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PROJECT GENIE®
Genomics Evidence Neoplasia Information Exchange

GENIE BPC PANC1.0-PUBLIC COHORT

MARCH 2025

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Release Notes

- The GENIE BPC PANC 1.0-public dataset contains 1,109 PANC patients from MSKCC, DFCI, VICC, and UHN.
- **Data Access:**
  - A subset of the data is available through cBioPortal. Both the genomic and clinical (phenomic) data can be evaluated in cBioPortal with opportunities for data exploration and visualization using a user-friendly interface.
  - The complete, post-processed data are available on Synapse.

- **What is included in GENIE BPC data?**
  - **Genomic Data:** Clinical-grade next-generation sequencing data for each patient from the GENIE Registry. Genomic profiling was performed between 2013 and 2018; patients were aged 24-88 at the time of genomic sequencing.
  - **Cancer Diagnosis:** Pancreatic cancer diagnosis is considered the index tumor for this patient cohort. There are data about other cancer diagnoses antecedent and subsequent to the pancreatic cancer.
  - **Pancreatic cancer-specific fields:** includes tumor resectability status (Resectable, Borderline Resectable, Unresectable/Locally Advanced or Metastatic) at the time of clinical staging prior to any cancer-directed treatment, were also collected.
  - **Pathologic Information:** Each pathology specimen from diagnosis through death or last follow-up is curated with specimen type, site, and histology.

Release Notes

- **Treatment Histories:** All anti-neoplastic systemic therapies–intravenous and oral chemotherapies–are included in the data set. Dates are provided as intervals from diagnosis to start and stop of each drug. Investigational drugs are masked; no dosing information is included.
- **Imaging Information:** Each CT, MRI, PET-CT scan from diagnosis through death or last follow-up is curated for the presence or absence of cancer and an evaluation of whether the cancer was stable, responding, or progressing. These data are used to compute progression-free survival-imaging (PFS-I). Sites of tumor involvement are also recorded.
- **Medical Oncologist's Evaluations:** Medical oncology notes (1/month) have been curated to ascertain the presence or absence of cancer and whether the cancer was stable, responding, or progressing. These data are used to compute progression-free survival-medonc (PFS-M) from diagnosis through death or date of last follow-up.
- **Overall Survival:** Overall survival is based on death, with censoring at the date last known alive. Ascertainment of death varies by institution.
- **Additional Relevant Biomarkers:** Information about select biomarkers not included on the NGS panels, including PD-L1, and MMR are also curated.
- **Patient-Reported Outcomes:** No patient-reported outcomes are available in this dataset.
- **Date Masking:** Exact dates are masked to preserve confidentiality; however, date intervals are available, allowing calculation of event times such as diagnosis, treatment start, treatment end, PFS-I, PFS-M, and OS

Release Notes

- **Analytical Data Guide:** A more comprehensive overview of the data can be found in the data guide, and a description and location of the variables collected can be found in the variable synopsis spreadsheet.

- **Other Resources:** There is a dedicated project wiki that describes each of the files.

- **Training Videos**:
  - Demo of GENIE Data on the Synapse and cBioPortal Platforms: here
  - BPC- specific cBioPortal video training playlist: here

- **PRISSMM™:** the BPC PANC dataset uses the PRISSMM™ system licensed and enhanced by Memorial Sloan-Kettering Cancer Center, Memorial Hospital for Cancer and Allied Diseases, and Sloan-Kettering Institute for Cancer Research (collectively "MSK") is for informational and research purposes only. The content is not intended as a substitute for professional medical advice, diagnosis, or treatment. Original system and improvements © 2019-2022 Dana-Farber Cancer Institute, Inc. Additional functionality and enhancements © 2023 MSK. All rights reserved. Additional information can be found in the analytic data guide and information about licensing PRISSMM™ can be obtained by emailing PRISSMM@mskcc.org

GENIE BPC Data Model

Core Patient-Level Data
- Demographics
- Smoking history
- Vital status

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Biomarkers

Pan-cancer biomarkers
- PD-L1
- MMR

Cancer Diagnosis-specific attributes
- Histology
- Stage
- Resectability status

Treatment Data

Systemic Therapies (Rx)
- International Nonproprietary Name (INN)
- Start and stop dates

Lines of therapy
- Standardized treatment ontology
- HemOnc.org (Creative Commons license)

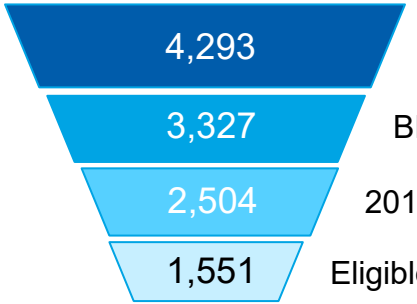
Clinical Outcomes

PRISSMM

rwOS – Real-world Overall Survival
rwDFS – Disease-Free Survival
rwPFS – Progression-Free Survival
rwTTNT – Time to Next Treatment
rwTTD – Time to Treatment Discontinuation
rwTTR – Time to Response

