



---

To: UPHS Physicians and Staff

From: **The Division of Precision and Computational Diagnostics (PCD)**  
**Salvatore F. Priore, M.D., Ph.D., Project Lead**  
**Jennifer Morrisette, Ph.D., Interim Director, Center for Personalized Diagnostics**  
**Vivianna Van Deerlin, M.D., Ph.D., Interim Director, PCD**

Date: May 6, 2024



Re: **Improvements and Changes to Cancer Genomic Reports and Variant Data Availability in PennChart**

---

The Center for Personalized Diagnostics, the genomic oncology laboratory in the Division of PCD, in partnership with SECTRA, are pleased to announce the launch of a new laboratory information system (LIS) that will manage all our Next-generation Sequencing (NGS) laboratory data. For providers, **this change means improved and easier to read reports and enhanced accessibility to discrete variant data in PennChart.**

**The new functionality described below begins May 2024.** During the transition period you will notice a mixture of new and old report formats. Functionality changes include:

1. New Report Format (sample data example below)



Final report, 4/25/2024 9:58 AM  
Page 1 of 6  
**Amendment**

**Update**

PATIENT INFORMATION		SAMPLE	
NAME Moore, Addison		SPECIMEN ID SM 24 0000, A1	SPECIMEN TYPE FFPE Slides
MRN 730023-3337 (Unspecified)	DATE OF BIRTH 3/23/1973	COLLECTION SOURCE & DATE Lung, right upper lobe, 4/16/2024	TUMOR IDENTITY u-40

**Variants with Clinical Significance for FDA-Approved Compounds (Tier 1)**

GENE	TYPE	DETAILS
EGFR	EGFR Exon 19 Deletion	p.E746_A750del, c.2235_2249del, NM_005228.5 VAF: 10%

**Other Variants with Strong or Potential Clinical Significance (Tier 2)**

GENE	TYPE	DETAILS
TP53	Missense variant	p.Arg273Cys, c.817C>T, NM_000546.5 VAF: 43%

**Variants of Unknown Clinical Significance (Tier 3)**

GENE	TYPE	DETAILS
BRAF	Missense variant	p.H58E, c.1753C>T, NM_004333.6 VAF: 50%

**Interpretation**

Tier 1 variants have an associated FDA-Approved compound and/or confer resistance to an FDA-Approved compound. Available FDA-Approved compounds are not necessarily indicated for all patients, tumor types, and/or clinical situations (please see <https://www.fda.gov/> for more information). Consult with a healthcare provider with expertise in clinical oncology care for your specific treatment options.

Tier 2 variants have clear or reasonable evidence of clinical significance in driving cancer development and progression. These variants may provide relevant prognostic and/or diagnostic information, but have no associated FDA-Approved compounds.

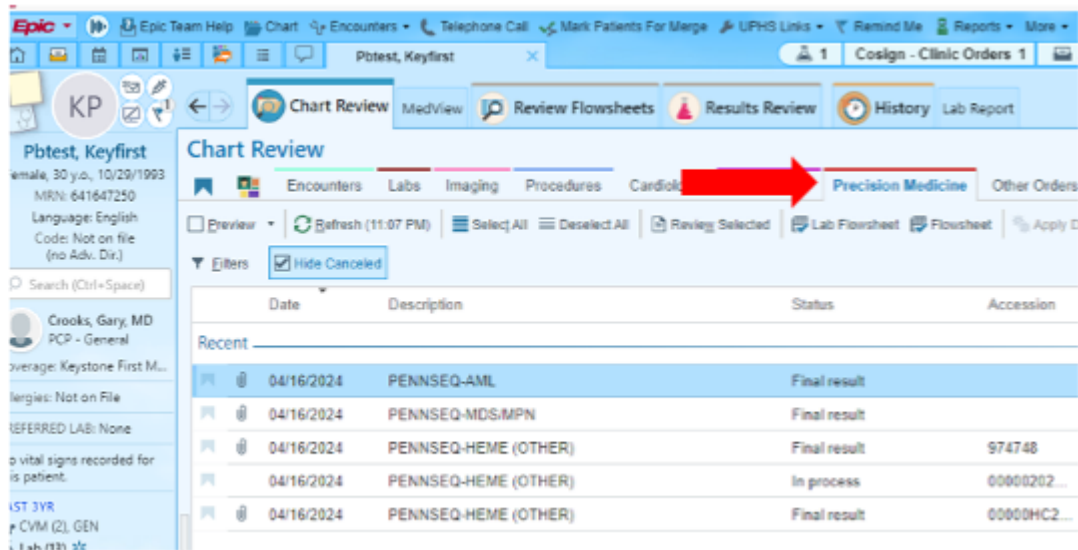
Tier 3 variants are variants of unknown clinical significance (VUS) and have little or no data to support a more specific classification. VUS have no clear or probable evidence that they drive the development and progression of cancer or provide diagnostic, prognostic, or therapeutic information. Interpret VUS with caution. Additionally, a VUS may represent a rare benign/likely benign genomic polymorphism.

