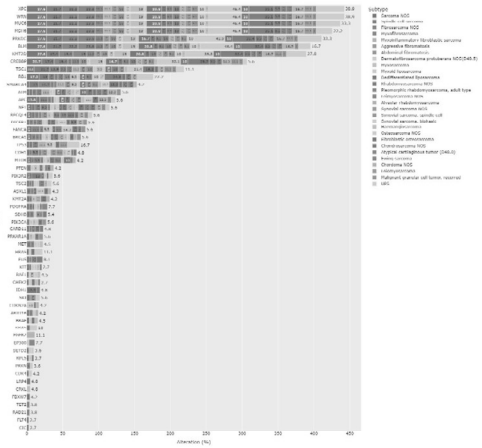
Celemics
Sarcoma Panel

The First Step Towards
Personalized Medicine for Rare Cancers

Key Features

- Molecular diagnostic panel for Sarcoma, including 129 clinically significant genes
- High capture efficiency ensuring low-VAF analysis performance





Manuscript in preparation

The Founding of KINGS

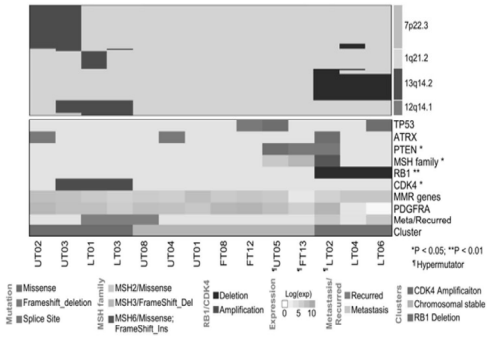
Korean Innovative Group for Sarcoma Research

KINGS brings researchers from different disciplines together to use BICSWAN resources, develop joint studies and advance precision medicine for patients with sarcoma.

KINGS
한국육종연구그룹
KOREAN INNOVATIVE GROUP
FOR SARCOMA RESEARCH

Foundations of the NCC Sarcoma Research Program





Complex-karyotype STS, BMC Med Genet (2018)

NCC Korea Sarcoma Research Group (2016–)

Clinicians and basic scientists analyzed banked sarcoma tissue and identified genomic and transcriptomic features across histologic subtypes.

Prospective biospecimen banking

The Rare Cancer Center's largest biospecimen collection established prospective protocols for fixation, freezing and single-cell isolation.

Standardized clinical database

An NCC-wide inventory, SOPs and e-Velos eCRFs captured ~1,500 retrospective records through March 2017.
The registry launched in 2018 with automated EHR transfer, weekly updates and quality checks.

Clinical and Laboratory Research at NCC Korea

국립암센터 NATIONAL CANCER CENTER

Article

Clinical Characteristics of Complex Karyotype Soft Tissue Sarcomas: A Single-Institution Cohort Study

Eun-Young Lee [1,†], June Hyuk Kim [2,3,4,†], Jong Woong Park [2,3,5], Hyun Guy Kang [2,3,4], Seog-Yun Park [6], Jiyu Sun [5,7,8], Seo-Young Kim [8], Ahyoung Cho [1], Bora Lee [2] and Hye Jin You [1,4,*]



Hye Jin You, PhD

Lee et al. Medicina. 2026

Clinical features and survival in three complex-karyotype soft tissue sarcoma groups: undifferentiated sarcoma (mainly UPS), myxofibrosarcoma, and leiomyosarcoma of soft tissue

The cohort provides a basis for linking clinical outcomes to molecular and multi-omics data

International Journal of *Molecular Sciences* — MDPI

Article

Biomarker candidates of complex karyotype soft tissue sarcomas with *CDK4*-amplification for better therapeutic strategies

Eun-Young Lee[1], Hyun Sang Cho[1], June Hyuk Kim[2,3], Hyun Guy Kang[2,3], Jong Woong Park[3,4], Ahyoung Cho[1,5], and Hye Jin You[1,2,*]

Lee et al. Int.J.Mol.Sci. 2025

cancers — MDPI

Article

New Established Cell Lines from Undifferentiated Pleomorphic Sarcoma for In Vivo Study

Eun-Young Lee [1,*], Young Ho Kim [2,*], MD Abu Rayhan [1], Hyun Guy Kang [3,4], June Hyuk Kim [3,4], Jong Woong Park [4,5], Seog Yun Park [6], Minjeong Kim [1] and Hye Jin You [1,3,†]

Lee et al. Cancers 2022.

