# Alessia Visconti, PhD

#### Assistant professor (tenure track)

Center for Biostatistics, Epidemiology, and Public Health Department of Clinical and Biological Sciences, University of Turin Regione Gonzole 10, 10043, Orbassano, Torino, Italy

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## Research Interests

- BIOSTATISTICS & BIOINFORMATICS
- Computational Biology & Medicine
- Data Mining & Machine Learning
- Multi-omics data analysis and integration
- Research Software Engineering

#### Brief Description

Alessia Visconti is an expert in medical statistics and computational medicine. Her research activity deals with the development and application of statistical and computational methods to identify multi-omics modifications influencing complex human phenotypes. She has also worked on the problem of knowledge discovery in biological data, developing new approaches tailored to solve biological tasks, and on the reverse engineering of gene regulatory networks.

## EDUCATION

Jul 2012 PhD in Computer Science, University of Turin.

Thesis title: Systems Biology: Knowledge Discovery and Reverse Engineering

Jul 2008 Master degree in Computer Science "summa cum laude", University of Turin.

Thesis title: SPOT: an algorithm for the extraction and the analysis of biological

patterns

MAR 2006 Bachelor degree in Computer Science "summa cum laude", University of Turin.

Thesis title: The Haskell language

## SKILLS

LANGUAGE SKILLS ITALIAN: native proficiency

ENGLISH: professional working proficiency Greek (Modern): elementary proficiency

Computing Skills Programming Languages: bash, C, C++, JAVA, php, python, R, ruby

OTHER LANGUAGES: CSS, LATEX, HTML, PyQt, XML

STATISTICAL SOFTWARE: R, SAS

DATABASE MANAGEMENT: MySQL, MariaDB

VERSION CONTROL SYSTEMS & REPRODUCIBILITY: GIT, nextflow, docker,

singularity

BIOINFORMATICS & GENETIC ANALYSIS: BBmap, BEDTools, DESeq2, GCTA, GWAMA, LDAK, limma, lmekin, metal, Merlin, PLINK, QTDT,

samtools, vcftools, ...

STRUCTURAL EQUATION MODELLING: openMX, Mplus

DATA VISUALISATION: dot, ggplot2

Dec 2023 - Present

Assistant professor (tenure track) at the Center for Biostatistics, Epidemiology and Public Health, Department of Clinical and Biological Sciences, University of Turin, Italy

RESEARCH ACTIVITY: Alessia Visconti is responsible for (a) the development and application of statistical and computational multi-omics approaches to understand the mechanisms and to improve the diagnosis and treatment of human diseases, (b) the design and deployment of Deep Learning approaches for monitoring and improving public health responses [P6], (c) the teaching of under- and post-graduates courses, and (d) the preparation of research grants, of which she has also the scientific responsibility. She has also an interest in the integration of patients' preferences in medical care and drug development [PP3].

Alessia Visconti remains lead the microbiome data analysis at the Department of Twin Research & Genetic Epidemiology, King's College London, UK, where she explores the effect of the gut microbiome on human health [J53, J54, J56, J58], with a particular interest on its phage component [J52, PP1], and continues her research on IgA glycosylation [J57] and in the field of genetic epidemiology [J55, PP4].

Oct 2023 - Nov 2023

**Senior bioinformatician** at the Genomics Research Centre, Human Technopole, Italy

RESEARCH ACTIVITY: Alessia Visconti was involved in a project performing genome-wide association study (GWAS) on phenotypes acquired from cardiac MRIs *via* an unsupervised deep-learning approach [PP2].

Aug 2022 - Apr 2023

Career break: Maternity leave

Aug 2017 - Sep 2023

Research fellow at the Department of Twin Research & Genetic Epidemiology, King's College London, UK

RESEARCH ACTIVITY: Alessia Visconti was responsible for (a) the development and application of statistical and computational multi-omics approaches to understand the mechanisms and to improve the diagnosis and treatment of human diseases, (b) the supervision of master and PhD students as well as postdocs, (c) the preparation of research grants, of which she has also the scientific responsibility

From her previous role, Alessia Visconti continued to lead the bioinformatics analyses for a set of studies aiming at dissecting the aetiology of melanoma and its risk phenotypes [J21, J24, J25, J29] and at studying IgA Nephropathy [J38]. She was also responsible for several new projects aiming at investigating, among the others, the influence of the gut microbiome on human health [J22, J31, J41, J44, J45, J48, J49, J50], and predicting melanoma response to immunotherapy [J43, J46], and at studying thyroid diseases [J23], atopic dermatitis [J39, J51], cardiovascular diseases and their risk factors [J35], immune system modifications [J36], and X-inactivation [J47]. During the SARS-CoV-2 pandemic, she (a) developed the analysis pipeline for the daily data provided by more than four million users [J40], (b) led two studies investigating skin manifestations of SARS-CoV-2 [J33, J42], and (c) performed the bioinformatics analysis for several other studies [J26, J28, J30, J32, J34, J37].

Jun 2016 - Jun 2019

Honorary research associate at CERN OpenLab, Switzerland

RESEARCH ACTIVITY: Alessia Visconti extended the ROOT library (https://root.cern/), developed by the CERN OpenLab for enabling the storage and scientific analyses and visualisation of large amounts of data from particle physics experiments, to allow the efficient storage of genomic data.

