

2024-07-15

Ontario Institute for Cancer Research

Djerba: Sharing and Updating a Modular System for Clinical Report Generation

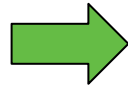
Iain Bancarz PhD, Felix Beaudry PhD, Aqsa Alam,
Oumaima Hamza, Alexander Fortuna, Trevor Pugh PhD FACMG



Clinical reporting for cancer



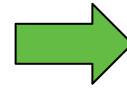
Genome
sequencing



47 clinical workflows



Variant
detection



Clinical
report



OICR
Ontario Institute for Cancer Research

Clinical Research Report
PATIENT & P1923040

CASE OVERVIEW

TREATMENT OPTIONS

RESULTS SUMMARY

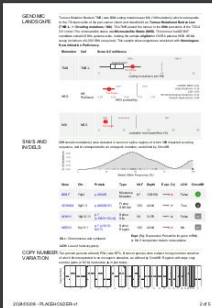
EMPIRIC INFORMATION

20180000-PLA0010200-01 1 of 5



Ontario Institute for Cancer Research
c/o Tissue Portal Sample Receiving MaRS Centre
West Tower 661 University Avenue, Suite 6-46
Toronto, Ontario, Canada, M5G 0A3
CAP: 8381376 ACDx: 0730 CLIA: 99D2270792

Clinical Research Report



CLINICAL AND TREATMENT INFORMATION

ABSTRACT

DISCUSSION

REFERENCES

APPENDIX

20180000-PLA0010200-01 1 of 5

APPENDIX

REFERENCES

DISCUSSION

20180000-PLA0010200-01 1 of 5

APPENDIX

REFERENCES

DISCUSSION

20180000-PLA0010200-01 1 of 5

GOCHR
Clinical Research Report
PATIENT & PHYSICIAN
CASE
TREATMENT
RESULTS & SUMMARY
SAMPLE INFORMATION

20240508-PLA300102001

Primary cancer: Ovarian
Site of biopsy/surgery: Paravertebral Mass
Study: PLACEHOLDER **Patient Study ID:** None
Patient LIMS ID: PLACEHOLDER **Tumour Sample ID:** PLACEHOLDER
Requisition Approved: 2024/02/02 **Blood Sample ID:** PLACEHOLDER
Date of Report: 2024/05/08 **Report ID:** PLACEHOLDER-v1

GENOMIC LANDSCAPE

20240508-PLA300102001

CLINICAL AND TREATMENT SUMMARY

20240508-PLA300102001

APPENDIX

20240508-PLA300102001

APPENDIX

20240508-PLA300102001

FDA Approved and/or NCCN Recommended Biomarker:

OncoKB	Treatment(s)	Gene(s)	Alteration
1	Dabrafenib+Trametinib	<i>BRAF</i>	p.V600E

ONCO

Clinical Research Report

PROTEIN & PIVOTAL

CASE

ONCOGENE

TREATMENT

OPTIONS

RESULTS

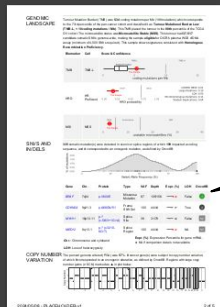
SUMMARY

EMPIRICAL

EVALUATION

2018-01-01 - 10:00

Protein	Type	VAF	Depth	Expr. (%)	LOH	OncoKB
p.V600E	Missense Mutation	67	105/156	—●98	False	1



CLINICAL

AND

RESEARCH

RESULTS

Figure 4: Clinical and research results. The plot shows the number of mutations per megabase (mut/Mb) across the genome. The x-axis represents the chromosome number (1-23, X, Y). The y-axis represents the number of mutations per megabase (mut/Mb). The plot shows a high number of mutations in the 1p and 11q regions, which are consistent with the presence of the p.V600E mutation.

APPENDIX

RESULTS

Figure 5: Appendix results. The plot shows the number of mutations per megabase (mut/Mb) across the genome. The x-axis represents the chromosome number (1-23, X, Y). The y-axis represents the number of mutations per megabase (mut/Mb). The plot shows a high number of mutations in the 1p and 11q regions, which are consistent with the presence of the p.V600E mutation.