KINGS Multidisciplinary Research Network (est. 2022)



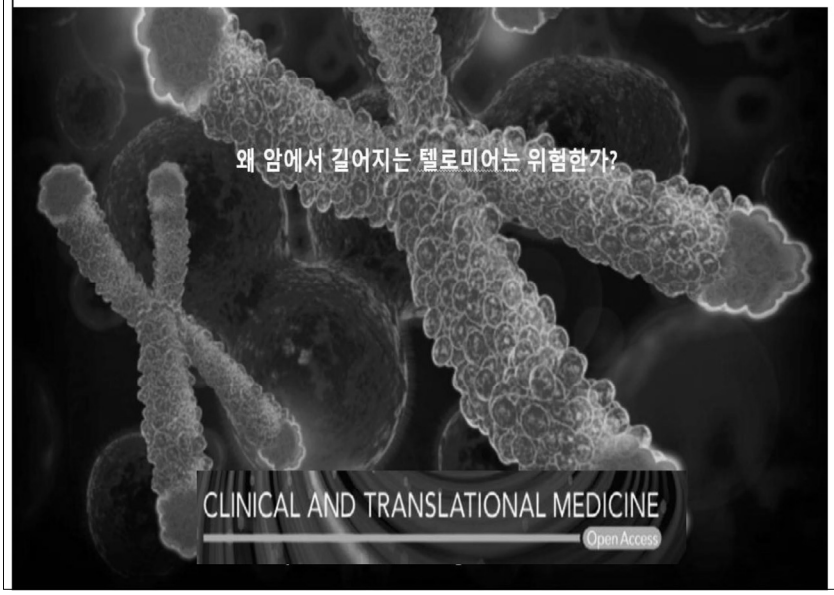
First KINGS Study with an External Collaborator

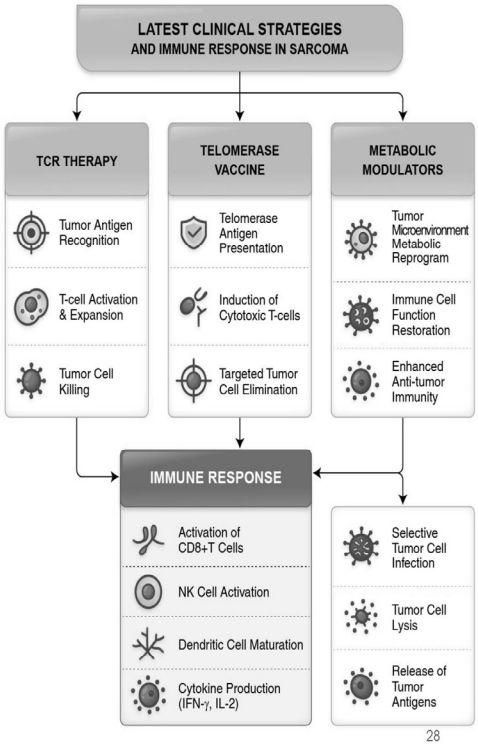
국립암센터 NATIONAL CANCER CENTER

Sung and Kim, Clinical and Translational Medicine, 2025

Telomere–metabolism–immunity axis in sarcoma: Immune evasion mechanisms and therapeutic strategies

Ji-Yong Sung[1,2] | June Hyuk Kim[3]





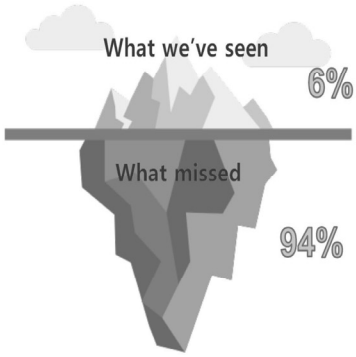
Ongoing BICSWAN and KINGS Studies

BICSWAN
육종 혁신형 바이오뱅킹 컨소시엄
BIOBANK INNOVATIVE CONSORTIUM FOR SARCOMA WITH ADVANCEMENTS AND NETWORKING

KINGS
한국육종연구그룹
KOREAN INNOVATIVE GROUP FOR SARCOMA RESEARCH

Sarcoma Whole-Genome Sequencing with NIH Korea

질병관리청 국립보건연구원



Rare variants
(< 1% among the people)