APR 2015 - JUL 2017

Research associate at the Department of Twin Research & Genetic Epidemiology, King's College London, UK

RESEARCH ACTIVITY: Alessia Visconti was mostly responsible for the development and application of statistical and computational multi-omics approaches to understand the mechanisms and to improve the diagnosis and treatment of human diseases. She also supervised master and PhD students and contributed to the writing of research grants. In this role, Alessia Visconti led the bioinformatics analyses for a set of projects aiming at dissecting the aetiology of melanoma, melanoma risk phenotypes, and their connection with ageing [J11, J15, J16, J19], and at studying IgA Nephropathy [J10], cognition and neurodevelopmental disease [J20], and epigenetic modification [J14]. She also reported on how to conduct metagenomic studies in microbiology and clinical research [J18] and developed a novel pipeline which ensures reproducibility in metagenomics research [J17].

Jan 2014 - Mar 2015

**Research associate** at the Department of Genomics of Common Disease, School of Public Health, Imperial College London, UK

RESEARCH ACTIVITY: Alessia Visconti (a) developed and implemented a novel approach for the population and pedigree association testing for quantitative data [J13], and (b) conducted the bioinformatics analysis for [J7, J8, J9, J12].

Jan 2012 - Dec 2013

**Research associate** at the Department of Computer Science, University of Turin, Italy

RESEARCH ACTIVITY: Alessia Visconti (a) developed and implemented a novel bi-clustering approach leveraging additional knowledge [J5], (b) contributed to the development of a novel exact algorithm for answering Maximum a Posteriori queries on tree structures [P5], (c) applied machine learning approaches for the prediction and interpretation of the lipophilicity of small peptides [J6], and (d) conducted the bioinformatics analysis to measure the ability of more than 200 compounds of acting as hydrogen bond donors [J4].

Jun 2011 - Dec 2011

**Visiting researcher** at the Center of Biological Sequence Analysis, Technical University of Denmark, Denmark

RESEARCH ACTIVITY: Alessia Visconti (a) continued her PhD project developing a new method for the reverse engineering of gene regulatory networks that uses a popular econometrics statistical hypothesis test, namely the Granger Causality [T3], and (b) developed an *ensemble* approach for the prediction of promoter activity.

Jan 2009 - Dec 2011

PhD student at Department of Computer Science, University of Turin, Italy RESEARCH ACTIVITY: Alessia Visconti developed and implemented: (a) a de novo framework (accompanied by a web interface) performing protein motifs identification and allowing the simultaneous associations between groups of protein sequences and groups of motifs thanks to a constrained co-clustering approach [P1], (b) a new methodology (accompanied by a web interface) that provides meaningful co-clusters whose discovery and interpretation are enhanced by embedding gene ontology (GO) annotations [BC1, P2], (c) a novel algorithm for the rewriting of the GO aiming at obtaining a more compact and informative ontology [J2, P3], (d) two algorithms for the reverse engineering of gene regulatory networks [P4, J3, T3], and (e) contributed to a modular framework for the analysis of metagenomics sequences leading the co-clustering module development [J1].

Sep 2008 - Dec 2008

Research assistant at the Department of Computer Science, University of Turin and in collaboration with the Department of Arboriculture and Pomology, University of Turin, Italy

RESEARCH ACTIVITY: During this post, Alessia Visconti contributed to the development of computational approaches for the classification and traceability of fruits produced in Piemonte.

## Awards

Summer 2019 Awarded a mini-grant (£1000) to hire an undergraduate student through the "King's Undergraduate Research Fellowship (KURF)".

Summer 2018 Awarded a mini-grant (£1000) to hire an undergraduate student through the "King's Undergraduate Research Fellowship (KURF)".

Winner of one of the challenges of the "BioDataHack 2018 – Genomic, Biodata and Improving Health Outcomes". The project presented by Alessia Visconti and the other members of her team (Jun Aruga, Oliver Giles, Ioannis Valasakis e Chen Zhang) advanced the vision of a device that will allow the constant monitoring of IBD by patients from the comfort of their own homes, ranked first on the ARM, Cavium, and Atos Challenge: How can we use mobile technology to transform biological data processing?. During the two-day BioData Hackathon, the team successfully ported the metagenomics pipeline developed by Alessia Visconti [J17] onto Arm's 64-bit architecture, where the data could be processed in a few hours, showing that the analysis of microbial data can be successfully taken out of centralised data centres.

The solution also implemented a neural network that, receiving as input the microbial profile produced by the analysis pipeline, could predict the disease status. Even with a very small set of training data, the proposed approach was able to separate patients with Crohn's disease and ulcerative colitis (the two main forms of IBD) from healthy controls with over 90% accuracy.

The team also proposed a derivative score from the neural network's output. Such a score, which could be tracked over time, would allow individuals to measure the effects of lifestyle/medication changes on their disease's progression to assess their efficacy in almost real-time. The next step would be to use the data generated in this monitoring process to craft suggestive models, able to offer individuals advice and treatments based on what has been effective in patients with similar microbiome profiles.

Summer 2017 Awarded a mini-grant (£1000) to hire an undergraduate student through the "King's Undergraduate Research Fellowship (KURF)".