2018-01-01 - 10:00

APPENDIX

RESULTS

Figure 6: Appendix results. The plot shows the number of mutations per megabase (mut/Mb) across the genome. The x-axis represents the chromosome number (1-23, X, Y). The y-axis represents the number of mutations per megabase (mut/Mb). The plot shows a high number of mutations in the 1p and 11q regions, which are consistent with the presence of the p.V600E mutation.

2018-01-01 - 10:00

APPENDIX

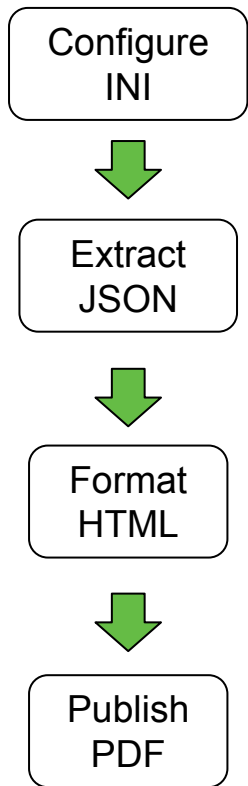
RESULTS

Figure 7: Appendix results. The plot shows the number of mutations per megabase (mut/Mb) across the genome. The x-axis represents the chromosome number (1-23, X, Y). The y-axis represents the number of mutations per megabase (mut/Mb). The plot shows a high number of mutations in the 1p and 11q regions, which are consistent with the presence of the p.V600E mutation.