UKB, ChinaMAP, TOGOVAR

	WGS analyzed			
Subtype	T-N Paired	T Only	N Only	Total
Bone sarcoma	63	39	1	103
Soft tissue sarcoma	131	21		152
Borderline	26	5		31
Total	**220**	**65**	**1**	**286**

Rationale

WGS extends coverage of CNAs, SVs and noncoding variants

Genomic instability may be central to sarcoma biology

Goal

To identify novel oncogenic variants and for molecular subtyping

~100X

Analysis and clinical linkage

1. Profile SNVs, indels, CNAs, SVs, fusions, rearrangements and chromothripsis
2. Link matched bulk RNA-seq to target-gene expression and ECM remodeling, DNA damage, immune and cell-cycle signatures
3. Identify candidate biomarkers for metastasis, recurrence and survival.
4. Use matched snRNA-seq to characterize cellular composition and the tumor microenvironment

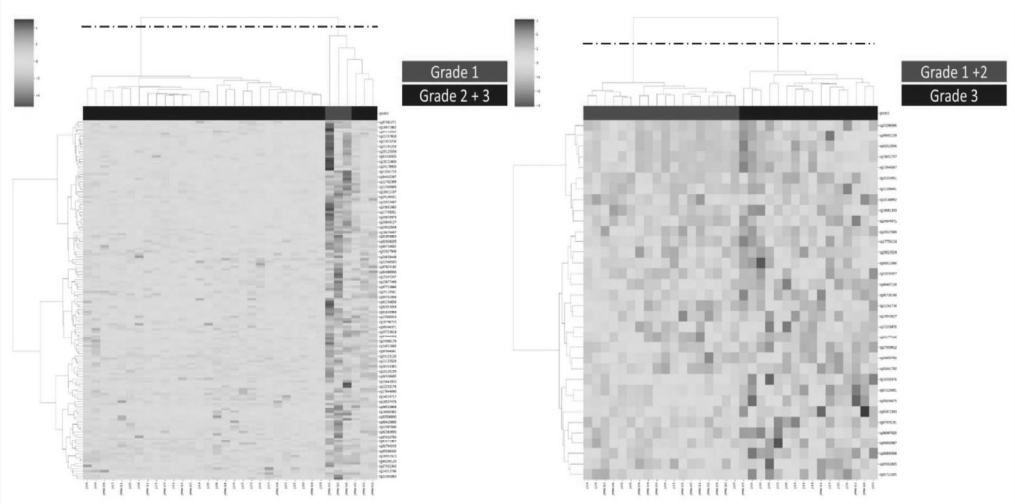
*Methylation analysis considered to be added

Sarcoma Whole-Genome Sequencing with NIH Korea

Can DNA Methylation Profiling Classify Histologic Subtypes and Grades in Soft Tissue Sarcoma?

Hyunho Kim MD, PhD[1], Min Wook Joo MD, PhD[2], Joohee Yoon MD, PhD[3], Hong Sik Park MD[4], June Hyuk Kim MD, PhD[5], Joo Hwan Lee MD[6], Sung Hwan Kim MD, PhD[6], Seul Ki Lee MD, PhD[7], Yang-Guk Chung MD, PhD[8], Yoon Joo Cho MD, PhD[2]

Kim et al. Clin Orthop Relat Res, 2024



Cluster analyses represent each tumor grade according to the DNA methylation pattern

Proteogenomics of Synovial Sarcoma and ASPS

- Combine protein and RNA data to identify high-risk groups in adolescent and young adult sarcoma.