Jan 2012 - Dec 2013 **Awarded two years postdoctoral fellowship** awarded by the Italian Minister of Education, University and Research.

Jan 2012 - Dec 2012 Awarded a postdoctoral training grant (€3000) awarded by the Regione Piemonte.

Summer 2011 Participation to the "DREAM6 – Promoter Activity Prediction Challenge". The proposed approach (developed with Ali Altıntas and Chris Workman) which combined results from two well-known machine learning approaches (regression trees and support vector machines for regression), ranked 8th out of 21 participants.

Summer of one of the challenges of the "DREAM5 – Network Inference Challenge". The ensemble approach [P4, J3] developed with the other team members (Roberto Esposito and Francesca Cordero), which was compared with other 35 approaches and which combines multiples approaches within a Naive Bayes classifier, despite its simplicity, ranked third for the reverse engineering of real organisms' gene regulatory networks and first for the reconstruction of the Saccharomyces Cerevisiae's network.

Jan 2009 - Dec 2011 Awarded three years PhD fellowship awarded by the Italian Minister of Education, University and Research (best PhD project).

# RESEARCH COLLABORATIONS

Only collaborators outside King's College London (for projects started between 2014 and 2023) and the University of Turin (for projects started before 2014 and after 2024) are reported.

#### Projects for which Alessia Visconti has scientific responsibility

2025 Title: "Read Assessment and Decision Support for ICU Readmission Prediction (READ-ICU)"

BUDGET: €43,200 - FUNDER: Horizon Enfield

Collaborators: National Technical University, Athens, Greece

OBJECTIVES: Developing a deep-learning ensemble predictive model to evaluate the risk of ICU readmission using MIMIC-IV and eICU datasets.

ROLE: co-PI. Within this project, she is co-supervising a PhD student (Emanuele Koumantakis).

2024-2025 Title: "Exploring the potential role of gut microbiota in improving outcomes of chronic myeloid leukaemia patients"

BUDGET: €80,000 - FUNDER: ESH John Goldman Research Award

Collaborators: Italian Institute for Genomic Medicine, Turin, Italy

OBJECTIVES: Understanding the cross-talk between gut microbiota and immune response and its role in treatment tolerance and success in patients with chronic myeloid leukaemia. Role: Responsible of the work-packages dealing with all the bioinformatics analyses.

2022-2025 Title: "A Collaborative Approach to the Borne Uterine Mapping Programme (BUMP) Feasibility Study"

Funder: BORNE - Budget: £500,000

COLLABORATORS: Imperial College London, London, UK, Newcastle University, Newcastle upon Tyne, UK, and University of Turin, Turin, Italy

OBJECTIVES: Studying normal term labour and map the uterus using state-of-the-art transcriptomics (bulk and single cells/nuclei), proteomics (bulk and single-proteomics), and spatial sequencing.

ROLE: Responsible for the work-packages aiming at the development of (a) pipelines for the analysis of single-cell and single-nucleus RNA sequencing and spatial transcriptomic data, and (b) deep-learning approaches for modelling their interaction.

2021-2024 Title: "Understanding phenotype and mechanisms of spontaneous preterm birth in sub-Saharan Africa (PRECISE-SPTB)"

Funder: MRC Medical Research Council - Budget: £458,80

COLLABORATORS: Aga Khan University, Nairobi, Kenya, MRC Unit The Gambia at LSHTM, Banjul, The Gambia, and Eduardo Mondlane University, Maputo, Mozambique OBJECTIVES: Determining the epidemiological and contextual nature of spontaneous preterm birth in three sub–Saharan African countries (Kenya, Nairobi and Mozambique), while developing technical infrastructure and training research scientists.

Role: Responsible for the preparation and delivery of workshops covering both basic and specialistic skills, namely: shell programming, version control and collaboration with Git/GitHub, programming in R, programming in python, workflow development with Nextflow, machine-learning approaches for biomedical data analysis, and metagenomic data analysis. Workshops have been rated as "exceptional" by the attendees.

2020-2021 Title: "A multi-omics study to dissect the role of the gut microbiome in IgA nephropathy risk"

Funder: King's College London - Peking University Health Science Centre Joint Institute for Medical Research — Budget: £74,000

Collaborators: Peking University, Peking, China

OBJECTIVES: Identify microbes and microbial functions associated with IgAN, and investigating their role in the disease by studying their interplay with blood glycomics and faecal metabolomics.

ROLE: Responsible for the work package dealing with *in silico* characterisation and validation, using bioinformatics models, of microbes associated with IgA nephropathy and/or IgA glycosylation profiles.

2016-2018 Title: "A high-resolution map of copy number and structural variation in Qatari genomes and their contribution to quantitative traits and disease"

Funder: Qatar Foundation - Budget: £160,521

Collaborators: Sidra Medical and Research Centre, Doha, Qatar

OBJECTIVES: Generating a database of structural variants in Qataris and leveraging the deep phenotyping on these samples to estimate, among the others, the contribution of SVs to CVD and related cardio-metabolic traits.

ROLE: Responsible for work-packages aiming at the development of (a) an approach for the storage of genomic data taking advantage of the ROOT library, and (b) an ensemble approach for the identification of structural variation. She also conducted the bioinformatics analysis for [J35, J55] and supervised a postdoctoral researcher (Niccoò Rossi).