2018-01-01 - 10:00

Level	Definition
1	FDA-recognized biomarker predictive of response

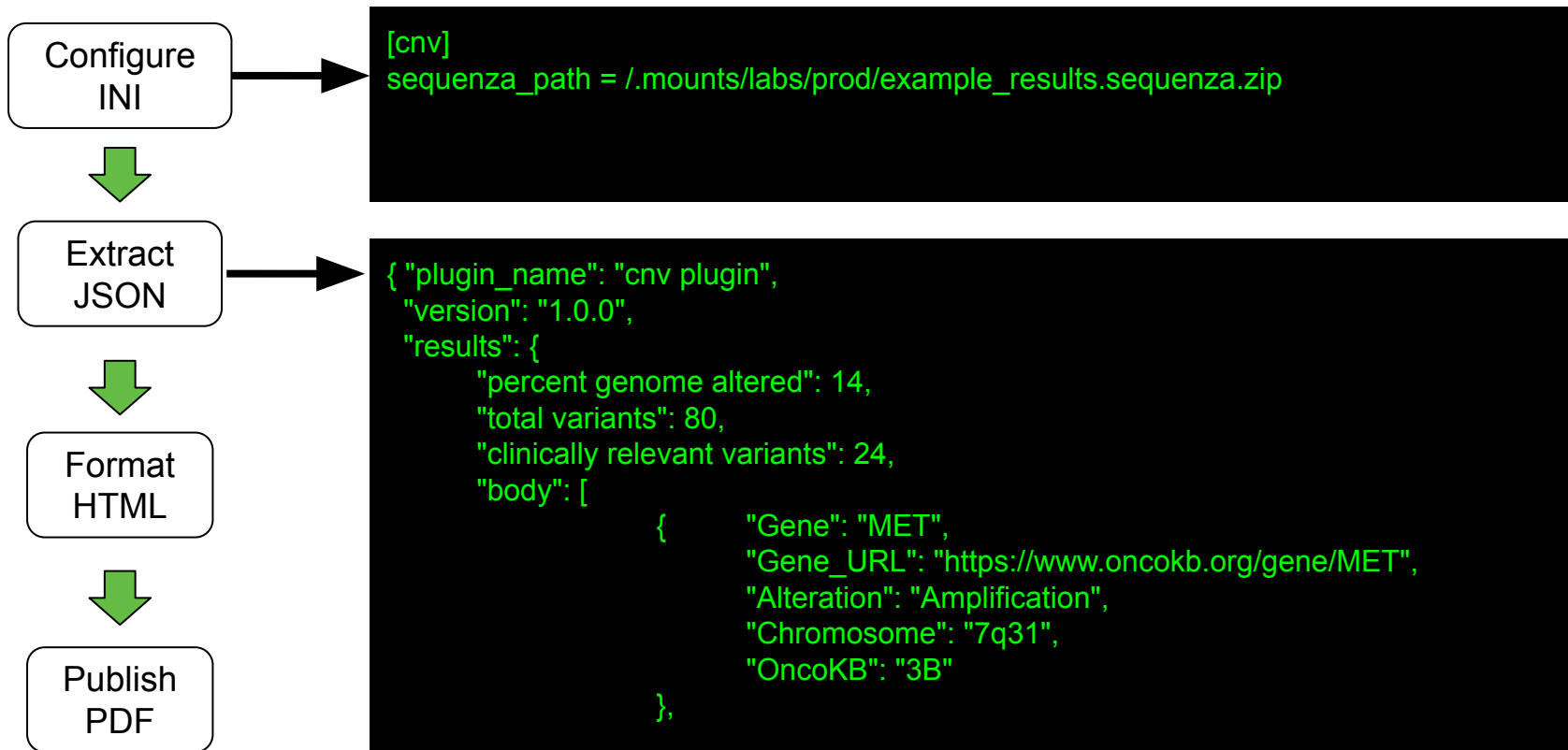
Djerba: Clinical reporting with plugins



Djerba: Clinical reporting with plugins



Djerba: Clinical reporting with plugins



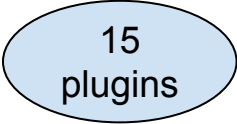
Djerba: Clinical reporting with a new plugin!



Djerba: The First Assay

Whole Genome & Transcriptome

core
report_title
patient_info
supplement.body
input_params_helper
provenance_helper
wgts.cnv_purple
case_overview
treatment_options_merger
summary
sample
wgts.snv_indel
fusion
expression_helper
gene_information_merger



15
plugins

Djerba: Reporting A New Assay

Whole Genome & Transcriptome

core
report_title
patient_info
supplement.body
input_params_helper
provenance_helper
wgts.cnv_purple
case_overview
treatment_options_merger
summary
sample
wgts.snv_indel
fusion
expression_helper
gene_information_merger

15
plugins

Plasma Whole Genome

core
report_title
patient_info
supplement.body
pwgs_provenance_helper
pwgs_cardea_helper
pwgs.case_overview
pwgs.summary
pwgs.sample
pwgs.analysis

10
plugins

NEW!

Shared
plugins for
faster
development!

Whole Genome & Transcriptome



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Director: Trevor Pugh, PhD, FACMG
Phone: 647-468-7844
Main contact: Alexander Fortuna, MSc
Phone: 416-673-8539
Hours of Operation: Mon-Fri, 9:00AM - 5:00PM

Clinical Research Report

PATIENT & PHYSICIAN

Patient Name: LAST, FIRST Patient DOB: yyyy/mm/dd
Patient Genetic Sex: SEX Requisitioner Email: NAME@domain.com
Physician Licence #: nnnnnnnnn Physician: LAST, FIRST
Physician Phone #: nnn-nnn-nnnn Physician Hospital: HOSPITAL NAME AND ADDRESS

CASE OVERVIEW

Assay: Whole genome and transcriptome sequencing (WGTS)-80X Tumour, 30X Normal (v3.0)
Primary cancer: Ovarian
Site of biopsy/surgery: Paravertebral Mass
Study: PLACEHOLDER Patient Study ID: None
Patient LIMS ID: PLACEHOLDER Tumour Sample ID: PLACEHOLDER
Requisition Approved: 2024/02/02 Blood Sample ID: PLACEHOLDER
Date of Report: 2024/05/08 Report ID: PLACEHOLDER-v1

TREATMENT OPTIONS

Review identified 1 option(s) indicating an FDA Approved and/or NCCN Recommended Biomarker 2 option(s) indicating investigational therapies, and 0 option(s) indicating NCCN-listed biomarkers.

