GENoML: AUTOMATED MACHINE LEARNING FOR GENOMICS

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ABSTRACT

GenoML is a Python package automating machine learning workflows for genomics (genetics and multi-omics) with an open science philosophy. Genomics data require significant domain expertise to clean, pre-process, harmonize and perform quality control of the data. Furthermore, tuning, validation, and interpretation involve taking into account the biology and possibly the limitations of the underlying data collection, protocols, and technology. GenoML’s mission is to bring machine learning for genomics and clinical data to non-experts by developing an easy-to-use tool that automates the full development, evaluation, and deployment process. Emphasis is put on open science to make workflows easily accessible, replicable, and transferable within the scientific community. Source code and documentation is available at https://genoml.com.

Keywords Genomics, multi-omics, machine learning, AutoML

1 Introduction

In recent years, the demand for machine learning (ML) expertise has outpaced the supply, despite the surge of people entering the field. To address this gap, there have been significant strides in the development of user-friendly machine learning software that can be used by non-experts. The first steps toward simplifying machine learning involved developing simple, unified interfaces to a variety of machine learning algorithms (e.g., scikit-learn [1], XGBoost [2], LightGBM [3], TensorFlow [4], PyTorch [5]). Although these packages have made it easy to experiment with machine learning, there is still a fair bit of knowledge and background in data science required to produce high-performing and usable machine learning models. This demand has given rise to the area of automated machine learning (AutoML [6]). Some of the recently developed and popular AutoML systems include Auto-WEKA [7], hyperopt-sklearn [8], Auto-sklearn [9], TPOT [10], and Auto-Keras [11].

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However, different data require different ML pipelines. The development of ML models for genomics (genetics and multi-omics) data, in particular, is notoriously difficult for a non-expert. These data modalities require significant domain expertise to clean, pre-process, harmonize and perform quality control (QC) [12]. Furthermore, tuning, validation, and interpretation involve taking into account the biology and the limitations of the underlying data collection, protocols, and technology.

For ML to truly be accessible to non-experts in the genomics and clinical research areas, we have designed an easy-to-use tool, called GenoML, that automates the full development, evaluation, and deployment process. GenoML provides an end-to-end framework for genomic datasets, including the most complex parts of the process, such as data pre-processing and cleaning, to more advanced training and tuning. GenoML intelligently explores many possible techniques to find the best model for the specific input data. GenoML is also helpful to advanced users; it provides a high-level wrapper performing many modeling tasks that would typically require many more lines of code.

Furthermore, GenoML is more than a package. Since its inception, it has evolved into a diverse community with integrative expertise in data science, bioinformatics, computer science, software engineering, biology, and healthcare. GenoML contributors are staunch advocates of open science, striving to make data and code easily accessible to the scientific community. Please join us and contribute to the development of GenoML.

2 GenoML Principles and Philosophy

GenoML developers advocate open science. The following are the underlying principles of GenoML development:

- Little to learn - The goal of GenoML is to democratize complex genomics and machine learning workflows. Thoughtfully designed for newcomers, if a user can ‘cd’ or ‘ls’, they should be able to use GenoML.
- Intuitive - Everything has to be simple, straightforward, and effective, from data munging to a tuned model in a few lines of code.
- Layered architecture - GenoML is more than a tool; it is an ecosystem that will continuously grow, experimenting with new ideas and innovations. Workflows are kept in logical layers; to change or update one module and not affect the others.
- Intelligent defaults - Systematic research is done to set optimized defaults for varying inputs. The default settings are sensible and validated for most workflows to keep modules un-cluttered and to run smoothly. At the same time, providing manual options for advanced users.
- No vendor lock-in - Integration with other code, products, and platforms should be hassle-free. GenoML is open source and will remain free and public under the Apache 2.0 license.
- Safe and inclusive community - GenoML is a community for positivity in research and open science for the public good. Code of Conduct is adapted from the Contributor Covenant, version 2.0 [13].

3 Project Vision

We foresee a GenoML expansion from primarily an AutoML to a more broadly applicable framework. The next phases of GenoML focus on building an ecosystem of machine learning tools for genomics. Other components would include:

- GenoML Genetics: general genetics pipeline tools
- GenoML Deploy: designed for deploying the ML models for inference in practice
- GenoML Portal: an interface designed for clinicians/physicians for use in practice. It also provides model explanation information
- GenoML Federated: federated learning of GenoML. A critical component in light of recent privacy regulations such as GDPR [14]. Enables learning across multiple data silos
- GenoML Meta: meta-learning aspect of GenoML. Enabling learning and selection across data across diverse datasets and study populations
- GenoML Python: library developed to integrate seamlessly with other scientific Python libraries
- GenoML Higher API: higher-level APIs, enabling more community developments
- GenoML Model Zoo: a place to share models trained on public or private datasets
4 Conclusion

GenoML automates a wide variety of machine learning pipelines for genomics and multi-omics. Since it relies on other open-source Python packages, it can easily be integrated into existing systems and analytical protocols. We hope for broad adoption and contributions by the community, enabling more unified pipelines producing transparent and reproducible results. Future work includes expansion from primarily an AutoML to an ecosystem of machine learning tools for genomics and more general-purpose AutoML in epidemiological and other health-related domains.

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