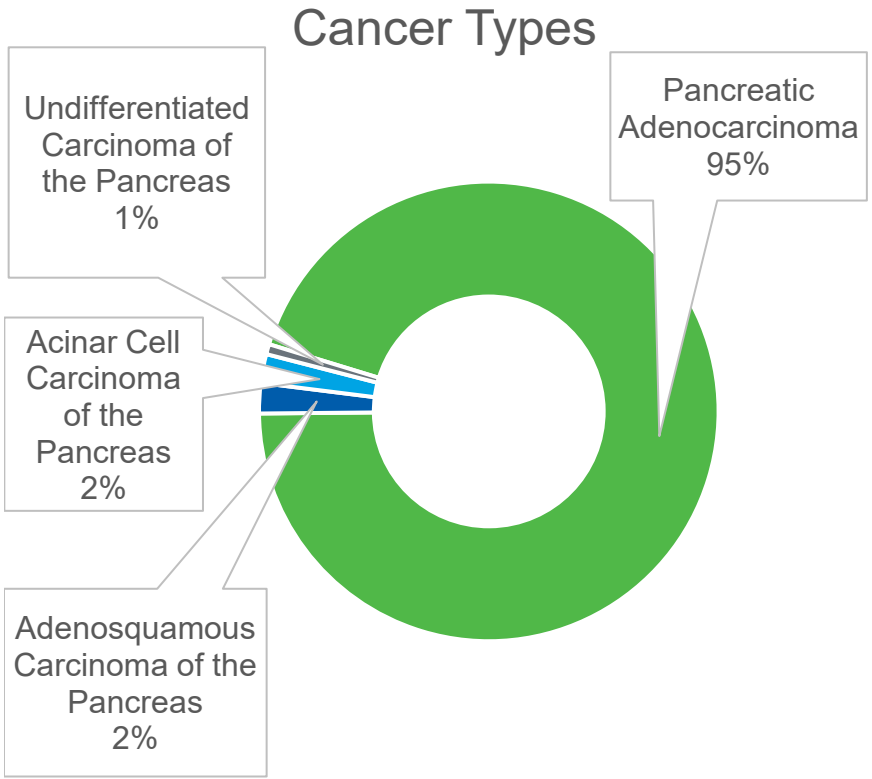
PRISSMM™ is licensed from the Dana-Farber Cancer Institute.

BPC PANC 1.0-public: Cohort



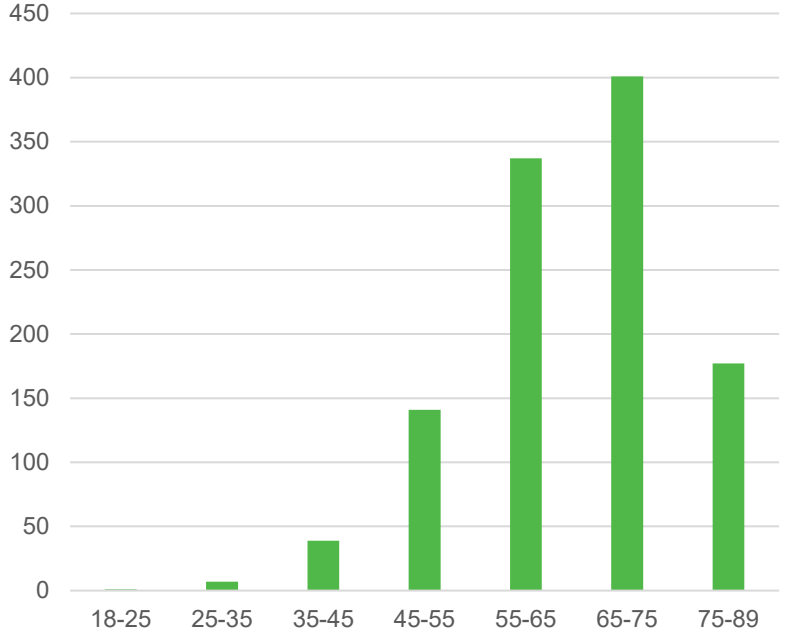
1,109 final cohort

Pancreatic Adenocarcinoma	1,075
Adenosquamous Carcinoma of the Pancreas	24
Acinar Cell Carcinoma of the Pancreas	23
Undifferentiated Carcinoma of the Pancreas	8

