

# Personalized multi-omics diagnostics of colon cancer

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**Abstract—ABSTRAAAAAAAAACT**

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## I. INTRODUCTION

Worldwide, colorectal cancer is the third most diagnosed cancer. An estimated 1,880,725 people were diagnosed with colorectal cancer in 2020. Two important features of CRC are high inter-patient variability and high spatial heterogeneity, which may influence the molecular characterization of tumor tissue. Hence, molecular subtyping of CRC is a yet not solved problem, the scientific resolution of which is essential for opportunities of precision medicine for colorectal cancer patients. Subtyping of CRC has been done using various methods previously.

## II. DATA AND PREPROCESSING

### A. Dataset

The dataset was obtained from the public database cBioPortal, from the study MSK-MET 2021 (Memorial Sloan Kettering - Metastatic Events and Tropisms 2021) study [cite 1]. It contains tumor genomic and clinical outcome data from a pan-cancer cohort of over 25,000 patients with metastatic diseases, including information about 50 different primary and metastatic cancer types. This paper is going to focus on colon and rectal cancer, which includes 3093 patients from the dataset, from which 2073 have primary and 1020 metastatic cancers (Fig 1). For model building primary cancer data is used.

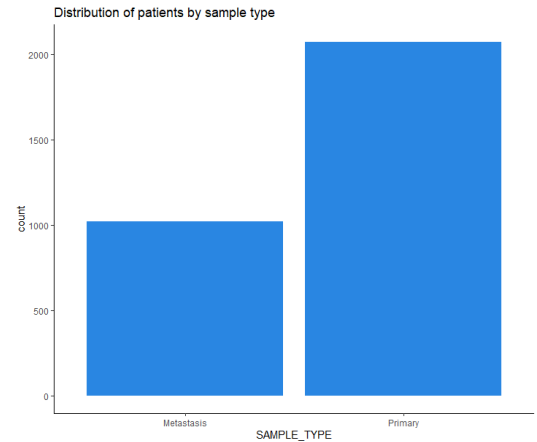


Fig. 1. Primary vs Metastatic cancer patients count

Exploratory data analysis showed that our data is very sparse (Fig2,3). This means there are many patients with very few number of mutations, which is typical for cancer mutation profiles.

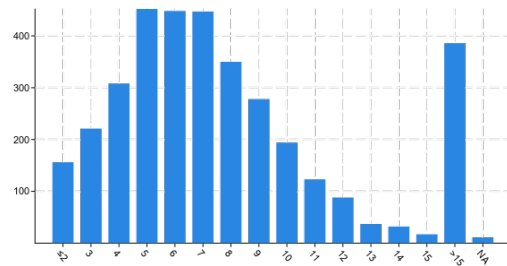


Fig. 2. Distribution of patients based on mutation count. Most patients have 5-8 mutations

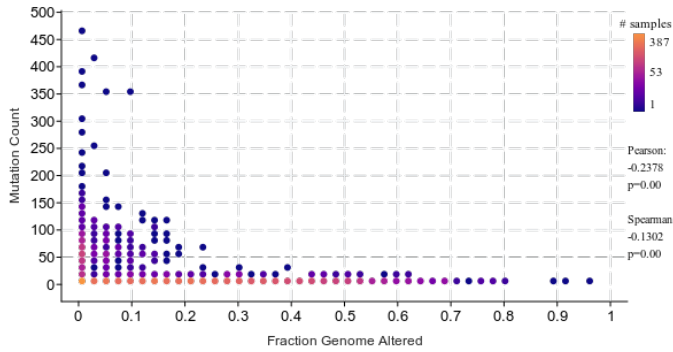


Fig. 3. Fraction of Genome Altered is the percentage of genome that has been affected by copy number gains or losses. Total Mutations refers to the number of mutations that are found in the tumor genomes

After obtaining relevant genomic (including mutations and copy number variations (cnv) and clinical data, preprocessing of the data was performed to

- remove non relevant for the task types of mutations, such as RNA, silent, splice site mutations
- remove cnv's that cause loss of heterozygosity
- map gene names to universal HGNC nomenclature
- transform dataframes to one-hot-encoding for each patient-gene pair

As the model was going to perform clustering, then classification on a single dataframe, we needed to combine the mutational and cnv data with some biological logic behind. Hence, the data were combined by counting cnv data as mutations in case

- cnv is a deletion in a tumor supressor gene
- cnv is an amplification of an oncogene

EDA of the patient clinical information showed general patterns of our patient sample. In terms of gender, our dataset is balanced (Fig.4), white race group is overrepresented (Fig.5), age is normally distributed within the range ;30 - 90 y.o. (Fig.6).