## Projects to which Alessia Visconti participates as researcher

Alessia Visconti has a prominent role in all projects, as shown by the number of publications in which she is first/last author.

2023-2027 Title: "PUZZLE: aPproccio integrato alla mUltimorbidità: ricerca di base, traslaZionale, clinica e formaZione muLtidisciplinarE"

OBJECTIVES: Developing a novel model for the study, management, and care of patients with multi-morbidity.

ROLE: Leading researcher in the application of Deep Learning approaches to identify multi-morbidity patterns from epidemiological data on several diseases (e.g., cardiometabolic diseases and cancer) and environmental exposures (e.g., smoking, alcohol consumption, physical activity). Within this project, she is supervising a postdoctoral researcher (Giulio Ferrero).

2022-2025 Title: "TrustAlert"

COLLABORATORS: Bruno Kessler Foundation, Trento, Italy, Links Foundation, Turin, Italy, Innovo Group, Turin, Italy, Local Health Authority Alba-Bra, Alba, Italy, Cottolengo Hospital, Turin, Italy

OBJECTIVES: Developing AI-based solutions for (a) real-time identification of health emergencies, (b) mapping vulnerable and multi-morbid populations, and (c) crate a *living lab* for micro and macro simulations.

ROLE: Co-leading researcher in the application of Deep Learning approaches for (a) the extraction reliable clinical information from structured and unstructured data for supporting medical decision [P6], and (b) the mapping of morbidity and medical vulnerabilities patterns that exist within communities. Within this project, she is supervising a mater student (Francesca Rondinone) and a research assistant (Emanuele Pietropaolo).

2022-2025 Title: "Repertor.IO: Incorporating Patient Preference Studies into Clinical Research and Decision Model"

Collaborators: University of Padua, Padua, Italy

OBJECTIVES: Developing a platform for the collection, annotation, and analysis of patients preference studies.

Role: Contributing to the data analysis [PP3].

2021-2022 Title: "Predicting Response to Immunotherapy for Melanoma with gut Microbiome and metabolomics - The PRIMM Study"

COLLABORATORS: University Medical Center, Groningen, The Netherland and University of Trento, Trento, Italy

OBJECTIVES: Predicting which patients with melanoma would benefit from the use of immune checkpoint inhibitors using multi-omics data.

ROLE: Leading researcher for the bioinformatics analysis for the identification of glycomarkers [J46] (first author) and collaborated to the analysis of proteomic data [J43] aiming at finding novel biomarkers of response and survival to identify those patients with melanoma who are most likely to benefit from immune checkpoint inhibitors. Within this project, she helped with the supervision of a PhD student (Karla Lee).

2019-2021 Title: "Dissecting the mechanisms of immune-mediated inflammation: a systems-immunology approach"

OBJECTIVES: Identifying targetable networks of circulating immune cells and cytokines involved in the normal immune-mediated inflammation process, and identify those disrupted in immune-mediated inflammatory diseases.

ROLE: Leading researcher for the bioinformatics analyses aiming at (a) the reverse engineering of immune cell co-expression networks and their involvement in a set of autoimmune diseases, and (b) the identification of genetic variations and microbes/metabolites responsible for the development of such diseases. Within this project, she supervised a postdoctoral researcher (Niccoò Rossi).

2018-2024 TITLE: "TREatment of ATopic eczema (TREAT) Registry Taskforce"

COLLABORATORS: more than 30 centers over 13 countries (see the registry website for details treat-registry-taskforce.org)

OBJECTIVES: studying the genetic and environmental basis of atopic dermatitis (consortium).

Role: Leading bioinformatician for the TwinsUK cohort [J39, J51, PP4].

2016-2018 Title: "Gut microbiome modulation of fasting glucose homeostasis and postprandial glycaemic response in TwinsUK and PREDICT: towards personalised diet for healthy aging"

Collaborators: University of Trento, Trento, Italy

Objectives: studying the influence of the gut microbiome on cardiometabolic health.

Role: Alessia Visconti (a) developed a tool for the analysis of metagenomic data which ensures the reproducibility of the scientific results [J17] (first author), (b) performed the bioinformatics analysis of metagenomics and metabolomics data [J22, J31] (co-first author in the first study), (c) collaborated to further studies [J48, J49, J50], and (d) supervised a PhD student for the work described in [J44] (co-senior author).

2014-2016 Title: "An integrative genomics approach for non-invasive diagnostic biomarkers discovery in IgA nephropathy"

Collaborators: Imperial College London, London, UK

OBJECTIVES: Identify glycomics and genetic biomarkers for helping IgAN diagnosis and treatment

ROLE: Alessia Visconti applied statistical and bioinformatics approaches for studying the role of IgA and its glycosylation profiles in the development of IgA nephropathy, as described in [J10, J38, J57] (co-first author in all).

2013-2015 Title: "Senescence and melanoma – An integrative systems biology approach to characterise the link between reduced biological senescence and melanoma susceptibility"

COLLABORATORS: Hospital Clinic of Barcelona, Barcelona, Spain, QIMR Berghofer Medical Research Institute, Brisbane, Australia, Beijing Institute of Genomics, Beijing, China, Institute of Cancer and Pathology, Leeds, UK, Erasmus MC University Medical Center, Rotterdam, The Netherland, Women's Hospital, Boston, US.