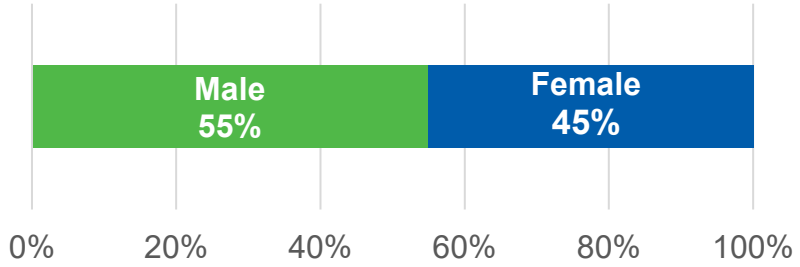


BPC PANC 1.0-public: Demographics

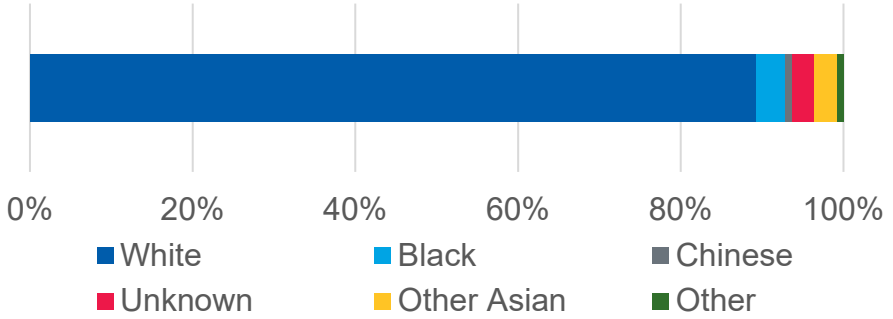
Age at Diagnosis



Sex

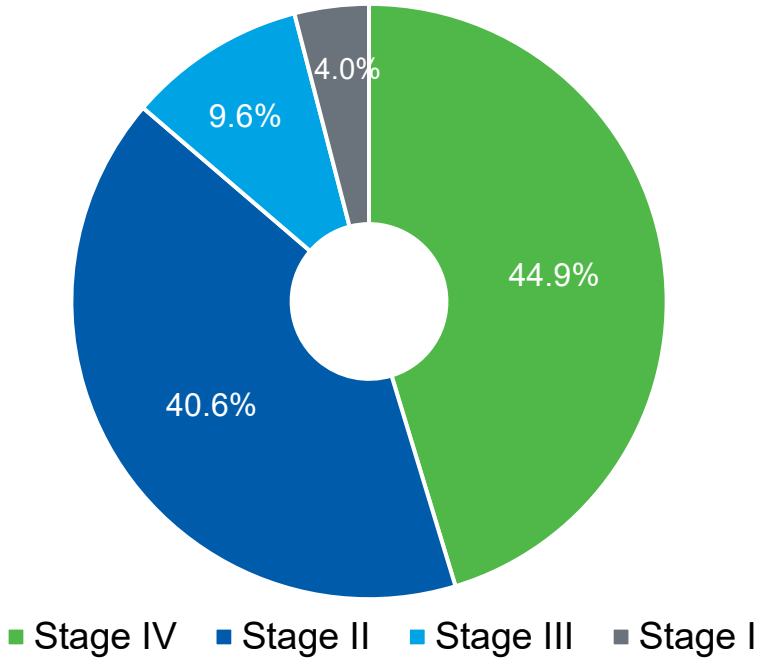


Primary Race

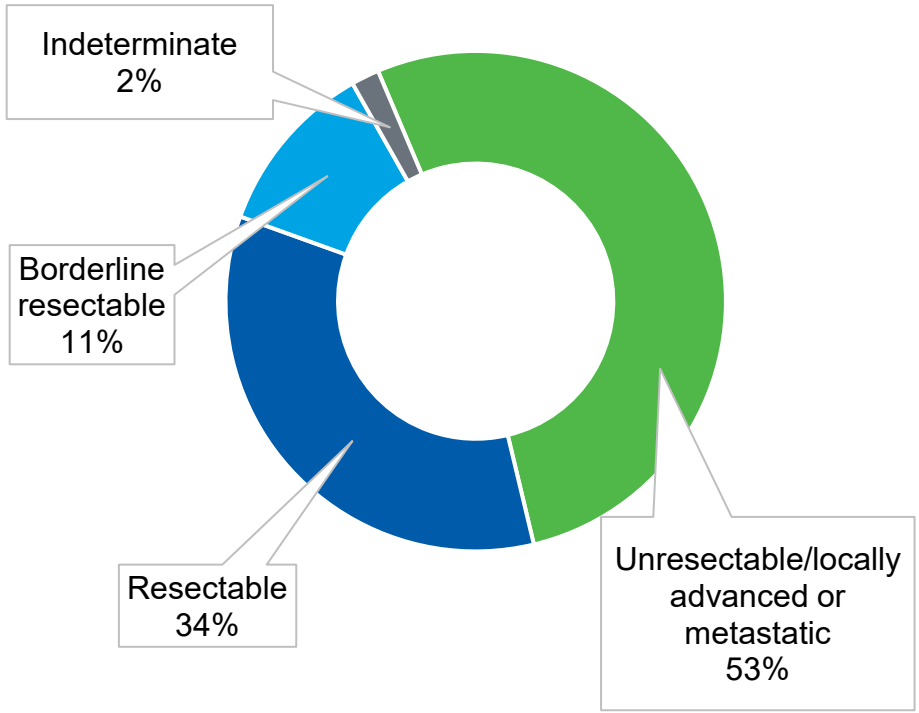


BPC PANC 1.0-public: Stage at Diagnosis and Resectability

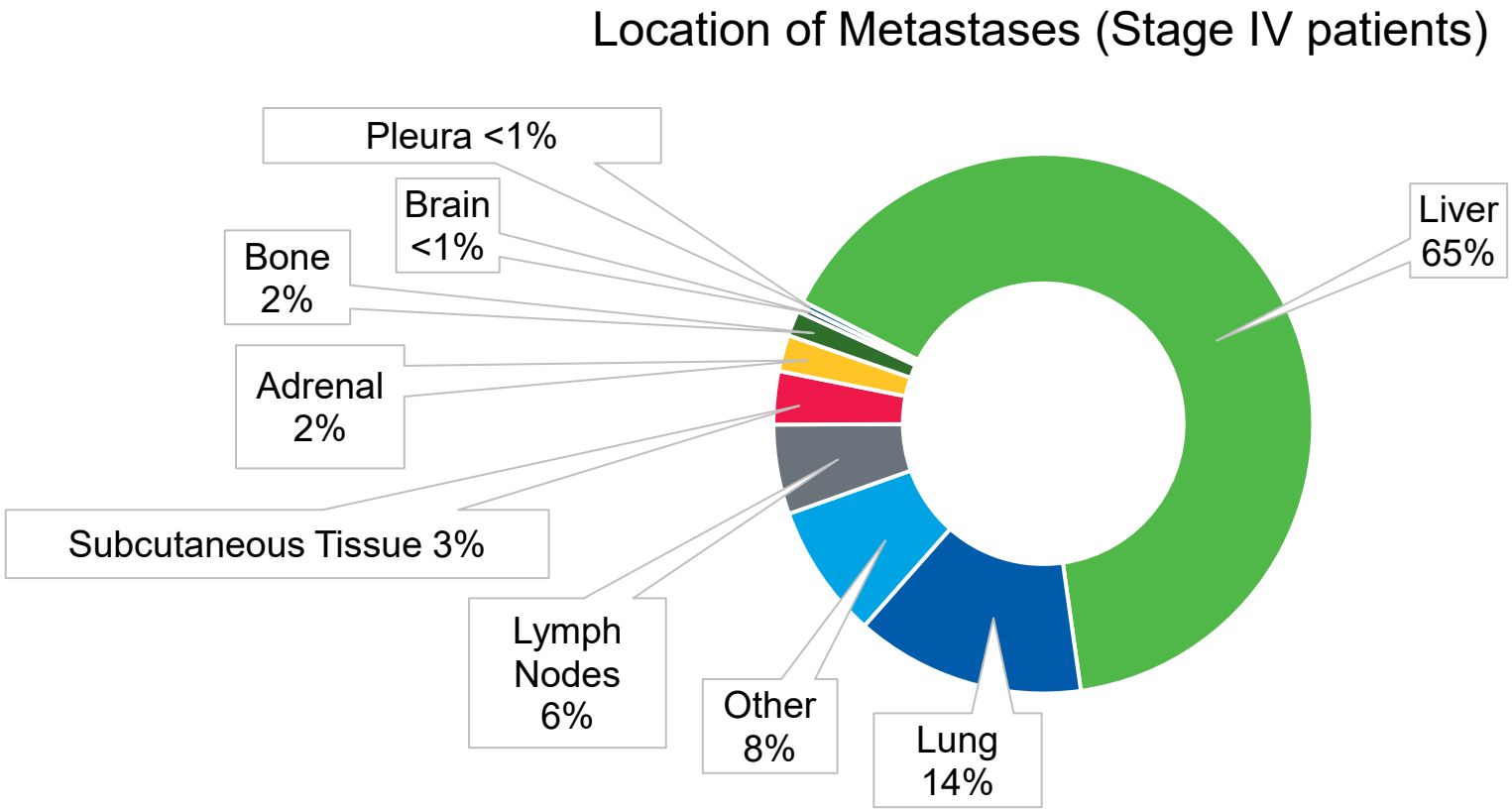
Stage at Diagnosis



Tumor Resectability Status



BPC PANC Cohort:
Sites of Metastases at Diagnosis



BPC PANC Cohort: Detailed Clinical Genomics

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Mutated Genes

Mutated Genes (1130 profiled samples)

Gene	# Mut	#	Freq
KRAS	979	970	85.8%
TP53	796	788	69.7%
DNAH9	14	11	64.7%
SYNE1	16	9	52.9%
RNF213	12	9	52.9%
PDE4DIP	10	9	52.9%
NCOR2	12	8	47.1%
NIN	8	8	47.1%
CDH23	8	6	35.3%
SAMD9	6	6	35.3%
NUMA1	6	5	29.4%
TGM7	6	5	29.4%
LIFR	5	5	29.4%
PKHD1	6	4	23.5%
CACNA1E	5	4	23.5%
EP400	5	4	23.5%
ANKRD24	4	4	23.5%

Structural Variants

Structural Variant Genes (1061 profiled samples)

