# Mutation-Disease-Drug database for Personalized Medicine

## Client Overview

The client is the pioneer of genome-scale diagnostics services and contract research organization specializing in genome guided medicine. The company is focused on producing the most accurate genetic sequence data from samples using analytics and proprietary content to draw accurate and reliable biomedical interpretations of the data. Its genome services provide academic, pharmaceutical, biotech, and clinical researchers an accurate and comprehensive end-to-end human genome sequencing and analysis solution.

## Challenges

As part of an international collaborative project to annotate the genome of an individual, the client required a comprehensive database of association data of mutations and human disease phenotypes along with drug data. A new database was required because no single public resource offered the capabilities necessary for the clinical annotation of a whole genome. The ultimate aim of the data is to aid the creation of new analytical tools and statistical approaches geared towards personalized medicine that would allow physicians to select evidence parameters of their choice.

Variations in individuals' genetic profiles many times correlate with differences in how individuals develop diseases and respond to treatment. Personalized medicine supported by genetic and genomic assays has the potential to facilitate optimal risk identification, disease screening, disease diagnosis, therapy and monitoring. The objective was curating, collating and analyzing all this plethora of information to cater the personalized needs of individuals to improve clinical outcomes significantly.

Optra was found to be a perfect partner with requisite experience and capabilities in data curation and scientific data mining to deliver client's expectations.

## Solution (Data curation for building mutation-disease-drug database facilitating decision support for personalized medicine)

Optra Systems built a new database of mutation-disease-drug entity culled from a number of publicly available sources. For each mutation it captured disease phenotypes, study and sample information, statistical data for association parameters and frequencies of the variants and drug data encompassing drug dosage and ADR (Adverse Drug Reaction) all under a single effort.

- A literature survey of published papers on mutation-disease- drug reaction association was carried out through PubMed to extract disease associations from tables, full text, and supplementary data.
- Broad level disease name or condition under study along with the corresponding MeSH qualifier was collected. Under this broad level, the individual associations between the mutation and the drug entity were recorded.
- Data was recorded for both case-control and cohort studies. The sample description included the sample size, gender, ethnicity and other information pertaining to the particular sample under study. The ethnicity was later mapped to the HapMap phase III or 1000 genome populations code.

# Benefits

- Optra System's domain expertise assisted the client to create a comprehensive and novel database of mutation-disease-drug associations as an initial step towards deriving clinically meaningful information about disease risk and response to drugs in patients with whole genome sequence data.
- The extensive manual curation involved in the project (400,000+ papers and ongoing) enabled the capturing of all relevant information onto a single schema for downstream automated usage given the lack of standardized reporting format for association studies.
- An important benefit of this new database over the preexisting ones is that it indicates the risk of adverse drug reaction and beneficial drug response specific to particular genotype which will optimize personalized medicine.
- Based on this database, healthcare professionals will be able to provide more tailored prevention & treatment programs for the patients; results in improved patients' outcomes and reduced adverse events making it more cost effective.



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# About Optra Systems

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