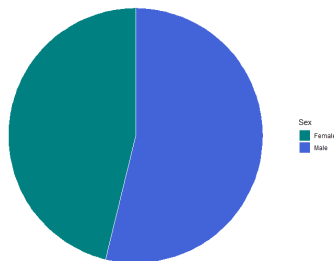


Fig. 4. Distribution of patients by sex

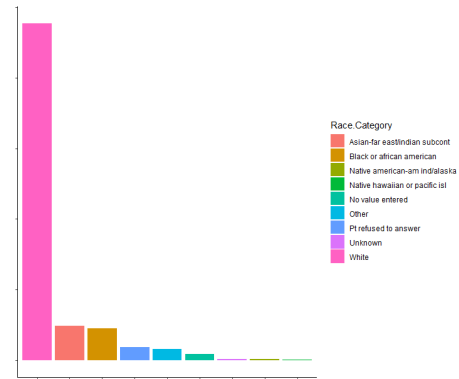


Fig. 5. Distribution of patients by race

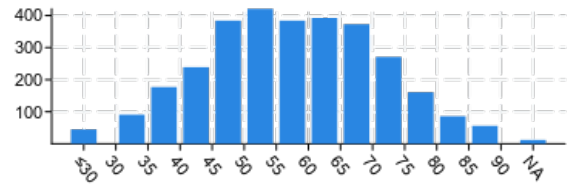


Fig. 6. Age of patients at the time of genetic sequencing of tumors (data collection)

### III. PREPARE YOUR PAPER BEFORE STYLING

Before you begin to format your paper, first write and save the content as a separate text file. Complete all content and organizational editing before formatting. Please note sections III-A–III-E below for more information on proofreading, spelling and grammar.

Keep your text and graphic files separate until after the text has been formatted and styled. Do not number text heads— $\LaTeX$  will do that for you.

#### A. Abbreviations and Acronyms

Define abbreviations and acronyms the first time they are used in the text, even after they have been defined in the abstract. Abbreviations such as IEEE, SI, MKS, CGS, ac, dc, and rms do not have to be defined. Do not use abbreviations in the title or heads unless they are unavoidable.

#### B. Units

- Use either SI (MKS) or CGS as primary units. (SI units are encouraged.) English units may be used as secondary units (in parentheses). An exception would be the use of English units as identifiers in trade, such as “3.5-inch disk drive”.
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- Do not mix complete spellings and abbreviations of units: “Wb/m<sup>2</sup>” or “webers per square meter”, not “webers/m<sup>2</sup>”.

Spell out units when they appear in text: “. . . a few henries”, not “. . . a few H”.

- Use a zero before decimal points: “0.25”, not “.25”. Use “cm<sup>3</sup>”, not “cc”).

### C. Equations

Number equations consecutively. To make your equations more compact, you may use the solidus ( / ), the exp function, or appropriate exponents. Italicize Roman symbols for quantities and variables, but not Greek symbols. Use a long dash rather than a hyphen for a minus sign. Punctuate equations with commas or periods when they are part of a sentence, as in:

$$a + b = \gamma \quad (1)$$

Be sure that the symbols in your equation have been defined before or immediately following the equation. Use “(1)”, not “Eq. (1)” or “equation (1)”, except at the beginning of a sentence: “Equation (1) is . . .”

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Please use “soft” (e.g., `\eqref{Eq}`) cross references instead of “hard” references (e.g., (1)). That will make it possible to combine sections, add equations, or change the order of figures or citations without having to go through the file line by line.

Please don’t use the `{eqnarray}` equation environment. Use `{align}` or `{IEEEeqnarray}` instead. The `{eqnarray}` environment leaves unsightly spaces around relation symbols.

Please note that the `{subequations}` environment in  $\LaTeX$  will increment the main equation counter even when there are no equation numbers displayed. If you forget that, you might write an article in which the equation numbers skip from (17) to (20), causing the copy editors to wonder if you’ve discovered a new method of counting.

$\BIBTeX$  does not work by magic. It doesn’t get the bibliographic data from thin air but from .bib files. If you use  $\BIBTeX$  to produce a bibliography you must send the .bib files.

$\LaTeX$  can’t read your mind. If you assign the same label to a subsection and a table, you might find that Table I has been cross referenced as Table IV-B3.

$\LaTeX$  does not have precognitive abilities. If you put a `\label` command before the command that updates the counter it’s supposed to be using, the label will pick up the last counter to be cross referenced instead. In particular, a `\label` command should not go before the caption of a figure or a table.

Do not use `\nonumber` inside the `{array}` environment. It will not stop equation numbers inside `{array}` (there won’t be any anyway) and it might stop a wanted equation number in the surrounding equation.

### E. Some Common Mistakes

- The word “data” is plural, not singular.