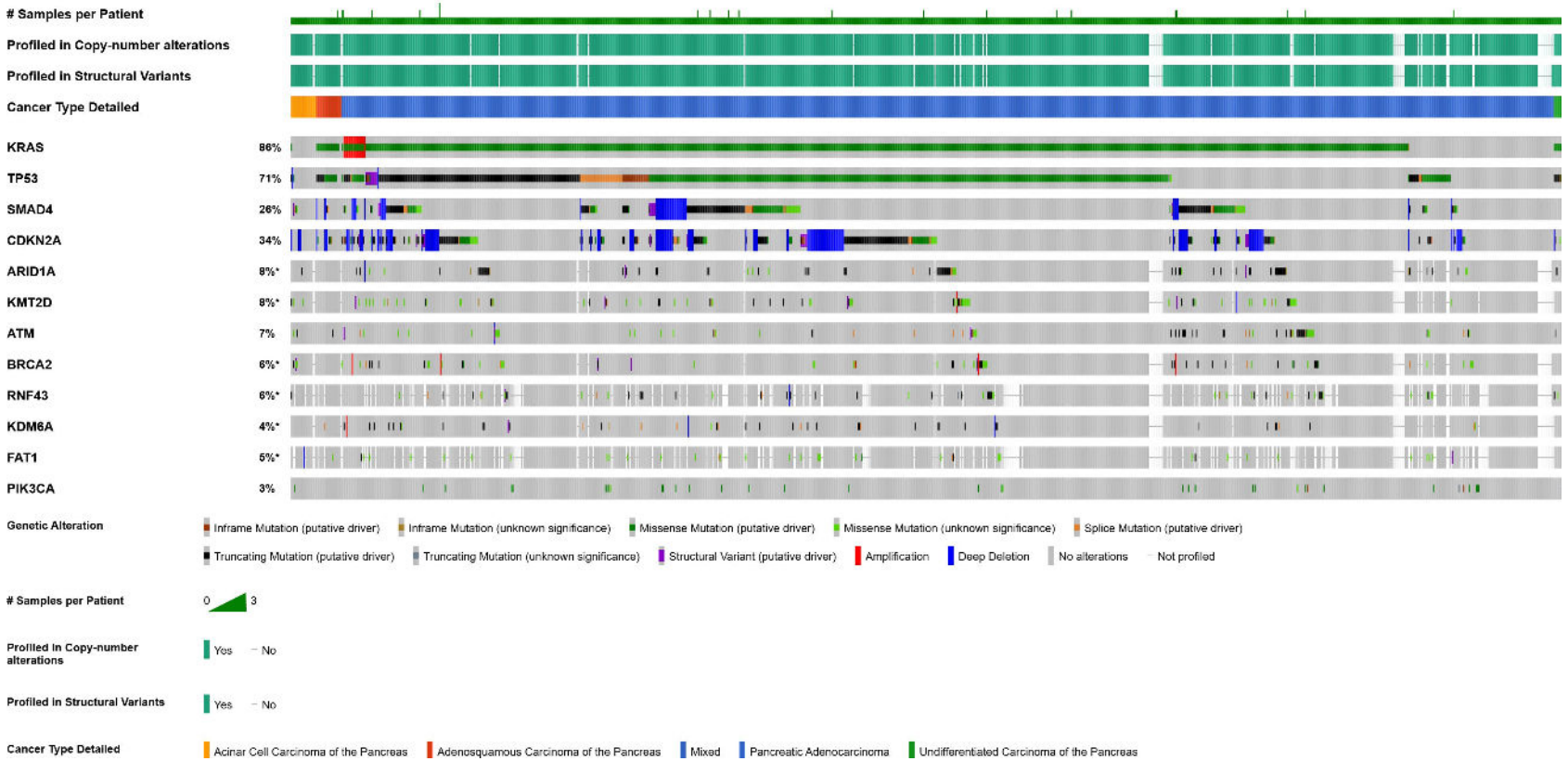
Gene	# SV	#	Freq
CDKN2A	16	16	1.5%
SMAD4	10	10	0.9%
TP53	10	10	0.9%
BRAF	8	8	0.8%
NTRK1	5	3	0.3%
BRCA2	4	4	0.4%
CDKN2B	4	4	0.4%
KMT2D	4	4	0.4%
SND1	4	4	NA
ALK	3	3	0.3%
ATM	3	3	0.3%
BRCA1	3	3	0.3%
CTRC	3	2	NA
EGFR	3	3	0.3%
ETV6	3	3	0.3%
KMT2A	3	3	0.3%
KMT2C	3	3	0.5%

