Bulley, Margaret

From: labclientservice
Sent: Friday, December 7, 2018 11:44 AM
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Subject: RE: PENN MEDICINE - Copy Number Variants (CNVs) in the Solid Tumor Sequencing Panel: Center for Personalized Diagnostics (CPD)

From: OMAProviderEmailDistribution
Sent: Friday, December 07, 2018 11:32 AM
Subject: PENN MEDICINE - Copy Number Variants (CNVs) in the Solid Tumor Sequencing Panel: Center for Personalized Diagnostics (CPD)

Penn Medicine
University of Pennsylvania Health System

To: UPHS Physicians and Staff
From: The Division of Precision and Computational Diagnostics (PCD)
Kojo Elenitoba-Johnson, M.D., Director, Center for Personalized Diagnostics
Jennifer Morrissette, Ph.D., Clinical Director, Center for Personalized Diagnostics
Jason Rosenbaum, M.D., Assistant Professor of Clinical Pathology and Laboratory Medicine
Jacquelyn Roth, Ph.D., Assistant Professor of Clinical Pathology and Laboratory Medicine
Date: December 7, 2018
Re: Copy Number Variants (CNVs) in the Solid Tumor Sequencing Panel: Center for Personalized Diagnostics (CPD)

The Center for Personalized Diagnostics (CPD) offers assays designed to detect genomic variants in oncology samples. The CPD is pleased to announce the validation and implementation of a new bioinformatics algorithm for the detection of selected Copy Number Variants (CNVs) on the **Solid Tumor Sequencing Panel**. The new algorithm will be active on all cases processed starting **Monday, December 10, 2018**. The new algorithm offers improved sensitivity over previous versions. To distinguish from prior bioinformatics pipelines, the new version will be designated in the Methodology section of reports as "Halo v1.3."

Copy number gains in the following genes will be reported:

AKT1	*CREBBP*	*GNA11*	*KIT*	*PIK3CA*
AKT3	*CTNNB1*	*GNAQ*	*KRAS*	*RET*
ALK	*DDR2*	*HRAS*	*MAP2K1*	
BRAF	*EGFR*	*IDH1*	*MET*	
BRCA1	*ERBB2*	*IDH2*	*NRAS*	
CHEK2	*ESR1*	*KDR*	*PDGFRA*	

Copy number losses will not be detected by this assay. Due to the nature of the assay, low-level copy number gains (i.e. polysomy) in many cells cannot be distinguished from high level copy number gain (i.e. amplification) in few cells. For this reason, please note that **CNVs will not be quantified** and **gains in copy number are not necessarily equivalent to gene amplification**. If clinical decision-making depends on the detection of true gene amplification, orthogonal testing is recommended using a methodology that can discriminate individual cells (e.g. fluorescence in situ hybridization [FISH]). Should such testing be clinically warranted, please contact the laboratory for assistance in test selection.

There are no changes in the ability of the assay to identify single-nucleotide variants (SNVs) and small insertions and deletions (indels). There are no changes to the sample requirements, accepted sample types, or ordering procedures for the assay.

Results previously issued from the Solid Tumor Sequencing Panel, v2 can be re-analyzed using the new algorithm by request. To request re-analysis, please enter a new order in APCONS, and use the free-text to indicate "re-analysis of case PD-##-####." Only cases previously successfully processed through the Solid Tumor Sequencing Assay, v2 (analyzed at CPD from approximately October 2016 to present) are eligible for reprocessing.

For additional information and questions: Call the Center for Personalized Diagnostics (215-615-3966) weekdays during regular business hours or visit our website: http://pennmedicine.sitecoreauthoring.uphs.upenn.edu/departments-and-centers/center-for-personalized-diagnostics/gene-panels

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