FDA Approved and/or NCCN Recommended Biomarker:

OncoKB	Treatment(s)	Gene(s)	Alteration
1	Dabrafenib+Trametinib	BRAF	p.V600E

Investigational Therapies:

OncoKB	Treatment(s)	Gene(s)	Alteration
3B	Dabrafenib, Encorafenib+Cetuximab, Selumetinib, Tovorafenib, Vemurafenib+Atezolizumab+Cobimetinib, Encorafenib+Panitumumab, Trametinib, Vemurafenib, Vemurafenib+Cobimetinib,	BRAF	p.V600E



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PATIENT & PHYSICIAN

Patient Name: LAST, FIRST Patient DOB: yyyy/mm/dd
Patient Genetic Sex: SEX Requisitioner Email: NAME@domain.com
Physician Licence #: nnnnnnnnn Physician: LAST, FIRST
Physician Phone #: nnn-nnn-nnnn Physician Hospital: HOSPITAL NAME AND ADDRESS

CASE OVERVIEW

Assay: plasma Whole Genome Sequencing (pWGS) - 30X (v1.0)
Primary cancer: Pancreatic Adenocarcinoma
Site of biopsy: Blood cell-free DNA
Study: PLACEHOLDER Patient Study ID: PLACEHOLDER
Patient LIMS ID: PLACEHOLDER WGS Report ID: PLACEHOLDER
Requisition Approved: 2024/05/28 Plasma Sample ID: PLACEHOLDER
Date of Report: yyyy/mm/dd pWGS Report ID: PLACEHOLDER

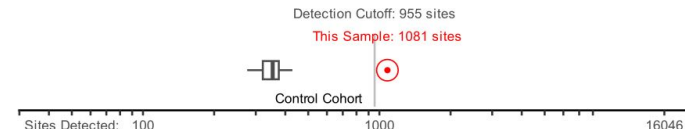
SUMMARY

Cell-free DNA tumour burden: Detected
Mutational sampling analysis: ctDNA DETECTED

SAMPLE QC

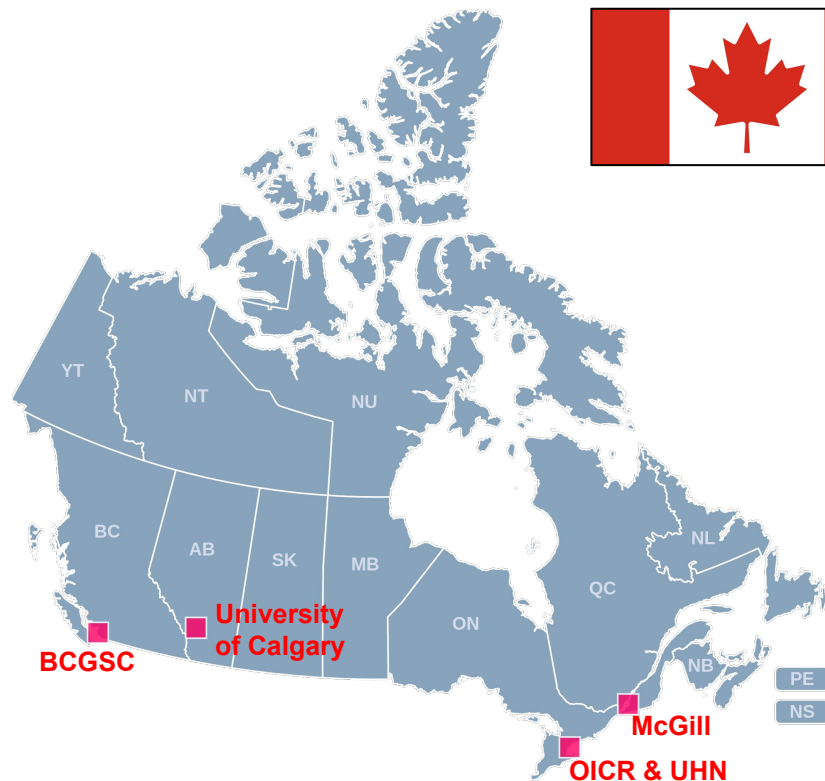
Type of Specimen: Plasma Candidate SNVs: 16,492
Median Insert Size (bp): 165 Coverage (mean): 31.0

MUTATIONAL SAMPLING



Sharing Djerba across Canada

- MOHCCN-O working group:
 - OICR (Ontario)
 - UHN Toronto
 - McGill (Montreal)
 - University of Calgary (Alberta)
 - BCGSC (British Columbia)
 - Broad (USA, observer)
- Can develop plugins independently
- Proof-of-concept done!
- TODO Custom reports for production



Djerba: A modular system for clinical report generation

- Open source; GPL 3.0 license
- **Main:** <https://github.com/oicr-gsi/djerba>
- **Demo:** <https://github.com/oicr-gsi/djerba-demo>





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is provided by the Government of Ontario

