

CANCERXOME™ APPROACH

PGDx has developed proprietary techniques for analysis of cancer exomes, including methods for extracting genetic material from frozen or fixed tumor samples, high-throughput next-generation sequencing (NGS), patented approaches for novel genetic analyses, and proprietary data analysis algorithms.

CancerXome™ captures and analyzes the coding regions of >20,000 genes. Both tumor and normal samples are requested and analyzed using proprietary methods that accommodate low abundance, poor quality sample DNA. Samples are sequenced at high coverage to identify tumor-specific (somatic) mutations and copy number changes.

CANCERXOME™ HIGHLIGHTS

- Detailed visual inspection and curation of tumor-specific mutations by world-class cancer bioinformatic experts
- DNA extraction methods that accommodate poor quality and low cellularity tumor samples
- Proprietary analysis algorithms to identify bona-fide sequence changes and to exclude sequence artifacts
- Identification of mutated genes with biologic or clinical implications in human cancer
- Analysis algorithms to evaluate genes and pathways enriched for alterations
- Patented Digital Karyotyping analysis for high resolution annotation of copy number alterations
- CHASM analysis algorithms to evaluate mutation properties and importance of non-hotspot mutations

COMPREHENSIVE CANCERXOME™ ANALYSIS DELIVERABLES

- Pathological evaluation of tumor sample
- Tumor-specific sequence alterations (single base and small indel alterations)
- Tumor-specific copy number alterations
- Functional impact of mutations (predicted protein alterations and domain consequences)
- Mutated genes and pathways with biological or clinical implications
- In-depth COSMIC analysis for recurrent mutations across tumor types
- Proprietary CHASM analysis for identification of driver mutations
- Data summary statistics (read data and depth distribution across target regions)
- Integrated Analysis Report (incidences and frequencies of mutations identified)

CANCERXOME™ SEQUENCING DELIVERABLES & ANALYSES

Analysis Metrics	CancerXome™
Regions Analyzed	>20,000 genes
Sample Prep and NGS Sequencing	✓
Sequence Mapping	✓
Somatic Mutation Analysis	✓
Copy Number Analysis	✓
Germline Variant Analysis	✓
Pathway and Functional Analysis	✓
Integrated Project Analyses	✓

*Germline variant analysis is optional

CANCERXOME™ SEQUENCING KEY METRICS

Regions Analyzed	Coding regions of >20,000 genes
Sequencing Method	Illumina next generation sequencing
Bioinformatics	Proprietary methods and visual inspection
Assay Sensitivity	>95%
Assay Specificity	>99.99%
Sequencing Coverage	150x
Turn-around Time	6 weeks (rush available)
Sample Requirements	Tumor only or tumor and matched normal (optimal results)
Sample Types	Frozen tumor, FFPE, cell lines, blood, saliva, and xenograft
DNA Input Required	1 ug (minimum 50 ng)

Related References

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