

# Umberto Perron

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## Profile

- Postdoctoral Researcher in Bioinformatics with 5+ years of experience.
- Experienced in building, processing, annotating, integrating, analysing large biomedical datasets and knowledge bases using statistical, machine learning methods.
- Development of tools and approaches for flexible, scalable, and reproducible data science and predictive modelling.
- Experience leading multidisciplinary biomedical research projects, interacting with diverse stakeholders; planning and coordinating data processing, and analysis.
- Experience working in and collaborating with large international research organizations, research hospitals, and pharma R&D teams.

## Research experience

**Aug 2020 - present** - Fondazione Human Technopole, Milano, IT  
*Postdoctoral researcher*

Team leader: Dr. Francesco Iorio,

- I processed and integrated multiomics data from a colorectal cancer PDX cohort and developed an ensemble ML classifier of Cetuximab sensitivity which outperforms SOTA biomarker signatures.
- I designed and developed a cancer dependency biomedical knowledge graph integrating multiple data sources. This will be served interactively via a web app as well as being used to train graph ML models for drug repurposing.

**Oct 2016 - June 2020** - European Bioinformatics Institute (EMBL-EBI), Cambridge, UK

*Predoc fellow / PhD student*

Supervisor: Dr. Nick Goldman, Dr. Iain Moal

- I developed a novel substitution model for amino acid sequence evolution that makes use of both amino acid identity and side chain rotamer configuration information.
- I designed scalable approaches for annotating protein sequences with structural features computed from X-ray crystallography data.

**May 2019 - Jul 2019** - Medicines Discovery Catapult, Alderley Edge, UK

*Visiting PhD student*

Supervisor: Prof. John P. Overington

- I investigated amino acid side chain rotamer configuration stability in different structural contexts.
- I prototyped a protein variant effect prediction ML model combining sequence conservation and structural features.

**Oct 2014 - Jul 2016** - Università degli Studi di Torino, Torino, IT

*Graduate research intern*

Supervisor: Prof. Paolo Provero

- I developed a pipeline to ingest public RNAseq data, compute a multi-species and multi-tissue co-expression network using Python, R, and Bash.
- I used this co-expression network to annotate uncharacterized lncRNAs with ontology terms associated with protein-coding genes.

## Education

- 2016-2020** *PhD* in Bioinformatics - University of Cambridge, UK / EMBL International PhD Programme  
**2014-2016** *MSc* in Molecular Biotechnology - Università degli Studi di Torino, IT  
**2011-2014** *BSc* in Biotechnology - Università degli Studi di Torino, IT

## Skillset

**Programming:** strong scientific software development, data analysis and visualization in Python, R. Basic C/C++ and Julia.

**Domain knowledge:** trained in molecular biology, biochemistry and pharmacology.

**Bioinformatics:** highly experienced in processing, integrating and analysing large multiomics/clinical datasets. Development of Python/R pipelines for protein sequence and structure analysis, NGS and gene expression analysis, variant and transcript annotation, biomarker and target discovery, drug repositioning, phylogenetics, molecular evolution

**Data Engineering:** ingestion, processing, and retrieval of biomedical and chemical resources. Backend, internal REST API, Neo4j graph database design and development. Basic SQL, MongoDB, Docker, Kubernetes, Unix/Linux, HPC.

**Biostatistics / Machine learning:** predictive modelling, inference, testing, statistical computing with NumPy and SciPy, machine learning with scikit-learn, Keras/TensorFlow. Basic PyTorch.

**Project lead:** experienced in coordinating computational researchers, clinicians, software engineers, data curators, facility staff; communicating results and priorities, drafting analytical plans, scientific grant and manuscript writing and editing.

## Selected publications

### Published

- Vinceti, A., R. R. De Lucia, P. Cremaschi, Perron, U., E. Karakoc, L. Mauri, C. Fernandez, K. H. Kluczynski, D. S. Anderson, and F. Iorio (2022). “CRISPRcleanR WebApp: an interactive web application for processing genome-wide pooled CRISPR-Cas9 viability screen”. *bioRxiv*.
- Vinceti, A., Perron, U., L. Trastulla, and F. Iorio (2022). “Reduced gene templates for supervised analysis of scale-limited CRISPR-Cas9 fitness screens”. *bioRxiv*.
- Kalkauskas, A., Perron, U., Y. Sun, N. Goldman, G. Baele, S. Guindon, and N. De Maio (2021). “Sampling bias and model choice in continuous phylogeography: Getting lost on a random walk”. *PLoS computational biology* 17.1, e1008561.
- Najgebauer, H., Perron, U., and F. Iorio (2021). “Redefining false discoveries in cancer data analyses”. *Nature Computational Science* 1.1, 22–23.
- Vinceti, A., E. Karakoc, C. Pacini, Perron, U., R. R. De Lucia, M. J. Garnett, and F. Iorio (2021). “CoRe: a robustly benchmarked R package for identifying core-fitness genes in genome-wide pooled CRISPR-Cas9 screens”. *BMC genomics* 22.1, 1–16.
- Perron, U., A. M. Kozlov, A. Stamatakis, N. Goldman, and I. H. Moal (2019). “Modeling Structural Constraints on Protein Evolution via Side-Chain Conformational States”. *Mol. Biol. Evol.* 36.9, 2086–2103.
- Perron, U., P. Provero, and I. Molineris (2017). “In silico prediction of lncRNA function using tissue specific and evolutionary conserved expression”. *BMC Bioinformatics* 18.Suppl 5, 144.

### In preparation

- Perron, U., A. Chatzipli, E. Grassi, M. Viviani, E. Karakoc, E. Zanella, C. Isella, I. Molineris, H. Klett, P. Penk, J. Schueler, A. Bertotti, L. Trusolino, N. Conte, U. McDermott, and F. Iorio (-). “Integrative ensemble modelling of Cetuximab sensitivity in Colorectal Cancer PDXs”.
- Perron, U., R. R. De Lucia, and F. Iorio (-). “A cancer dependency knowledge graph”.