Copy Number Alterations

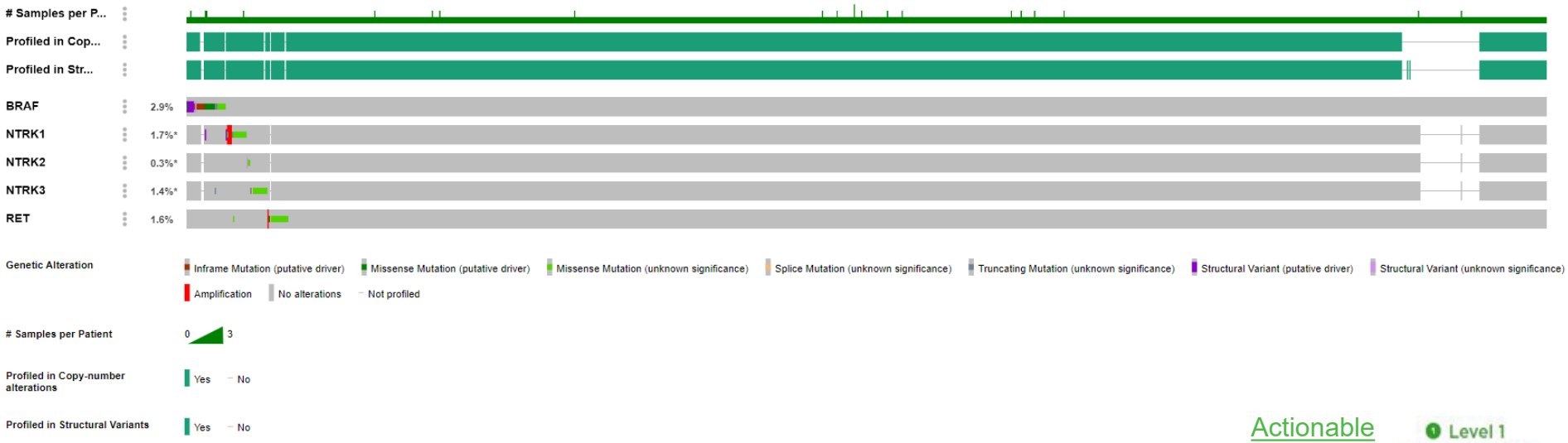
CNA Genes (1058 profiled samples)

Gene	Cytoband	CNA	#	Freq
RUNX1T1	8q21.3	AMP	1	0.3%
FANCM	14q21.2	AMP	1	0.3%
STK19	6p21.33	AMP	1	0.3%
INPPL1	11q13.4	AMP	1	0.3%
RRAGC	1p34.3	AMP	1	0.3%
PRDM14	8q13.3	AMP	1	0.3%
CYSLTR2	13q14.2	HOMDEL	1	0.3%
TAP2	6p21.32	AMP	1	0.3%
TAP1	6p21.32	AMP	1	0.3%
EPAS1	2p21	AMP	1	0.3%
SMYD3	1q44	AMP	1	0.3%
CYSLTR2	13q14.2	AMP	1	0.3%
DROSHA	5p13.3	AMP	1	0.3%
LYN	8q12.1	AMP	1	0.2%
TEK	9p21.2	AMP	1	0.2%
CRTC1	19p13.11	AMP	1	0.2%
STAT6	12q13.3	AMP	1	0.2%

BPC PANC: Top 12 Mutated Genes



BPC PANC: Clinically Actionable Genes



Actionable Genes

OncoKB

Level 1
FDA-approved drugs

Level R1
Standard care

Chakravarty et al., JCO PO 2017

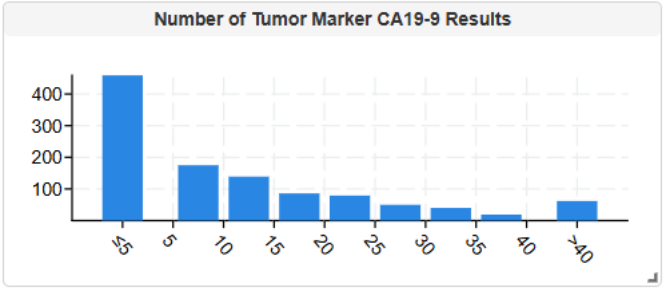
BPC PANC Cohort: Cancer Specific Fields

Tumor Resectability Status: captured at the time of clinical staging prior to any cancer-directed treatment

Tumor's Resectability Status

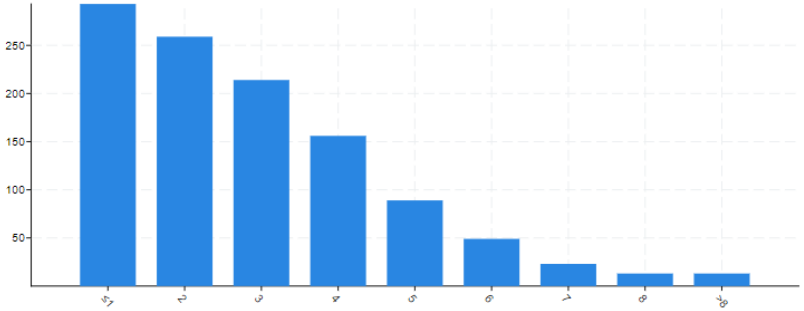
	#	Freq
Unresectable/locally advanced or …	585	52.8%
Resectable	378	34.1%
Borderline resectable	125	11.3%
Indeterminate	20	1.8%
NA	1	<0.1%