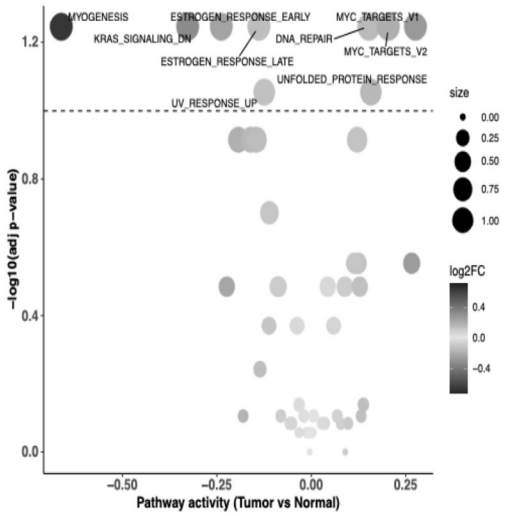


Prof. Yong Wook Kim

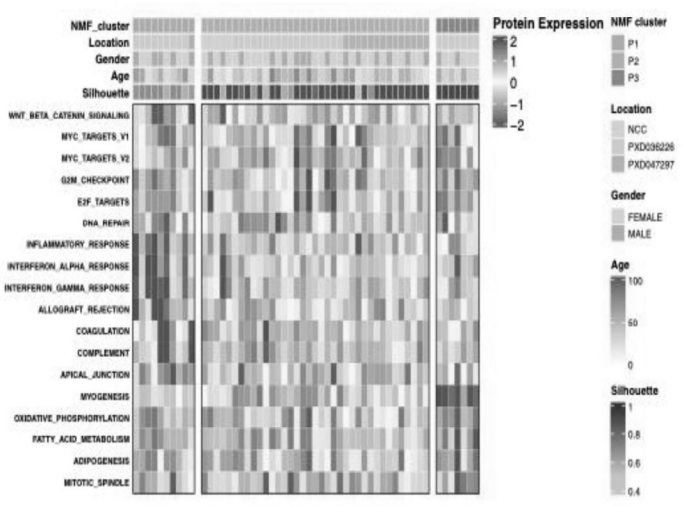


Hye Jin You, PhD

ASPS



Synovial sarcoma



Manuscript in preparation.

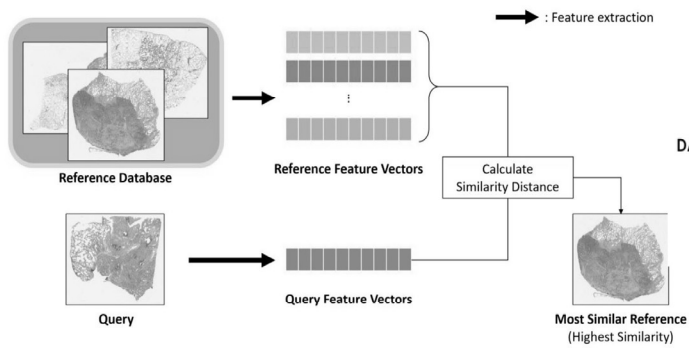
Digital Pathology for Sarcoma Diagnosis


Pf. Yoon Rha Choi


Pf. David Ho


Pf. Seok Yeon Park

General Concept of Reverse Image Search



scientific data

Article in Press

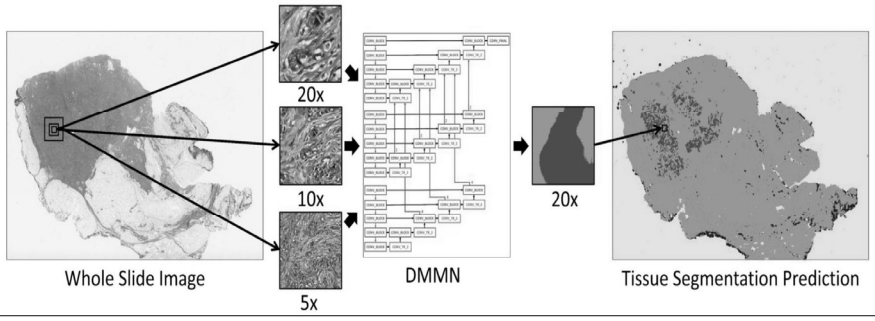
OPEN

DATA DESCRIPTOR

A region-level H& E histopathology image dataset for bone and soft tissue tumors

Gyu Yeong Kim[1,15], Yongjun Jeon[2,3,15], Hoyeon Jeong[4,8,9,14,15], Donggeon Lee[5,9,14], Seungkyun Lee[1], Hyungbin Kim[1], Yurimi Lee[1], Jihwan Kim[9,14], Seog-yun Park[10], Kyu-Hwan Jung[2,3], David Joon Ho[6,7], June Hyuk Kim[11,12,13] & Yoon-La Choi[1,6,9,14,15]

Kim et al., Scientific Data, 2026



MRI Segmentation and Radiomics


Pf. Chae Jung Park

An automated MRI segmentation and standardized radiomics pipeline for bone and soft tissue sarcomas

Image collection and QC

Index and de-identify DICOM data.
Identify imaging sequences.

