To: UPHS Physicians and Staff

From: The Division of Precision and Computational Diagnostics (PCD)

Vivianna Van Deerlin, MD, PhD

Interim Director of PCD and Director of Molecular Pathology Laboratory

Jennifer Morrissette, PhD

Interim Director of the Center for Personalized Diagnostics

Salvatore Priore, MD, PhD

Assistant Professor of Clinical Pathology and Laboratory Medicine

Date: November 16, 2023

Re: Update to PennSeq™ Report Format

The Center for Personalized Diagnostics (CPD) performs testing designed to detect genomic variants in oncology samples. The CPD is pleased to announce that we will implement format changes to our PennSeq[™] clinical reports to improve readability of genomic results. This change is an initial step in a long-term plan to improve CPD reports. With these new edits, there will be no loss of previously provided critical information. Clinicians can expect the following changes in reports:

1. Variant Descriptions:

- 2. Variants will be listed with relevant information combined at the top of the report;
 - a. Interpretative information will appear with each variant (e.g., Variant Allele Frequency (VAF)).
 - Descriptive text to specifically identify important and actionable variants will appear next to the gene name (e.g. EGFR Exon 19 Deletion).
 - c. Alternate p. and c. nomenclature when necessary to help with interpretation (e.g. BRCA1 variants).

3. Improved "Indeterminate" Variant Reporting:

- Separate "Indeterminate" section on the report will be removed to avoid previous confusion regarding these variants.
- b. These variants will now be listed under their appropriate disease category group (e.g. Disease-Associated, VUS, etc.) with a comment in the VAF field, "Indeterminate, See Comment" to flag these variants as being borderline calls

4. Updates to the Interpretation Section:

- a. Excessive descriptive text will be removed.
- Relevant information will be captured in the new variant descriptions as described in #1.
- This section will now contain general interpretive text relevant to the variants seen in the case.

5. Introduction of New Report Sections:

a. Patient past and concurrent testing at CPD and relevant pertinent negative variant review will now populate in dedicated sections of the report. This replaces text that appeared in the "Comments" section of the report.

Example
DISEASE ASSOCIATED VARIANTS [3 Total]:
EGFR, Potential Copy Number Gain (INDETERMINATE)
EGFR Exon 19 Deletion p.E746_A750del; c.2235_2249del; NM_005228.3 VAF: 39%
TP53 p.R213*; c.637C>T; NM_000546.5 VAF: Indeterminate, See Comment

This new report format will go into effect beginning Wednesday, Mar. 1, 2023. You may observe a mix of old and new reports for your patients during the transition period.

For additional information and questions, please call the CPD at 215-615-3966 on weekdays during regular business hours or visit

 $\frac{https://www.pennmedicine.org/departments-and-centers/center-for-personalized-diagnostics.}{}$

unsubscribe