OBJECTIVES: Identifying the genetics basis of melanoma skin cancer and of its related phenotypes.

ROLE: Alessia Visconti applied statistical and bioinformatics approaches for studying melanoma, melanoma risk phenotypes, and their connection with ageing, as described in [J12, J11, J15, J16, J19, J21, J24, J29, J25] (first author in three manuscripts).

2013-2018 Title: "Genomic analysis of Type 2 Diabetes in Qatar, towards diabetes personalized medicine"

Collaborators: Weill Cornell Medicine-Qatar, Doha, Qatar

Objectives: Identifying the genetics and epigenetics basis of Type 2 Diabetes

Role: Alessia Visconti (a) developed and implemented an approach for the population and pedigree association testing for quantitative data [J13] (first author), and (b) conducted the bioinformatics analysis for [J8] and [J14].

2012-2013 Title: "LIMPET – Isotropic And Anisotropic Lipophilicity To Model Permeability Of New Therapeutic Peptides"

OBJECTIVES: Applying machine Learning approaches to predicts peptides permeability on the basis of experimental non-conventional lipophilicity indexes.

ROLE: Alessia Visconti (a) evaluated the ability of some combinations of descriptors/algorithms to find the best model to predict the lipophilicity of small peptides [J6] (first author), and (b) performed the bioinformatics analyses to measure the ability of more than 200 compounds of acting as hydrogen bond donors [J4].

2007-2011 Title: "BioBITs – Developing white and green biotechnologies by converging platforms from biology and information technology towards metagenomics"

Collaborators: University of Eastern Piedmont, Alessandria, Italy

OBJECTIVES: Identifying and characterising populations of uncultivable bacteria living inside a symbiotic, arbuscular mycorrhizal fungus.

ROLE: Alessia Visconti contributed to the development of a modular framework for the analysis of metagenomics sequences, and was responsible for the co-clustering module [J1].

## Projects with industrial partners to which Alessia Visconti participates

2020 - 2022 PARTNER: ZOE Ltd https://health-study.joinzoe.com/)

ROLE: Alessia Visconti (a) developed the analysis pipeline for the daily data provided by more than four million users [J40], (b) led two studies investigating skin manifestations of SARS-CoV-2 [J33, J42], and (c) performed the bioinformatics analysis for several other studies [J26, J28, J30, J32, J34, J37].

- 2019 2020 PARTNER: Sanofi (https://www.sanofi.com) ("Sanofi iAwards Europe 2019")
  ROLE: Alessia Visconti carried out part of the bioinformatics analyses, and supervised a postdoctoral researcher (Niccolò Rossi).
- 2018 2020 PARTNER: Danone Nutricia Research (https://www.nutriciaresearch.com)
  ROLE: Alessia Visconti performed the metagenomic data analyses, as described in [J41].

# REVIEW ACTIVITY

# EDITORIAL BOARD MEMBERSHIP

Since September 2019, Alessia Visconti is *Review Editor* of Human and Medical Genomics (speciality section of Frontiers in Genetics).

Reviewer

Alessia Visconti serves as a reviewer for the following international journals: Bioinformatics, BioData Mining, BMC Cancer, BMC Nutrition, BMC Supplements, British Journal of Cancer, Computational and Structural Biotechnology Journal, Communications Biology, European Journal of Human Genetics, Frontiers in Cellular and Infection Microbiology, GigaScience, Gut Microbes, Journal of the European Academy of Dermatology and Venereology, Knowledge and Information Systems (KAIS), Machine Learning, PLoS Computational Biology, PLoS One, Nature Communications, Scientific Reports

## INVITED TALKS

17 Jun 2019	"Reproducible shotgun metagenomic analysis with Nextflow and containers" at the 1st London Bioinformatics Frontiers Frontiers conference, London, UK
23 Nov 2018	"Nextflow on the go" at the 2nd Nextflow workshop, Barcelona, Spain
6 Dec 2017	"YAMP: a framework enabling reproducibility in metagenomics research" at the Computational Biology seminar series, Francis Crick Institute, London, UK
13 Dec 2017	"My reproducible adventure in (meta)genomics (A tale of workflow development, research reproducibility & open science)" at the Researc/hers code seminar series, London, UK
15 Sep 2017	"Simplifying shotgun metagenomics analysis with Nextflow" at the 1st Nextflow workshop, Barcelona, Spain
7 Jul 2012	"Knowledge-driven Co-clustering of Gene Expression Data" at the Center for Biological Sequence Analysis, Technical University of Denmark, Lyngby, Denmark
8 May 2009	"Using a priori knowledge for the reverse engineering of gene regulatory networks" at the Computer Laboratory, Cambridge University, Cambridge, UK

# DEVELOPMENT OF RESEARCH SOFTWARE

The software is available at https://github.com/alesssia or upon request.

