Penn Medicine

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To: **UPHS Physicians and Staff**

From: **The Division of Precision and Computational Diagnostics (PCD)**
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Re: **Launch of NEW Solid Tumor NGS Panel at 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 transition to **PennSeq™** for solid tumors, detecting single nucleotide variants, insertions and deletions, and select copy number alterations with diagnostic, prognostic and therapeutic utility in various solid tumor malignancies. PennSeq™ will also provide an estimate of tumor mutational burden (TMB). **PennSeq™Solid, will replace the current Solid Tumor Sequencing Panel (STP) for samples received in-lab beginning April 26, 2022.** PennSeq™ Solid sequences all coding regions and segments of the flanking intronic regions of the 152 genes currently reported in the STP, plus 31 additional genes (indicated by bold text in the gene list below). While results between the STP and PennSeq™ were highly concordant during validation, technical factors may affect detection of variants, especially at the validated limits of detection of each assay. If you have any questions about interpreting historical STP and PennSeqTM results for the same patient, please contact the appropriate PCD faculty member listed in the clinical report.

This panel will not detect fusion transcripts. For more details on assay design, please

contact the CPD (see below). At this time, there is no change to the PennSeq™ Heme, Fusion Transcript, and Penn Precision Panels.

SAMPLE REQUIREMENTS: The acceptable sample types for this assay include: peripheral blood, bone marrow aspirates, formalin-fixed paraffin embedded (FFPE) tissues, and tissue or fluid in PreservCyt solution. This assay is agnostic regarding the tissue-type of the specimen. Decalcified specimens and specimens containing less than 10% viable nucleated neoplastic cells are not acceptable.

TURNAROUND TIME: 14-21 business days

ORDERING INFORMATION: In PennChart, select "Anatomic Pathology Consult" (APCONS). Choose from "Special Studies" drop down menu:

- **CPD: PennSeq Solid. If limited specimen, perform Penn Precision Panel***
- **CPD: PennSeq Solid only (no reflex testing)**

*The Penn Precision Panel is a "hot spot" cancer sequencing panel that covers only select regions of 59 common cancer genes. Since it may not be appropriate for all tumor types, please refer to the Lab Test Service Guide or CPD Website for more information about the panel design before ordering.

For additional information and questions: Call the Center for Personalized Diagnostics (215-615-3966) weekdays during regular business hours or visit our website: https://www.pennmedicine.org/departments-and-centers/center-for-personalized-diagnostics.

Genes included (183 total genes):

ABL1, AKT1*, AKT2, AKT3*, ALK, APC, AR, ARAF, ARID1A, ARID2, ATM, ATRX, AURKA, **AXIN1, B2M**, BAP1, **BCL2**, BRAF*, BRCA1, BRCA2, BRIP1, BTK, CCND1, CCND2, CCND3, CCNE1, CDH1, CDK4, CDK6, CDKN2A, **CDKN2B**, CHEK2, CIC, CREBBP, CRKL, CSF1R, CTNNB1*, DAXX, DDR2*, **DDX41, DICER1**, DNMT3A, EGFR*, EIF1AX, EP300, **EPCAM**, EPHA3, ERBB2*, ERBB3, ERBB4, ERCC2, ERG, ESR1*, ESR2, EZH2, FBXW7, FGF3, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, **FOXL2**, FUBP1, GATA3, GNA11, GNAQ, GNAS, H3-3A, **HNF1A**, HRAS*, IDH1, IDH2, IGF1R, **IKZF1**, JAK1, JAK2, JAK3, KDM5A, KDM5C, KDM6A, KDR*, KIT*, KMT2C, **KMT2D**, KRAS*, MAP2K1, MAP2K2, MAP2K4, MAPK1, MAPK3, MAX, MCL1, MDM2, MDM4, MED12, MEN1, MET*, MITF, MLH1, **MLH3, MPL**, MRE11,

MSH2, **MSH3**, MSH6, MTOR, **MUTYH**, MYC, MYCN, NBN, NF1, NF2, NKX2-1, NOTCH1, NOTCH2, NOTCH3, **NPM1**, NRAS*, NTRK1, NTRK2, NTRK3, PAK1, PALB2, PBRM1, PDGFRA*, PIK3CA*, PIK3CB, PIK3R1, **PMS1, PMS2, POLD1, POLE, POT1, PPM1D, PRPF8,** PTCH1, PTEN, PTPN11, RAB35, RAC1, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAF1, RB1, RET*, RHOA, RNF43, **ROS1, SDHA, SDHB, SDHC, SDHD,** SETD2, SF3B1, SLIT2, **SMAD2,** SMAD4, SMARCA4, **SMARCB1,** SMO, SPOP, SRC, STAG2, STK11, SUFU, SUZ12, SYK, **TERT#**, TET2, TGFBR2, TP53, TRAF7, TSC1, TSC2, TSHR, U2AF1, VHL, WT1 and XRCC2

Bold gene names are new to the PennSeq^{TM} Solid Assay
* Copy number gain is assessed for the following genes: AKT1, AKT3, BRAF, CTNNB1, DDR2, EGFR, ERBB2, ESR1, HRAS, KDR, KIT, KRAS, MET, NRAS, PDGFRA, PIK3CA, RET
# Promoter hotspot analysis also included