

1158-92-287

Mario Banuelos* (mbanuelos22@csufresno.edu), **Omar DeGuchy** (odeguchy@ucmerced.edu), **Suzanne Sindi** (ssindi@ucmerced.edu) and **Roummel F. Marcia** (rmarcia@ucmerced.edu). *Related Inference: A Supervised Learning Approach to Detect Genomic Variation.*

The human genome is composed of 3 billion nucleotides, given by the letters A,C,G,T. Typically, organisms in the same species have similar genomes that differ by only a few sequences of varying lengths at varying positions. These differences may be observed in the form of regions where letters are inserted, deleted, or inverted. These anomalies, or novel rearrangements, are known as structural variants (SVs) and are often difficult to detect. Standard approaches for identifying such variation involves comparing fragments of DNA from the genome of interest to a reference genome. This process is usually complicated by errors produced in both the sequencing and mapping process which may result in an increase in false positive detections. In this work, we propose a gradient boosting and neural network approach for reducing the number of false positives. We focus our attention on the deletions detected by the popular SV tool delly. In particular, we consider the ability of simultaneously considering sequencing data from a parent and a child using a neural network and gradient boosting. We compare the performance of each method on simulated and real parent-child data and show that including related individuals in training data greatly improves the ability to detect true SVs. (Received March 03, 2020)