Standard preprocessing

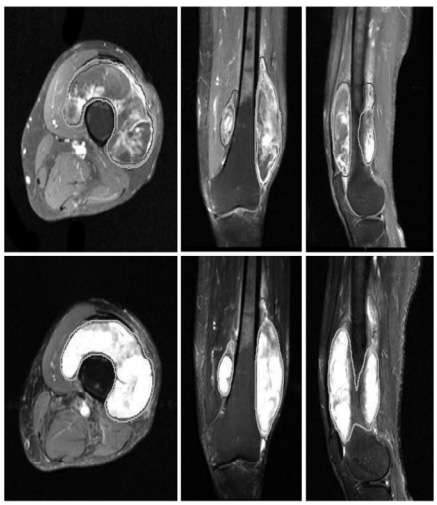
Normalize each MRI sequence.
Check CT and PET measurements.

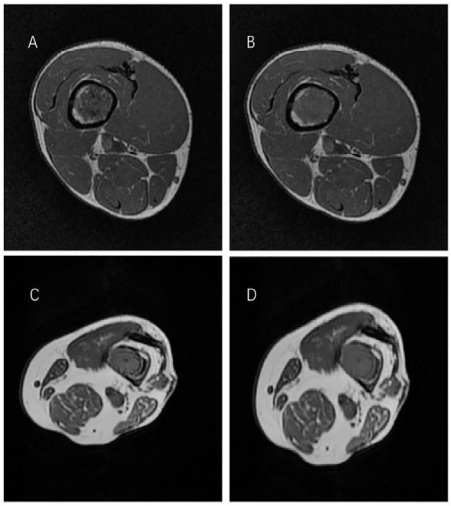
Expert annotation

Mark 3D tumor boundaries, peritumoral regions, and necrosis.

AI and external testing

Build a baseline model and a feature repository. Test on external data.





Updating the BICSWAN Sarcoma CGP Panel

Panel Gene Utility Score (PGUS)

Diagnostic value + actionability + cohort yield + subtype specificity + assay suitability

Evidence from approximately 350 previous cases

- Patients with pathogenic or likely pathogenic alterations
- Patients with variants of uncertain significance (VUS)
- Recurrent hotspots and copy-number alteration yield
- Enrichment within specific histologic subtypes

Gene	Current panel	Recommendation	Priority	Sarcoma relevance	Molecular role
ABL1	Current 129	KEEP	Moderate	Multiple/unspecified sarcoma context	Cancer-related / exploratory
ANKRD30B	Current 129	REMOVE	Low	Multiple/unspecified sarcoma context	Cancer-related / exploratory
APC	Current 129	KEEP	High	Desmoid/FAP context	WNT tumor suppressor
ARAF	Current 129	KEEP	Moderate	Multiple/unspecified sarcoma context	Cancer-related / exploratory
ARID1R	Current 129	KEEP	Moderate	Multiple/unspecified sarcoma context	Cancer-related / exploratory
ASPSCR1	Current 129	MOVE TO RNA	High	ASPS / TFE3-rearranged tumors	Fusion/rearrangement partner
ASXL1	Current 129	KEEP	Moderate	Multiple/unspecified sarcoma context	Cancer-related / exploratory
ATG2A	Current 129	REMOVE	Low	Multiple/unspecified sarcoma context	Cancer-related / exploratory
ATM	Current 129	KEEP	High	DDR subset	DDR signaling
ATRX	Current 129	KEEP	High	LMS, MPNST, complex STS	Chromatin/telomere regulation
BLM	Current 129	KEEP	High	Cancer predisposition / DDR	DNA helicase
BNIP5	Current 129	REMOVE	Low	Multiple/unspecified sarcoma context	Cancer-related / exploratory
BRAF	Current 129	KEEP	High	Selected sarcomas	MAPK driver

Use the findings to design BICSWAN Sarcoma CGP Panel v2.