AID-ISA	extracts biologically relevant biclusters from microarray gene expression data by leverag-
	ing additional knowledge (described in [J5])
CDoT	is a novel exact algorithm for answering Maximum a Posteriori queries on tree-structured
	Probabilistic Graphical Models (described in [P5] and developed in collaboration with
	Roberto Esposito)
FAMCNV (v2.0)	enables genome-wide association of copy number variants with quantitative phenotypes
	in families (developed in collaboration with Mario Falchi)
GOCLUST	performs a co-clustering of microarray gene expression data using Gene Ontology-derived
GOCLOSI	constraints (described in [P2])
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MotifsLinker	associates clusters of proteins with their frequent motifs
PicNic	extracts topologies and discovers patterns in sets of chemical compound
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PopPAnTe	enables pairwise association testing in related samples (described in [J13])
RGO	is a reorganization of the Gene Ontology emphasising regulative information and provid-

ing better structure for gene functional analysis (described in [J2])

SPOT performs an exhaustive search of frequent motifs in sets of biological sequences (described in [T2])

YAMP allows processing raw metagenomic sequencing data up to the functional annotation

(described in [J17])

## SERVICE ACTIVITY

Jan 2025 – Present	Co-coordinator of the Working Group in Machine Learning in Clinical Research of the Italian Society of Medical Statistics and Clinical Epidemiology (SISMEC, https://www.sismec.info/sismec/gruppi-di-lavoro/gruppi-di-lavoro-sismec/machine-learning-nella-ricerca-clinica/)							
Oct 2024 – Present	Member of the Scientific Board for the Medical Residency program in Med-							
	ical Statistics and Biometrics, at the University of Turin							
Jun $2024$ – Present	Member of the Scientific Board for the PhD program in Complex Systems for							
	Quantitative Biomedicine, at the University of Turin (phd-csqb.campusnet.unito.it)							
Dec 2023 – Present	DISC (Department Internal Staff Contact) for the platform MedCapu (www.							
220 2020 110202111	medcap.unito.it) at the University of Turin							
DEC 2019 - SEP 2023 Co-organiser of a series of workshops aiming at teaching basic comp								
	literacy and data analysis skills with the KCL Carpentries, King's College							
	London (kcl-carpentries.github.io)							
Nov $2016 - Jul 2022$	Co-organiser of the Regulatory Genomics journal club at the Department of							
	Twins Research & Genetic Epidemiology, King's College London.							
Jan 2012 - Dec 2013	Faculty member as representative of postdoctoral fellows at the Department							
	of Computer Science, University of Turin							
Jan 2009 – Dec 2011 Faculty member as representative of PhD students at the De								
J 2000 BEO <b>2</b> 011	Computer Science, University of Turin							

## Outreach activity

Jul 2017 - Feb 2023

Member and mentor of the Artificial Intelligence Club for Gender Minorities, which aims at promoting gender diversity in the artificial intelligence and scientific community via meetups, and mentorship. Alessia Visconti organised workshops on collaborative data science via Git and GitHub. Since May 2018, she is also co-organising the group monthly journal club

Mar 2016 - Sep 2017

Member, tutor, and mentor of the *RLadies London* community and of *Researc[her] Research* community. These groups aim at promoting gender diversity in the R and STEM community via meetups, mentorship and global collaboration. With *Researc[her]*, Alessia Visconti gave speeches on reproducibility, and workflow development

## TEACHING ACTIVITY

## In English

#### To PhD students, postdoctoral researchers, and PIs

2025 - PRESENT

Lecturer for the Collaborative tools for reproducible research workshop for the PhD program in Complex Systems for Quantitative Biomedicine, at the University of Turin. The workshop (8h) is centred around the idea of "live coding", where attendees code along with the instructors thus getting useful hands-on experience and improving their ability to explore the topics on their own.

**Lecturer** for the *Machine Learning in clinical epidemiology and pre-clinical research*) workshop for the PhD program in Complex Systems for Quantitative Biomedicine, at the University of Turin. The workshop includes frontal lessons (12h) and a hands-on session (8h, lead by Alessia Visconti).

2019 – 2023 Instructor for several programming and data analysis workshops:

- The Unix Shell
- Version Control and collaboration with  $\mathrm{Git}/\mathrm{GitHub}$
- Python Programming

- R Programming
- Introduction to Working with Data
- OpenRefine

These workshops are designed for PhD students and early career researchers but are open to researchers at every level, including PIs, are offered regularly (roughly twice a year), and have always been evaluated as "excellent" or "exceptional" by the attendees (details on some of the workshops can be found at <a href="https://kcl-carpentries.github.io/">https://kcl-carpentries.github.io/</a>). The workshops are centred around the idea of "live coding", where attendees code along with the instructors thus getting useful hands-on experience and improving their ability to explore the topics on their own.

- 2016 2023 **Co-organiser** of the Regulatory Genomics journal club at the Department of Twins Research & Genetic Epidemiology, King's College London. The journal club discusses papers on the latest achievements and methods in the fields of regulation of gene expression, epigenetics, splicing, evolution, and related topics.
- 2021 2022 Instructor for The Unix Shell and the Version Control and collaboration with Git/GitHub workshops for students and researchers of the MRC Gambia and the AKU Nairobi Research Centres. Both workshops were rated as "exceptional" by the attendees.

  Instructor for the Metagenomics Data Analysis: Investigating the invisible world of microbes (details at https://alesssia.github.io/metagenomic\_workshop/). The workshop, which included frontal lessons and hands-on sessions, was designed for researchers at every level (including but not limited to PhD students, post-doctoral researchers and PIs) without any previous knowledge of the topics and tools presented.

