# Client Overview

The client is one of the largest businesses supporting genome biology for transforming cancer care by application of genome technology, cancer biology, clinical oncology and information science. By means of harnessing new genomic technologies, the client aids disease gene discovery and helps in drug designing for personalized medicines.

# Challenges

The company envisioned to offer software and services for cancer researchers, pharmaceutical companies, and health-care organizations. The goal was to provide the data processing and analysis workflows required for personalized cancer therapy, in which treatments are matched to the specific genetic aberrations found in an individual patient's cancer cells. The client was facing a daunting task of development and execution of appropriate next generation sequencing bioinformatics pipelines to support analysis of cancer genomics data. For the same, the client needed to augment its internal resources with additional experts in NGS bioinformatics. Given the complexity of the project and the domain expertise it demanded, the client was challenged to find a competent Life Science informatics organization with multidisciplinary expertise in the field of genomics. In addition to knowledge of various NGS platforms, the client's project required specialized knowledge in cancer genetics, both with respect to research and clinical experience in cancer genetics to test and validate the bioinformatics pipeline and to interpret DNA sequence information obtained from cancer tumors. Clearly, a highly skilled and specialized partner was needed to address the client's challenges.

# Solution

Optra HEALTH, with its multidisciplinary team and vast genomics experience - all under one roof - was found to be the perfect partner to address the client's unmet needs. Given that Optra had domain experts on their team, they were able to train the rest of the team quickly and efficiently.

> ➢ Optra systems helped the client to develop, execute and validate bioinformatics pipelines to analyze huge amount of raw of cancer genomics data
> ➢ The raw data output from ChiSeq data from Illumina and SOLiD platform was analyzed by using Galaxy Server.
> ➢ Optra with its expertise on Galaxy Server analyzed and annotated tumor and control sample data.
> ➢ Systematic sample handling and workflow was maintained.
> ➢ Workflow for Primary analysis like Quality estimations, mapping, Peak calling, Annotations, Read density plots and heatmap generation was rigorously tested and executed.
> ➢ Pipeline had functionalities to rapidly filter variants and identify those which were biologically relevant. Domain experts validated the filtered variants to ensure that they were indeed clinically relevant and significant.
> ➢ Functionalities to perform Pedigree analysis and study segregation were also executed in the pipeline.
> ➢ Python scripts were incorporated in the pipeline for retrieving annotations from various Cancer Databases.
> ➢ Finally an entirely automated workflow was created on Galaxy server for complete analysis of cancer genome.

Optra HEALTH was proactive in taking a simplified, stepwise approach in rapid assessment of significant variants with rich annotations. This approach displayed a high precision and sensitivity in identifying causal, de novo gene disease relationships.

# Technology Environment

> ➢ Tools used: Galaxy Server
> ➢ Scripting languages: Perl, Python

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# Benefits

- By understanding the requirements for automated NGS data analysis pipeline, Optra successfully delivered reproducible workflow for next generation sequencing analysis
- Optra designed, executed and validated a comprehensive analysis package to answer a specific problem i.e. cancer genomics, in which most the analysis algorithms were specifically written for Galaxy
- Using this pipeline the user could run a NGS data file through a series of preexisting tools, with context-specific parameters for Galaxy workflow
- Streamlined annotations were made possible by linking pipeline to public data sources like dbSNP, 1000 genomes, OMIM, COSMIC, IPA and more.
- Simplified reporting was possible using descriptive maps, charts and tables.

# About Optra Health

Optra Health is an ISO-certified global organization with deep domain expertise in medical devices, lab automation, life science informatics and healthcare IT solutions. The company provides a fully-scalable, cost-effective OptiShore™ delivery model. This enables customers to choose the optimal balance between on-site, on-shore, and off-shore development that will best address their budget and collaboration requirements. With Optra Health, customers are able to shrink their time-to-market by leveraging practical, building-block based solutions. Committed to clear communication and total transparency, the company consistently meets or exceeds its clients' expectations. Offering a full complement of expert engineering and consulting services, Optra Health is aligned to real business needs applied over the entire product development lifecycle. The robust, scalable and efficient IT infrastructure of the company, together with its outstanding project management team, consistently ensures superior results. Optra Health's global delivery model helps its customers cut costs by about 50% without compromising on quality and realize a 200% improved production cycle. Visit Optra Health today: http://www.optrahealth.com

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