Target Discovery and Drug Screening

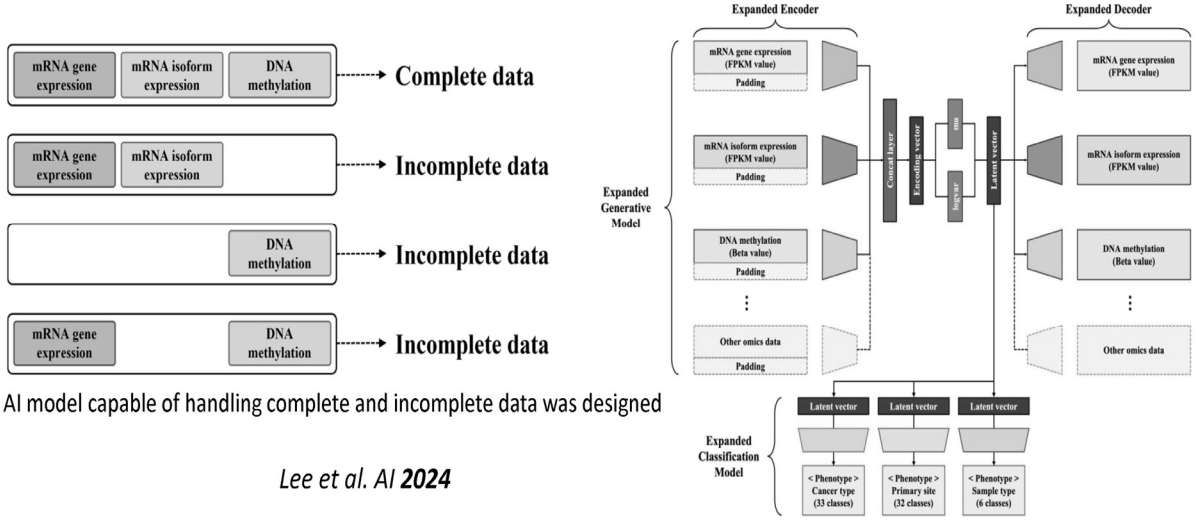
AI

MDPI

Article

Generative Models Utilizing Padding Can Efficiently Integrate and Generate Multi-Omics Data

Hyeon-Su Lee [1], Seung-Hwan Hong [1], Gwan-Heon Kim [1], Hye-Jin You [2,3], Eun-Young Lee [2], Jae-Hwan Jeong [4], Jin-Woo Ahn [4] and June-Hyuk Kim [5,*]



AI model capable of handling complete and incomplete data was designed

Lee et al. AI ***2024***

Target Discovery and Drug Screening



Prof. Sung Wook Seo



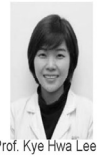
Prof. Joo Sang Lee



Prof. Jin Hong Kim



Prof. Yi Joon Kim



Prof. Kye Hwa Lee

Fusion discovery and clinical linkage

Prioritize sarcoma-specific fusions and assess outcomes by alteration status

AI and computer-aided drug design

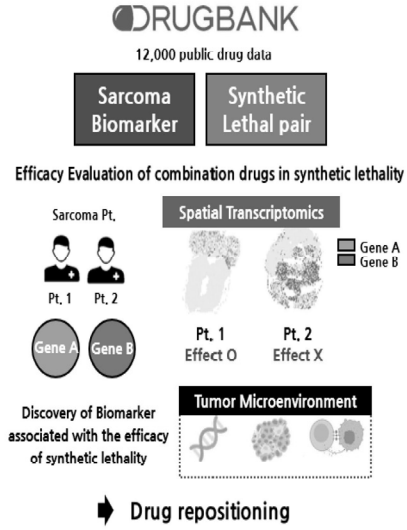
Validate fusion and variant protein structures. Identify binding si tes and run two-stage virtual screening.

Optimize peptide and small-molecule leads, study structure–activity relati onships, and test candidates in cell lines as they advance toward TRL 5.

특허출원(10-2025-0100442)

[VEGFR2 및 TIE2를 이중 타겟하여 억제하는 신규 화합물 및 이를 포함하는 암 예방 또는 치료용 약학 조성물]