CA19-9: count of tumor marker results per patient



BPC PANC Cohort: Complete Treatment Histories

Number of cancer-directed drug regimens curated for each patient



Cancer-directed Drug Regimens (Includes regimens ever received for PANC irrespective of line or stage)

Regimen	# of patients
FULV (Fluorouracil & LeucoVorin)	777
Gemcitabine + Radiation Therapy	309
Fluorouracil + Radiation Therapy	290
GemCap (Gemcitabine & Capecitabine)	270

Treatments by Patient

Treatment	#
Gemcitabine HCL	850
Fluorouracil	800
Leucovorin	780
Oxaliplatin	752
Irinotecan HCL	675
Nabpaclitaxel	628
Capecitabine	298
Investigational Drug	200
Irinotecan liposome	110
Cisplatin	65
Olaparib	22
Pembrolizumab	15
Carboplatin	11
Nivolumab	9
Mitomycin	9
Docetaxel	9

Treatments by Sample (pre- and post-)

Treatment	Pre / Post	#
Gemcitabine HCL	Pre	752
Gemcitabine HCL	Post	119
Fluorouracil	Pre	653
Fluorouracil	Post	167
Leucovorin	Pre	633
Leucovorin	Post	166
Oxaliplatin	Pre	603
Oxaliplatin	Post	167
Nabpaclitaxel	Pre	571
Nabpaclitaxel	Post	70
Irinotecan HCL	Pre	533
Irinotecan HCL	Post	158
Capecitabine	Pre	268
Capecitabine	Post	42
Investigational Drug	Pre	189
Investigational Drug	Post	18