#### To graduate students

2025 - PRESENT

Co-lecturer for the Leveraging Large Language Models for hands-on learning in medical education and research elective teaching activities for the Faculty of Medicine and Surgery - MedInTO, Department of Clinical and Biological Science, University of Turin. The workshop (10h) is organised as a set of lectures, hands-on activity, and discussions and explains what Large Language Models are, how they work, and how to use them in a critical way.

#### To postgraduate students

- 2013 2014 **Teaching assistant** for the "Human Molecular Genetics" MSc Department of Genomics of Common Diseases, Imperial College London. Alessia Visconti was offering support during the practical sessions on R programming as well as one-to-one meetings with students attending the following courses:
  - The Unix Shell
  - R Programming
  - Exploratory Data Analysis and Probability
  - Quantitative genetics
  - Next Generation Sequencing Data Analysis.

#### To medical residents

2025 - PRESENT

**Co-lecturer** for the *Integrating Large Language Models into clinical work and research:* a critical approach workshop for medical residents at University of Turin, University of Turin. The workshop (14h) is organised as a set of lectures, hands-on activity, and discussions and explains what Large Language Models are, how they work, and how to use them in a critical way in the clinic and in clinical reasearch.

#### Outside academia

2021 - 2022 Instructor for a Version Control and collaboration with Git/GitHub workshop at the UK Health Security Agency (UKHSA)

#### In Italian

#### To undergraduate students

2024 - PRESENT **Lecturer** for the "Medical statistics" module within the "Evidence-based nursing" course, Nursing School, Department of Clinical and Biological Science, University of Turin. Alessia Visconti was the sole responsible for the module, which covers the basics of biostatistics. The module is run in 2 parallel sessions of 15h each.

**Lecturer** for the "Medical statistics" module within the "Preparatory to research" course, Degree in Psychiatric Rehabilitation Techniques, Department of Clinical and Biological Science, University of Turin. Alessia Visconti was the sole responsible for the module, which covers the basics biostatistics and the basics of R programming and data analysis (24h).

Co-lecturer for the "Methodology of social-health research" course, Degree in Professional Education, University of Turin. Alessia Visconti was the responsible for the part of the course covering the basics biostatistics (20h).

2013 - 2014 **Teaching assistant** for the "Data analysis" course, Department of Biological Science, University of Turin. Alessia Visconti was offering support during the practical sessions on R programming as well as one-to-one meetings with students.

**Teaching assistant** for the "Operating System" course, Department of Computer Science, University of Turin. Alessia Visconti was offering support during the practical sessions on R programming as well as one-to-one meetings with students.

- 2012 2013 Lecturer for the "Operating System and Networking" course, Interfaculty School of Strategic Studies, University of Turin. Alessia Visconti was the sole responsible for the practical sessions covering the basis of GNU/Linux, the Unix shell, and process management. She designed the final project, i.e., the development of a basic client/server application in C (details at https://alesssia.github.io/sistemi\_elab\_info\_I\_2012\_13)

  Teaching assistant for the "Operating System" course, Department of Computer Science, University of Turin. Alessia Visconti was offering support during the practical sessions as well as one-to-one meetings with students.
- 2011 2012 **Teaching assistant** for the "Database" course, Department of Computer Science, University of Turin. Alessia Visconti was offering support during the practical sessions as well as one-to-one meetings with students.
- 2010 2011 **Teaching assistant** for the "Database" course, Department of Computer Science, University of Turin. Alessia Visconti was offering support during the practical sessions as well as one-to-one meetings with students.

**Teaching assistant** for the "Formal Language" course, Department of Computer Science, University of Turin. Alessia Visconti was offering support during the practical sessions as well as one-to-one meetings with students.

**Teaching assistant** for the "Statistics and data mining with SAS" course, Department of Mathematics, University of Turin. Alessia Visconti prepared slides and recorded 3 hours of video lessons on SAS Enterprise Miner. She also prepared a self-evaluation questionnaire for the students.

2009 - 2010 Lecturer for the "Computer Science" course, Department of Letters and Philosophy, University of Turin. Alessia Visconti was the sole responsible for the practical sessions covering the MS Office suite and the students' evaluation (details at https://alesssia.github.io/lab\_lettere\_2009\_10/).

- 2006 2007 **Teaching assistant** for the "Program Languages JAVA" course, Department of Computer Science, University of Turin. Alessia Visconti was offering support during the practical sessions.
- 2005 2006 **Teaching assistant** for the "Program Languages JAVA" course, Department of Computer Science, University of Turin. Alessia Visconti was offering support during the practical sessions.
- 2004 2005 **Teaching assistant** for the "Program Languages C" course, Department of Computer Science, University of Turin. Alessia Visconti was offering support during the practical sessions.

#### To medical residents

2023 - PRESENT **Lecturer** for *Introduction to Statistics* short course for medical residents at University of Turin (9h). Alessia Visconti was the sole responsible for the module, which covers the basics of biostatistics.

# SUPERVISION ACTIVITY

Thanks to her multidisciplinary experience, which combines a BSc, MSc, and PhD in computer science with more than 10 years of research activity in the biomedical field, Alessia Visconti has been able to supervise students with different backgrounds (e.g., computer science, bioinformatics, molecular biology, medicine, neurobiology, and engineering) offering a stimulating and engaging environment.