Compound Screening for Sarcoma

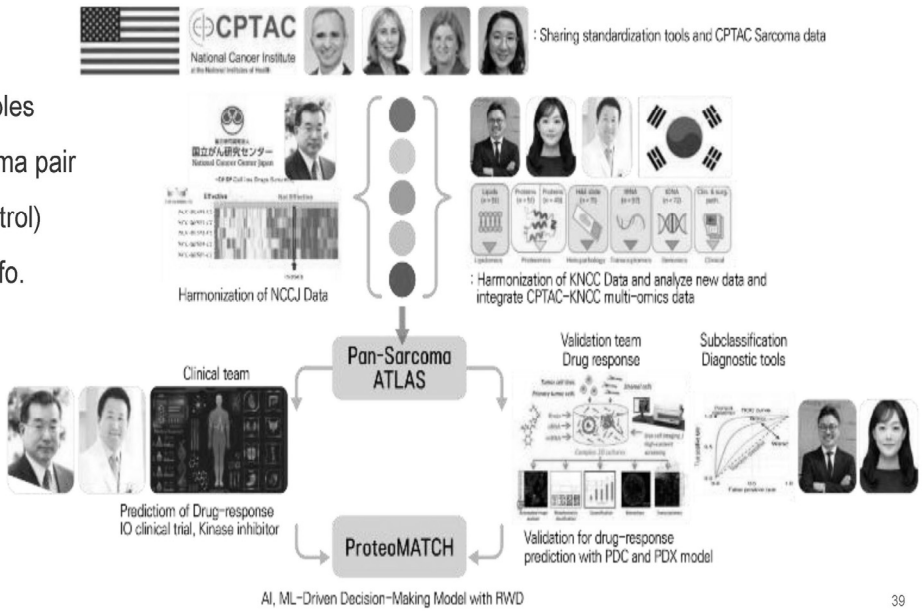


Unpublished Data

Pan-Sarcoma Proteomics Atlas

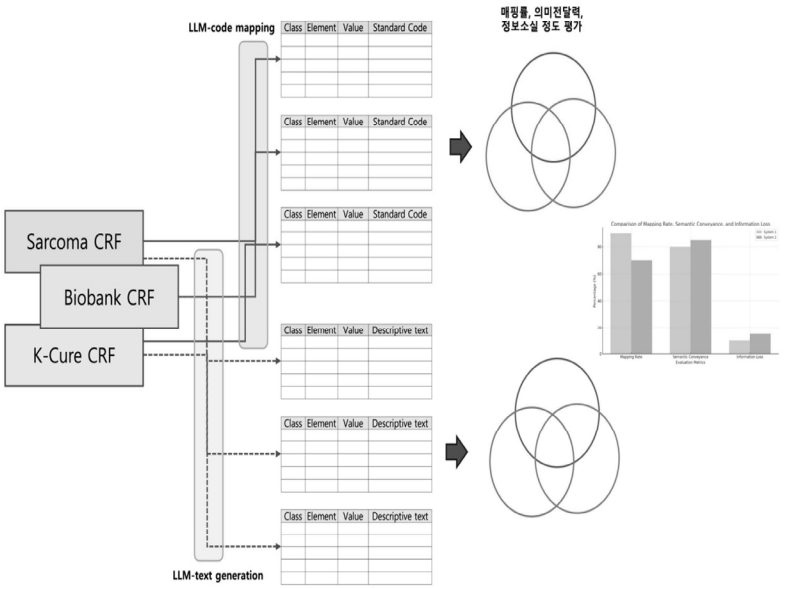
An ongoing Korea–Japan–US study involving NCC Korea, NCC Japan, and the US National Cancer Institute.
Profile protein expression and post-translational modifications to identify potential therapeutic targets.

- NCCJ: 649 sarcoma samples
- NCCK: 376 sarcoma-plasma pair
  (add. 34 normal control)
  w/ detailed clinical info.
- NCI: CPTAC protocol



Expansion into a National Research Project on Rare Cancers

임상 연구 증례기록서 데이터의 의미론적 상호운용성 향상을 위한
전통적 표준 용어 매핑(예: SNOMED CT, LOINC)과 텍스트 생성 기반 매핑 방식 비교 연구



특성	NCC Sarcoma DB	KBN_CDM
환자 수	66명	별도 환자군
필드 수	208개	398개
결측치 비율	변수에 따라 다양함 (0~100%)	제공된 통계에서는 명확하게 제시되지 않음
강점	추적 관찰 데이터 (생존, 재발, 전이)	상세 병력, 가족력, 임상검사 결과
한계	치료 데이터의 높은 결측치	추적 관찰 데이터 부족

Unpublished Data

"Let's collaborate. Why not?"

"If you want to go quickly, go alone. If you want to go far, go together."
African proverb

Thank you

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