Gene Mutation AI

H2O.ai and Snowflake Partner in Health

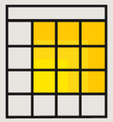
2022

Automated Machine Learning for

DNA Analysis

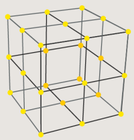
The Data

DNA information is distributed across more than 20,000 genes and half a million clinically relevant mutations. There are nearly 22,000 columns of data in the gene sequences of each patient which H2O.ai and Snowflake can process in mere seconds.



Autogenerated Code

Convert VCF files to a dataset that is suitable for machine learning. The SQL is dynamically generated and predictions are automatically created for every single patient in the table in a matter of seconds.

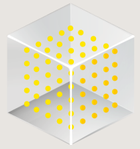


Machine Learning

Evaluate patient risk of disease and understand how common this risk has been among past patients. The pretrained model that best fits the data is automatically selected from all available models.

The Solution

H2O Gene Mutation AI is a machine learning-powered patient risk assessment application that uses the Snowflake Data Cloud to bring intelligent clinical decision support directly to clinicians and provide advanced analytics for pharmacogenomics and pharmacovigilance pipelines.



Model Interpretability

Automated drift detection identifies any variance between the training data and the specific table being used for model scoring. Model transparency reveals which specific cellular pathways have played a role for the diagnosis.

Explainable Insights

Shapley reason codes provide insights at the local gene level and at the global level for the entire population represented in a given table. Different distributions show how an individual's results compare to those of similar patients.

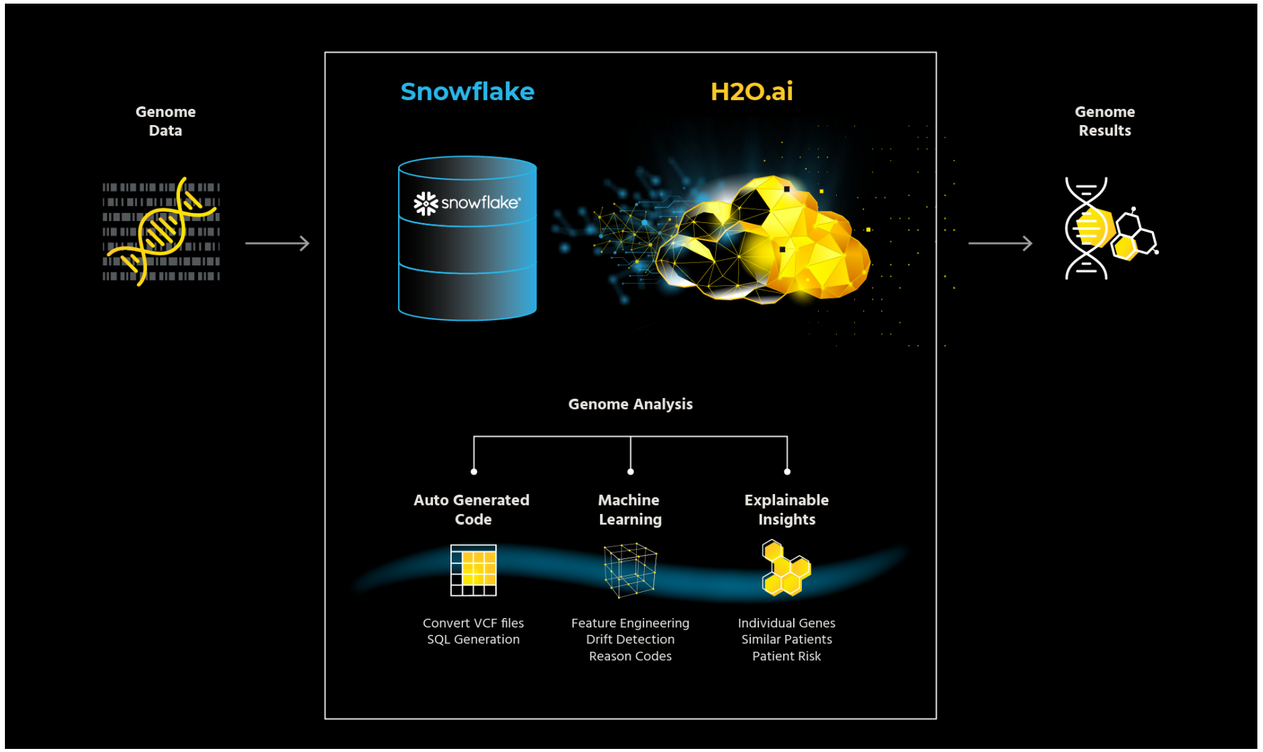
Gene Mutation AI

How it Works

H2O Gene Mutation AI combines the Snowflake database and the H2O AI Cloud to bring intelligent clinical decision support directly to clinicians and provide advanced analytics for pharmacogenomics and pharmacovigilance pipelines.