**Report prepared by**  
Salvatore Priore  
The Center for Personalized Diagnostics

- New reports are .pdf format and contain the same basic content as current CPD reports.
- Variants are organized in an easier to read table layout and variant classification is changing to better conform with industry standards:
  - Variants previously described as “Disease-Associated” or “Probably Disease-Associated” will split into two categories:
    - “Variants with Clinical Significance for FDA-Approved Compounds (Tier 1)” for variants with associated FDA-Approved compounds or variants that confer resistance to FDA-Approved compounds.
    - “Other Variants with Strong or Potential Clinical Significance (Tier 2)” for all other disease-associated and probably disease-associated variants detected.
  - Variants of Uncertain Significance will appear under the category “Variants of Unknown Clinical Significance (Tier 3)”.
- Ongoing additional report enhancements are expected after the initial LIS launch.

## 2. Discrete Data in PennChart

- Variants in the .pdf report will have discrete entries in the PennChart Precision Medicine tab which allows for easier access to CPD variant data and paves the way for building clinical decision support tools to help providers identify actionable variants in their patient’s tumor samples.
- Discrete data is accessed by clicking on the relevant test result in the Precision Medicine tab and scrolling down to view all variant entries.



- Discrete data are organized into two parts:
  - A component header (red box below) that contains descriptive information about the tested specimen.
  - Genomic variant results (blue box below) which are a list of variant(s) identified in the sequencing study.

Component

Case Number: #2249488

Specimen Type: Blood

Collection Date: 2024/04/16

Collection Source:

Specimen ID:

Disclaimer: These variants represent results from a cancer genomic sequencing study (i.e. tumor only testing). Tumor only testing cannot distinguish germline variants from somatic variants. Additional germline testing, in consultation with an expert in genetic medicine, is needed to confirm any suspicion for an inherited cancer predisposition variant. Please refer to the final written report for additional interpretive text and information.

Resulting Agency: CENTER FOR PERSONALIZED DIAGNOSTICS

Genomic Variant Results Expand All Collapse All

Somatic

Ⓢ IDH1 (Tier 1 or 2: Strong or potential significance) IDH1, p.R132H VAF: 50%

Type: Somatic Variant Allele Freq.: 50.00%

Sequence Change & Region

DNA Change: c.205G>A Transcript: NM\_005895.4 AA Change: p.R132H Position: 208248288

Genome Assembly: hg38 Chromosome: 2

Specimen Collected: 04/16/24 1028 Last Resulted: 05/02/24 23:01

- Any variant classified as either Tier 1 or Tier 2 in the report will carry a red label of “Tier 1 or 2: Strong or potential significance”. Other information includes, the

DNA and amino acid changes (using HGVS nomenclature), reference transcript, chromosome, genomic position, and the reference genome assembly.

- Always refer to the final and most up-to-date PDF report for the most complete information for your patient.
- **Discrete variant data availability is prospective.** Cases prior to LIS launch will not contain discrete data entries.

### 3. Access to CPD Reports after LIS

- **LaunchReports created in the new LIS will only file in PennChart.** We recommend using the Precision Medicine tab to access CPD reports.
- **Medview and CERNER will refer the user to PennChart to access reports.** We recommend discontinuing use of these systems for CPD reports.

**Contact Information:** Please contact the project lead listed above with any questions or concerns related to this letter. For general information, call the CPD (215-615-3966) weekdays during regular business hours (8:30am – 5:00 pm) or consult our website (<https://pathology.med.upenn.edu/clinical-services/precision-and-computational-diagnostics/center-personalized-diagnostics>)

[unsubscribe](#)