Genomic profiling to enable personalized patient care

Next-generation sequencing of 33 genes involved in melanoma for efficient and cost-effective somatic variant analysis

Genes tested include AKT1, ALK*, BAP1, BRAF, CDK4, CDKN2A, CTNNB1, EGFR, ERBB2, ERBB4, FGFR1, FGFR2*, FGFR3*, GNA11, GNAQ, HRAS, KIT, KRAS, MAP2K1, MAP2K2, MET, MTOR, NF1, NRAS, PDGFRA, PDGFRB, PIK3CA, PTEN, RAC1, RB1, RET*, ROS1*, TP53 (*rearrangements also detected)

Concise, expert interpretations by board-certified clinical genomicists, reported back to the ordering physician

Expert consultation available to physicians in result interpretation and in other technical and clinical considerations

Advantages of our improved tests

Additional predictive targets for improved and more immediate clinical utility of tests

Improved reports with easy-to-understand result summaries and interpretations

Improved sensitivity, enabling detection of variants at low allelic fraction

Lower DNA input requirement for successful results from limited tissue samples
Testing methodology

Tests are performed using targeted hybridization capture of tumor-derived genomic DNA coupled with next-generation sequencing (NGS). This approach enables deep, comprehensive coverage of all coding exons and key introns of ordered genes, and allows assessment of the molecular complexity of each DNA specimen, minimizing sampling bias even in cases of low DNA mass or quality.

Types of variation detected include single nucleotide variants (SNVs), small insertions and deletions (indels), selected larger indels, and structural rearrangements involving selected genes.

This test is routinely performed using formalin fixed paraffin embedded (FFPE) tissues and is able to detect SNVs under 10% allelic fraction in the sequenced tissue.

Results and interpretation

DNA sequence data are analyzed by GPS’ clinically validated bioinformatics pipeline to identify and annotate somatic variants associated with cancer.

Identified tumor mutations are interpreted by a board-certified clinical genomicist in the context of the patient’s disease and other clinical findings, highlighting mutations associated with specific treatment options based on evidence from the medical literature.

Results are returned to the ordering physician in a concise clinical report.

Specimen requirements

Specimen types accepted include excisional biopsies, core needle biopsies, cell blocks or bone marrow aspirate.

Acceptable materials for submission include tumor-containing formalin fixed paraffin embedded (FFPE) blocks, unstained slides from tumor-containing FFPE block or bone marrow in a lavender-top EDTA tube.

Tissue fixation protocols must be compatible with molecular testing; EDTA decalcification is acceptable, acid decalcification is not.

Turnaround time

The turnaround time for testing and interpretation is three weeks from the time a specimen is received.

Ordering

To order a test, submit a completed requisition form (available on our website) by fax or email. Testing is covered by most insurance. GPS performs insurance preauthorization.

In the case of archival specimens, GPS coordinates sample acquisition. For surgical pathology materials requested from outside BJC Healthcare, please include the release form available on our website. Please contact us for more information.

Ancillary testing

Other available services include expert pathologic evaluation by specialized dermatopathologists and chromosomal microarray (CMA) analysis for copy number interpretation of the submitted specimen. Contact us for more information.