DNA data can be massive, which makes the Snowflake Data Cloud an ideal place to store, cleanse and prepare the data for analysis.

H2O.ai's advanced machine learning pipeline processes massive datasets and extracts valuable information based on a patient's mutation profile.



Gene Mutation AI

Core Capabilities

State-of-the-Art Clinical Decision Support

Make AI part of the triage process, leveraging ML and patient data tables to evaluate disease risk.

Discover the Drivers for Individual Risk

Visualize and explore drivers automatically extracted from ML algorithms to estimate patient risk and store that output in table format.

Explore Avenues for Therapeutic Interventions

Understand how patient risk is affected by various factors, in a dose-response type what-if analysis, and store the results in tables.

Identification of Similar Patients

Look up tables of historical patients whose outcomes are known, find the most similar ones to the specific patient and understand the similarities.

Evaluation of Demographic Biases

Use historical patient tables to evaluate any unintended biases in the model due to lack of demographic diversity in the patient population.

Integration of Multiple Data Types

Leverage a variety of data types, tables with different molecular results, clinical measurements, or medical notes for the same patients to further refine the disease model.

Gene Mutation AI

Local and Global Insights

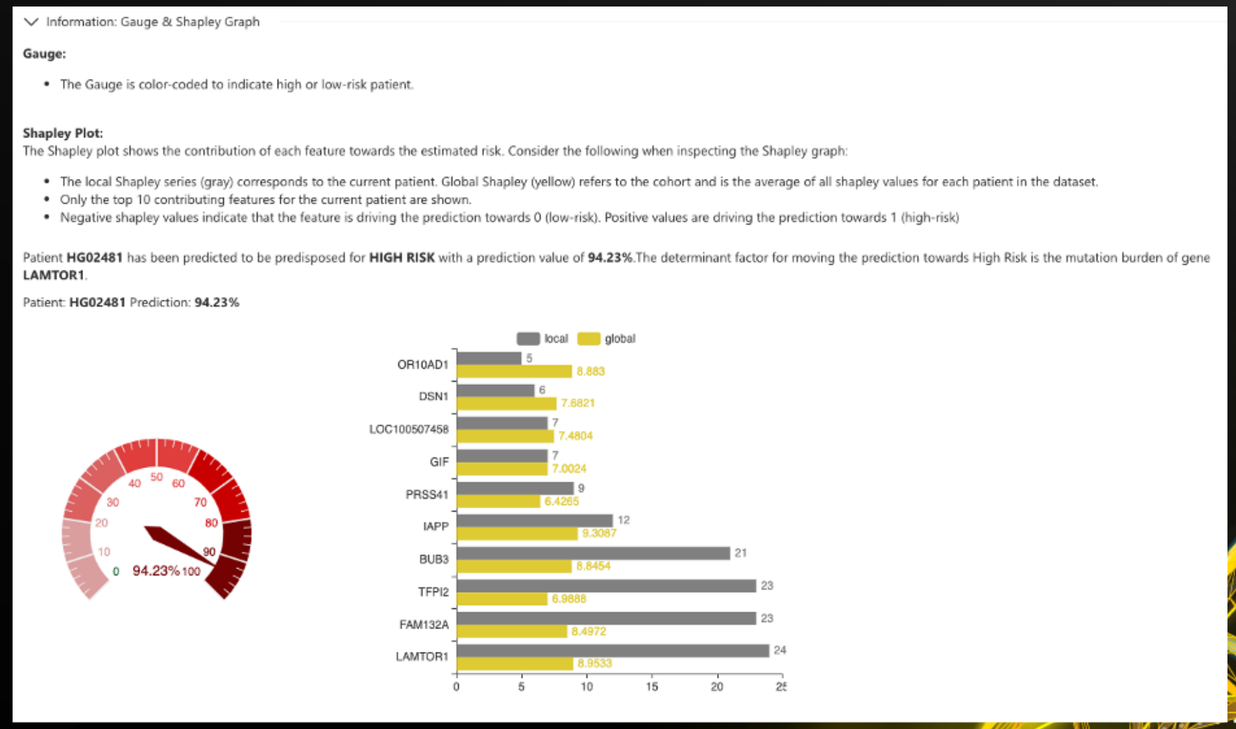
01

Information Gauge and Shapley Graph

The Gauge offers visually the patient's risk to the disease. The same information is presented within the relevant text at the top of the panel. The text also offers the classification of this patient as having HIGH or LOW RISK based on the risk score.

