



FINAL PROJECT PROPOSAL

INFO 6105 Data Science Engineering Methods

Advising Professor
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Title:

Personalized Medicine: Redefining Cancer Treatment

Predict the effect of Genetic Variants to enable Personalized Medicine

And

Classifying clinically actionable genetic mutations

Team Members

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Description:

A lot has been said during the past several years about how precision medicine and, more concretely, how genetic testing is going to disrupt the way diseases like cancer are treated.

But this is only partially happening due to the huge amount of manual work still required. Once sequenced, a cancer tumor can have thousands of genetic mutations. But the challenge is distinguishing the mutations that contribute to tumor growth (drivers) from the neutral mutations (passengers).

Currently, this interpretation of genetic mutations is being done manually. This is a very time-consuming task where a clinical pathologist has to manually review and classify every single genetic mutation based on evidence from text-based clinical literature.

Problem Statement:

We need to develop a Machine Learning algorithm that, using this knowledge base as a baseline, automatically classifies genetic variations.

This problem was a competition hosted on Kaggle and launched by Memorial Sloan Kettering Cancer Center (MSKCC), accepted by NIPS 2017 Competition Track

The Link to the competition on Kaggle: <https://www.kaggle.com/c/msk-redefining-cancer-treatment>

Data Description:

In this competition, we will develop algorithms to classify genetic mutations based on clinical evidence (text).

There are nine different classes a genetic mutation can be classified on.

This is not a trivial task since interpreting clinical evidence is very challenging even for human specialists. Therefore, modeling the clinical evidence (text) will be critical for the success of your approach.

Both training and test, data sets are provided via two different files. One (training/test_variants) provides the information about the genetic mutations, whereas the other (training/test_text) provides the clinical evidence (text) that our human experts used to classify the genetic mutations. Both are linked via the ID field.

Therefore the genetic mutation (row) with ID=15 in the file training_variants, was classified using the clinical evidence (text) from the row with ID=15 in the file training_text

File descriptions

- training_variants - a comma separated file containing the description of the genetic mutations used for training. Fields are ID (the id of the row used to link the mutation to the clinical evidence), Gene (the gene where this genetic mutation is located), Variation (the amino acid change for this mutations), Class (1-9 the class this genetic mutation has been classified on)
- training_text - a double pipe (||) delimited file that contains the clinical evidence (text) used to classify genetic mutations. Fields are ID (the id of the row used to link the clinical evidence to the genetic mutation), Text (the clinical evidence used to classify the genetic mutation)
- test_variants - a comma separated file containing the description of the genetic mutations used for training. Fields are ID (the id of the row used to link the mutation to the clinical evidence), Gene (the gene where this genetic mutation is located), Variation (the amino acid change for this mutations)
- test_text - a double pipe (||) delimited file that contains the clinical evidence (text) used to classify genetic mutations. Fields are ID (the id of the row used to link the clinical evidence to the genetic mutation), Text (the clinical evidence used to classify the genetic mutation)