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TEST UPDATE

SmartGenomics™ – 35 Gene Solid Tumor NGS and aCGH

Pranil Chandra, DO, Director of Molecular Pathology Services
James Prescott, PhD, HCLD (ABB) Director of Molecular Diagnostic Services
P. Alan Lennon, PhD, FACMG Director of Clinical Cytogenetics, FISH and Microarray Services

Effective February 3, 2014

PathGroup has increased regional coverage within our SmartGenomics™ solid tumor 35 gene next generation sequencing (NGS) panel from 112 to 140 amplicons in conjunction with a move of the test from the Ion PGM™ platform to the Illumina® MiSeq platform. Additionally, PathGroup has changed array comparative genomic hybridization (aCGH) platforms from the CombiMatrix DNArray™ Tumor Profile BAC array to the Illumina 850k CNV SNP array.

With the change in NGS platforms, PathGroup will now offer another 28 amplicons of coverage within the 35 gene assay at no additional cost, including KRAS and NRAS mutation detection for exons 2, 3 and 4. These regions have been reported to be involved in the pathogenesis of numerous human cancers, as well as resistance to anti-EGFR therapeutics in colorectal cancer (CRC) as noted at the 2014 ASCO GI Meeting.

The addition of the Illumina 850k CNV SNP array allows PathGroup to report on copy number variations (CNV) across >22,000 genes, as well as copy neutral loss of heterozygosity (CNLOH). CNLOH is frequently reported in a variety of human cancers and provides both prognostic and predictive information. As with NGS, there is no additional cost for this methodology improvement.

Sensitivity and Specificity:

SmartGenomics NGS Sensitivity*: 99% for base substitutions at ≥5% frequency of mutant allele
SmartGenomics NGS Specificity*: >99% PPV
SmartGenomics aCGH: Detection of malignant cell populations at greater than 20% of the total cell population, with the potential to detect gains/losses of entire chromosomes or chromosomal arms down to 10% of the total cell population.
*Dependent upon specific mutation, sequence context of mutation, and coverage of amplicon.

Ordering Information:

Specimens sent after January 30, 2014 will be tested via the above methodologies. Results reporting will reflect the new methodologies.

New Test Name:	New Test Code:
SmartGenomics Profile (NGS and Array)	SMGPROFILE (NGS and aCGH)
SmartGenomics Next Generation Sequencing (NGS only)	SMGNGS02 (NGS only)
SmartGenomics Cytogenomic Array (aCHG)	SMGACGTUM (aCGH only)

Specimen Collection and Storage:

- Specimen requirements have not changed.

Resulting:

- Result format will include new methodology.

For further questions, please contact Oncology Customer Support at 615-651-0897