The Shapley plot to the right of the panel shows the patient's genetic contribution to the reported risk (grey bars, label local), in comparison to the average contributions across the cohort (yellow bars, label global).

Each bar is one gene of the top ten most contributing genes for this patient and indicate a contribution towards high or low risk, respectively.



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Similar Patient Comparisons

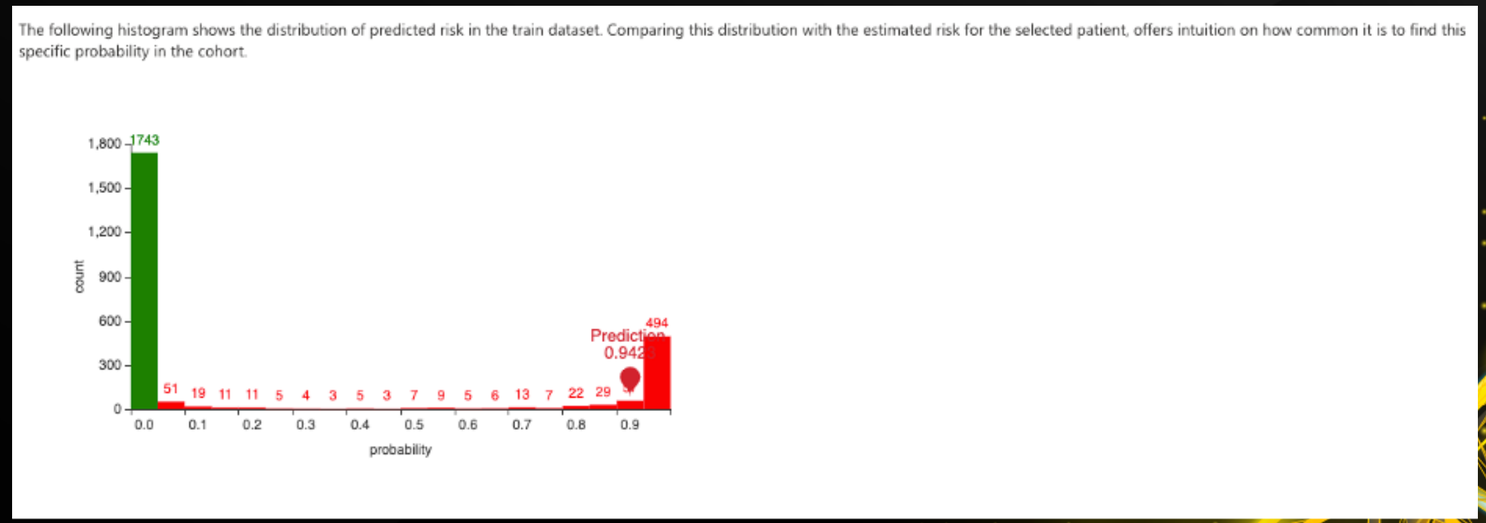
02

Histogram

This panel helps evaluate how confident we should be in declaring a patient with this specific risk score (Gauge) as HIGH or LOW RISK.

The histogram shows how risk is distributed across the entire patient cohort. LOW RISK is marked with green color and HIGH RISK with red. The selected patient's risk is highlighted with a pin in the right position.

The further right a HIGH RISK patient is the better confidence in the evaluation. On the other side, the further left a LOW RISK patient is the better.



