ELIXIR-GR: Bioinformatics tools for analysis of next-generation sequence data and an application example

Fotis Tsetsos[1] Margaritis Tsifintaris[1] Manolis Apostolos[2] Papageorgiou Aristotelis[2] Alexis Galanis[1] Peristera Paschou[1]

[1]Department of Molecular Biology and Genetics, Democritus University of Thrace, Alexandroupolis, Greece
[2]Department of Forestry and Management of the Environment and Natural Resources, Democritus University of Thrace

Summary

Here we present three web-based platforms (PCASNPs, VariantRanker, NetworkAnalyzer) for the analysis of high-throughput genetic experiments, along with a framework for genomics pipelines, within the framework of the ELIXIR-GR Infrastructure.

ELIXIR-GR

ELIXIR-GR is the Greek National Node of the ESFRI European RI ELIXIR, a distributed e-Infrastructure aiming at the construction of a sustainable European infrastructure for biological information. ELIXIR-GR supports life-science research and its translation to medicine, biological sciences and society. It offers a catalogue of tools, services and benchmarks, ensuring best practices as well as sustainability and interoperability with other biological and medical science infrastructures.

VariantRanker

Variant Ranker is a tool for ranking and annotation of coding and non-coding variants and facilitates the identification of causal variants based on novelty, effect and annotation information. Users can query and filter large amounts of high-throughput data based on user custom filter requirements and apply different models of inheritance.

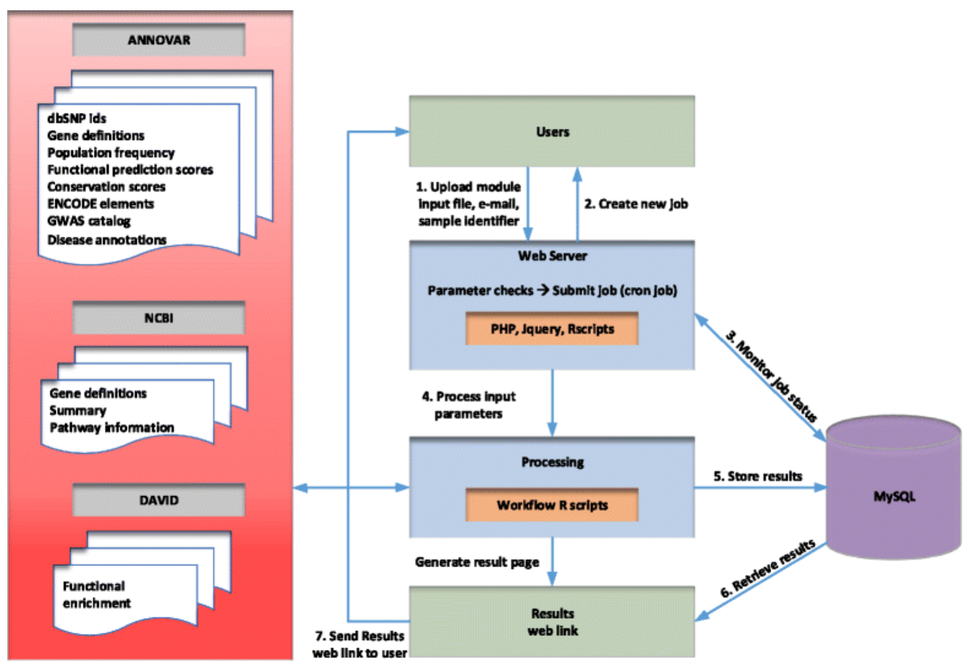

Figure 1: Workflow depiction of VariantRanker

NetworkAnalyzer

NetworkAnalyzer generates functional networks out of identified genes. Users can submit top candidate gene symbols and identify overlapping genes from different functionally enriched annotation categories like pathways/ontologies/diseases.

PCASNPs

PCASNPs is a robust and widely popular method for the identification of ancestry informative markers for diverse populations. It may be used to place individuals within a few miles from their region of origin with applications in forensics but also in order to correct for stratification in genetic association studies and to investigate the historic relationships of human populations.

	Anaemia (*PKLR*, recessive model)	Pfeifer (*FGFR2*), dominant model)	Miller (*DHODH*, recessive model)
VariantRanker	1	1	1
eXtasy	436	628	1588
wANNOVAR	12	90	12
PhenIX	1	1	6
wKGGSeq	1	6	3

Figure 2: Candidate rank comparison using similar web-tools with three of our validation data sets

Genomic pipeline framework

The toolset is also acompanied by a highly sophisticated genomic pipeline framework designed for the simple, fast and accurate analysis of high-throughput genomic experiments, focusing on DNA-seq, RNA-seq, ChIP-seq, CLIP-seq and DNAase-seq.

Acknowledgements

We acknowledge support of this work by the project “ELIXIR-GR: Hellenic Research Infrastructure for the Management and Analysis of Data from the Biological Sciences” (MIS 5002780) which is implemented under the Action “Reinforcement of the Research and Innovation Infrastructure”, funded by the Operational Programme “Competitiveness, Entrepreneurship and Innovation” (NSRF 2014-2020) and co-financed by Greece and the European Union (European Regional Development Fund). This work was also conducted within the framework of the project "INTERDISCIPLINARY TRAINING NETWORK FOR TOURETTE SYNDROME; EUROPEAN TRAINING CAPACITIES FOR NEURODEVELOPMENTAL DISORDERS (MATCHING FUNDS)", code KE-81523.

References

[1] John Alexander, Dimitris Mantzaris, Marianthi Georgitsi, Petros Drineas, and Peristera Paschou. Variant Ranker: A web-tool to rank genomic data according to functional significance. *BMC Bioinformatics*, 18(1):341, dec 2017.

Ευρωπαϊκή Ένωση
Ευρωπαϊκό Ταμείο Περιφερειακής Ανάπτυξης
ΕΛΛΗΝΙΚΗ ΔΗΜΟΚΡΑΤΙΑ ΥΠΟΥΡΓΕΙΟ ΟΙΚΟΝΟΜΙΑΣ & ΑΝΑΠΤΥΞΗΣ ΕΙΔΙΚΗ ΓΡΑΜΜΑΤΕΙΑ ΕΤΠΑ & ΤΣ ΕΙΔΙΚΗ ΥΠΗΡΕΣΙΑ ΔΙΑΧΕΙΡΙΣΗΣ ΕΠΑνΕΚ
ΕΠΑνΕΚ 2014-2020 ΕΠΙΧΕΙΡΗΣΙΑΚΟ ΠΡΟΓΡΑΜΜΑ ΑΝΤΑΓΩΝΙΣΤΙΚΟΤΗΤΑ ΕΠΙΧΕΙΡΗΜΑΤΙΚΟΤΗΤΑ ΚΑΙΝΟΤΟΜΙΑ
ΕΣΠΑ 2014-2020 ανάπτυξη - εργασία - αλληλεγγύη
Με τη συγχρηματοδότηση της Ελλάδας και της Ευρωπαϊκής Ένωσης