Longitudinal Data Collection

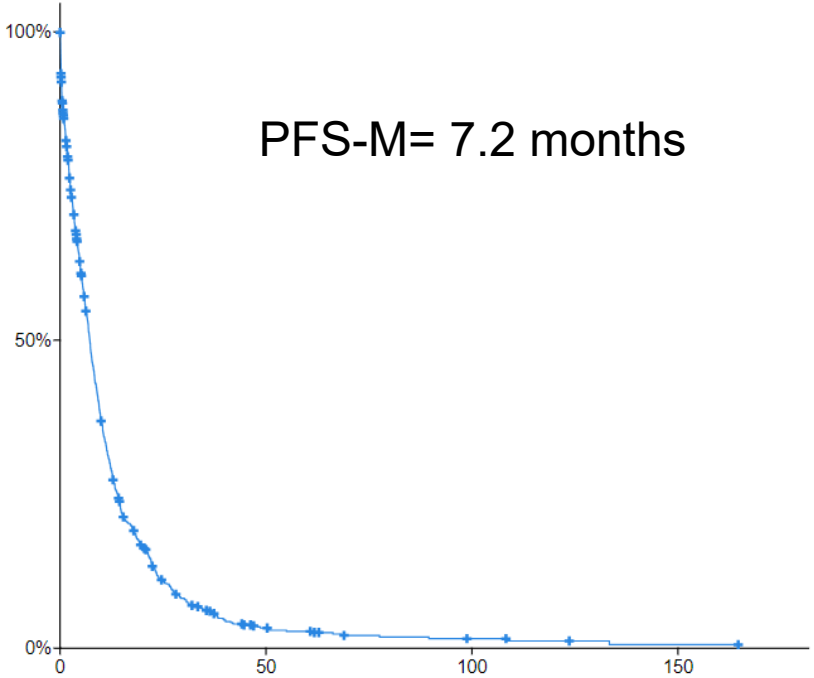
Sample Patient

Disease Status by Medical Oncology Note

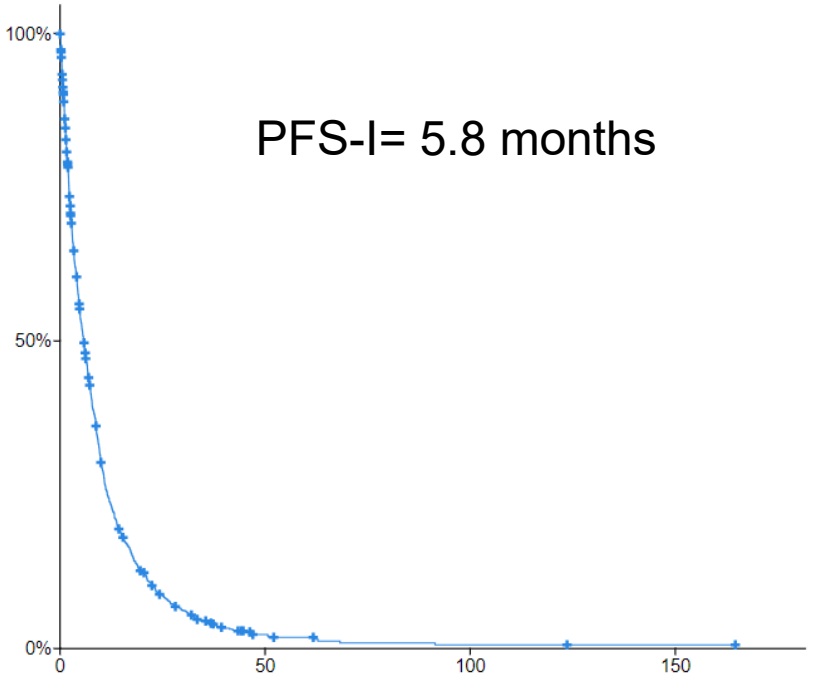
Disease Status by Imaging

Pathology Reports

BPC PANC Cohort: High Quality Clinical Data



PFS-M= 7.2 months

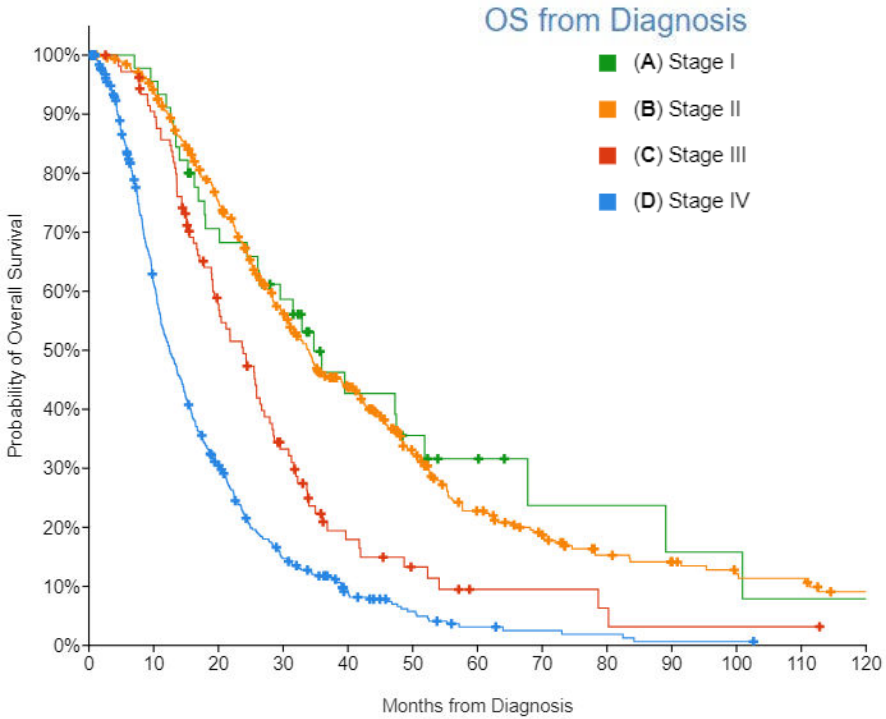


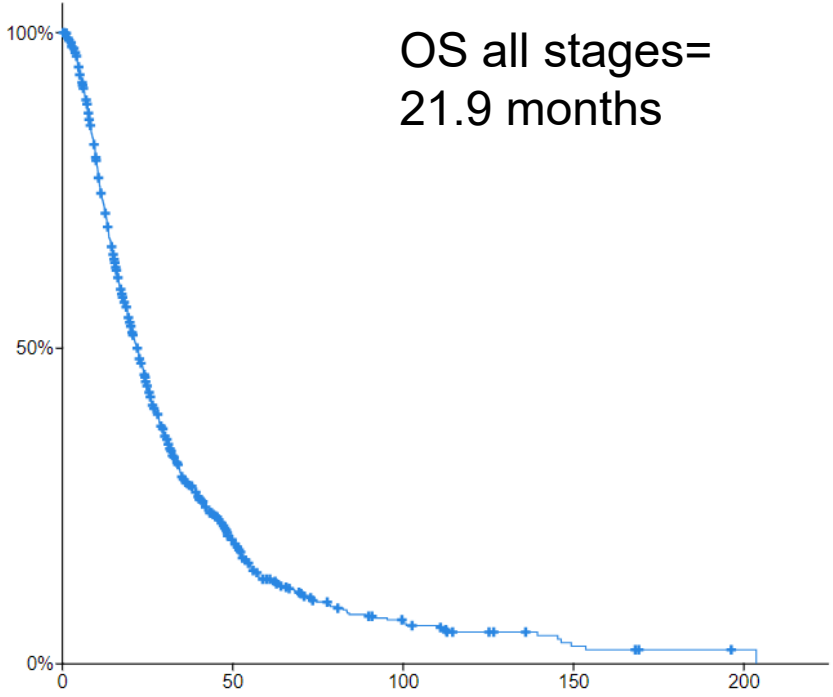
PFS-I= 5.8 months

- PFS-M and PSF-I are available by regimen for those regimens containing greater than 10 patients

* Not adjusted for delayed entry

BPC PANC Cohort: OS by Stage





OS all stages= 21.9 months

* Not adjusted for delayed entry

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