- The subscript for the permeability of vacuum  $\mu_0$ , and other common scientific constants, is zero with subscript formatting, not a lowercase letter “o”.
- In American English, commas, semicolons, periods, question and exclamation marks are located within quotation marks only when a complete thought or name is cited, such as a title or full quotation. When quotation marks are used, instead of a bold or italic typeface, to highlight a word or phrase, punctuation should appear outside of the quotation marks. A parenthetical phrase or statement at the end of a sentence is punctuated outside of the closing parenthesis (like this). (A parenthetical sentence is punctuated within the parentheses.)
- A graph within a graph is an “inset”, not an “insert”. The word alternatively is preferred to the word “alternately” (unless you really mean something that alternates).
- Do not use the word “essentially” to mean “approximately” or “effectively”.
- In your paper title, if the words “that uses” can accurately replace the word “using”, capitalize the “u”; if not, keep using lower-cased.
- Be aware of the different meanings of the homophones “affect” and “effect”, “complement” and “compliment”, “discreet” and “discrete”, “principal” and “principle”.
- Do not confuse “imply” and “infer”.
- The prefix “non” is not a word; it should be joined to the word it modifies, usually without a hyphen.
- There is no period after the “et” in the Latin abbreviation “et al.”.
- The abbreviation “i.e.” means “that is”, and the abbreviation “e.g.” means “for example”.

An excellent style manual for science writers is [7].

### F. Authors and Affiliations

**The class file is designed for, but not limited to, six authors.** A minimum of one author is required for all conference articles. Author names should be listed starting from left to right and then moving down to the next line. This is the author sequence that will be used in future citations and by indexing services. Names should not be listed in columns nor group by affiliation. Please keep your affiliations as succinct as possible (for example, do not differentiate among departments of the same organization).

### G. Identify the Headings

Headings, or heads, are organizational devices that guide the reader through your paper. There are two types: component heads and text heads.

Component heads identify the different components of your paper and are not topically subordinate to each other. Examples include Acknowledgments and References and, for these, the correct style to use is “Heading 5”. Use “figure caption” for your Figure captions, and “table head” for your table title. Run-in heads, such as “Abstract”, will require you to apply a style (in this case, italic) in addition to the style

provided by the drop down menu to differentiate the head from the text.

Text heads organize the topics on a relational, hierarchical basis. For example, the paper title is the primary text head because all subsequent material relates and elaborates on this one topic. If there are two or more sub-topics, the next level head (uppercase Roman numerals) should be used and, conversely, if there are not at least two sub-topics, then no subheads should be introduced.

## H. Figures and Tables

a) *Positioning Figures and Tables:* Place figures and tables at the top and bottom of columns. Avoid placing them in the middle of columns. Large figures and tables may span across both columns. Figure captions should be below the figures; table heads should appear above the tables. Insert figures and tables after they are cited in the text. Use the abbreviation “Fig. 6”, even at the beginning of a sentence.

TABLE I  
TABLE TYPE STYLES

Table Head	Table Column Head		
	Table column subhead	Subhead	Subhead
copy	More table copy <sup>a</sup>		

<sup>a</sup>Sample of a Table footnote.

Figure Labels: Use 8 point Times New Roman for Figure labels. Use words rather than symbols or abbreviations when writing Figure axis labels to avoid confusing the reader. As an example, write the quantity “Magnetization”, or “Magnetization, M”, not just “M”. If including units in the label, present them within parentheses. Do not label axes only with units. In the example, write “Magnetization (A/m)” or “Magnetization {A[m(1)]}”, not just “A/m”. Do not label axes with a ratio of quantities and units. For example, write “Temperature (K)”, not “Temperature/K”.

## ACKNOWLEDGMENT

The preferred spelling of the word “acknowledgment” in America is without an “e” after the “g”. Avoid the stilted expression “one of us (R. B. G.) thanks ...”. Instead, try “R. B. G. thanks...”. Put sponsor acknowledgments in the unnumbered footnote on the first page.

## REFERENCES

Please number citations consecutively within brackets [1]. The sentence punctuation follows the bracket [2]. Refer simply to the reference number, as in [3]—do not use “Ref. [3]” or “reference [3]” except at the beginning of a sentence: “Reference [3] was the first ...”

Number footnotes separately in superscripts. Place the actual footnote at the bottom of the column in which it was cited. Do not put footnotes in the abstract or reference list. Use letters for table footnotes.

Unless there are six authors or more give all authors’ names; do not use “et al.”. Papers that have not been published,

even if they have been submitted for publication, should be cited as “unpublished” [4]. Papers that have been accepted for publication should be cited as “in press” [5]. Capitalize only the first word in a paper title, except for proper nouns and element symbols.

For papers published in translation journals, please give the English citation first, followed by the original foreign-language citation [6].

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