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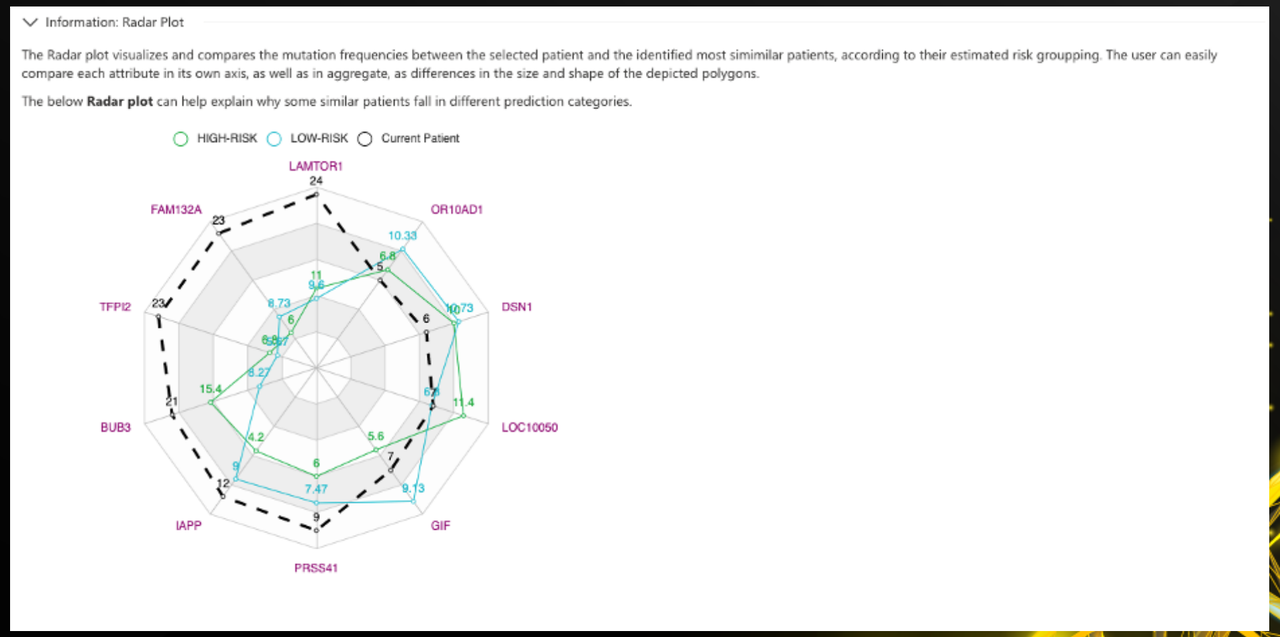
Individual Patient Risk

03

Radar Plot

A radar plot shows the individual patient's risk and identifies the specific factors behind those predictions.

he radar plot offers an explanation of the gene-level similarities and differences between the selected patient and the twenty most similar other patients in the cohort. The results are presented comparatively for the different risk groups. A group can be selected or deselected by clicking on the corresponding label at the plot's legend.



Gene Mutation AI

Key Users

Data Scientists

Genomic data is complex to handle at scale. Gene Mutation AI leverages H2O.ai models and Snowflake Data Cloud for speed and scalability, so that each of the technologies work cooperatively.

Pharmacologists

Easily understand genetically-driven, patient-specific drug efficacy and adverse effects.

Physicians

With an easy-to-navigate interface that is easily accessible from a tablet or smartphone, physicians can examine a patient's genetic profile and gain insights on-demand.

Gene Mutation AI

Benefits

Accuracy

Additional data like other molecular results, clinical measurements, medical notes, images and more can be used to augment the initial model, gain more comprehensive insights and ultimately improve the quality of the patient's care plan.

Trust

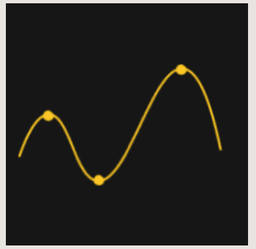
Doctors can visually discover the genetic factors that impact individual risk and explore personalized avenues for therapeutic intervention. Model transparency allows doctors to see which specific cellular pathways have played a role for the diagnosis.

Speed

The pretrained model that best fits the data is automatically selected from all available models based on metadata within Snowflake data tables and metadata within the H2O machine learning models. Thousands of rows of DNA data are analyzed in seconds.

Scale

H2O.ai's gene mutation solution is flexible enough to analyze different types of disease outcomes without any additional coding requirements. The application supports multiple pretrained models, so the same patient can easily be examined for their risk of multiple diseases.



Flexibility

The power of Gene Mutation AI is accessible on any computer, tablet or smartphone, allowing physicians to explore the results while making rounds and discussing results at patient bedsides.

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Free Trial

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