#### BSc & MSc Theses

2025 - Present	Co-supervise	or of Mr Bori	s Turcan	ı's MSc projec	$\operatorname{ct}$ in $Computer$	Science at th	e University
	of Turin, Italy	·.					
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**Co-supervisor** of Mr Samuele Perrotta's MSc project in *Computer Science* at the University of Turin, Italy.

- 2024 2025 **Co-supervisor** of Francesca Rondinone's MSc project in *Stochastics and Data Science* at the University of Turin, Italy.
- 2021 2022 **Co-supervisor** of Ms Darvina Magandran's MSc project in *Microbiome in Health and Disease* at King's College London.
- 2020 2021 **Co-supervisor** of Ms Raphaela Joos's MSc project in *Microbiome in Health and Disease* at King's College London.

**Co-supervisor** of Ms Petra Blackburn's MSc project in *Microbiome in Health and Disease* at King's College London.

Co-supervisor of Ms Natalie Falshaw's MSc project in *Microbiome in Health and Disease* at King's College London.

- 2019 2020 **Co-supervisor** of Ms Xinyu Huang's MSc project in Pharmacology at King's College London.
- 2018 2019 **Co-supervisor** of Ms Miriam Margari's MSc project in Genomic Medicine at Imperial College London.
- 2017 2018 **Co-supervisor** of Dr Robin Mesnage's MSc project in Bioinformatics at Birkbeck University of London
- 2013 2014 **Co-supervisor** of Mr George Powell's MSc project in Human Molecular Genetics at Imperial College London.
- 2010 2011 **Co-supervisor** of Mr Marco Gallizio's bachelor thesis in Computer Science at the University of Turin, Italy

#### Post-master students

2019 - 2020 **Co-supervisor** of Mr Simon Couvreur in his PhD rotation project at King's College London

Co-supervisor of Ms Helen King in her PhD rotation project at King's College London.

#### PhD students

- 2021 2023 Co-supervisor of Mr Roger Compte Boixader in his PhD project at King's College London.
   2019 2024 Co-supervisor (unofficial) of Ms Xinyuan Zhang in her PhD project at King's College London (Publications: [J44]).
- 2020 2022 **Assistance with the supervision** of Ms Karla Lee in her PhD project at King's College London (supervision limited at the multi-*omics* data analyses; publications: [J43, J46]).
- 2016 2019 Co-supervisor (unofficial) of Mr Niccolò Rossi during his research visit at King's College London (Publications [J35, J55]).
- 2018 2020 **Supervisor** of Ms Giulia Piaggeschi during her research visit at King's College London (Publications: [J36]).
- 2014 2020 **Co-supervisor** (unofficial) of Ms Marianna Sanna in her PhD project at King's College London and her research activity at Imperial College London (Publications: [J11, J19, J21, J24, J29]).
- 2015 2019 Assistance with the supervision of Mr Taghi Aliyev during his PhD at the CERN OpenLab (supervision limited to the biomedical part of the project).

## Postdoctoral researchers

- 2024 PRESENT Co-supervision of Dr Idrissa SY during his postdoc at the University of Turin.
  - 2019 2023 **Assistance with the supervision** of Dr Niccolò Rossi during his postdoc at King's College London (already supervised during the PhD; publications [J43]).
  - 2016 2018 Assistance with the supervision of Dr Harriet Cullen during her research fellowship at King's College London (Publications: [J20])

## Research assistants

2024 - PRESENT **Supervision** of Mr Emanuele Pietropaolo research at the University of Turin as part of the TrustAlert project (Publications: [P6]).

## Interns

- Spring/summer 2025 Supervisor of Mr Triparna Day's internship as part of her MSc in Biotechnological and Chemical Sciences in Diagnostics.
  - Summer 2019 **Supervisor** of Mr Yuhao Lin's summer project as part of the King's Undergraduate Research Fellowships (KURF)

Supervisor of Ms Olivia Castellini Pérez's summer internship as part of the Erasmus+program

- Summer 2018 Supervisor of Ms Lechun Huo's summer project as part of the King's Undergraduate Research Fellowships (KURF).
- Summer 2017 **Supervisor** of Ms Yuri Nemoto's summer project as part of the King's Undergraduate Research Fellowships (KURF).

Co-supervisor of Ms Fudi Wang's research visit at King's College London

- Summer 2016 Co-supervisor of Ms Esther Kok's summer internship at the CERN OpenLab.
- Summer 2014 Co-supervisor of Mr Marcin Świstak's internship at Imperial College London.

## **Publications**

### International Journals

- [J58] Saravi S.S.S, Pugin B., Constancias F., Shabanian K., Spalinger M., Thomas A., Le Gludic S., Shabanian T., Karsai G., Colucci M., Menni C., Attaye I., Zhang X., Allemann M.S., Lee P., Visconti A., Falchi M., Alimonti A., Ruschitzka F., Paneni F and Beer J.H., Gut microbiota-dependent increase in phenylacetic acid induces endothelial cell senescence during aging, Nature Aging, 2025, doi:10.1038/s43587-025-00864-8
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- [J56] Valdes A.M.<sup>†</sup>, Louca P.<sup>†</sup>, Visconti A., Asnicar F., Bermingham6 K., Nogal A., Wong K., Wolf J., Michelotti G., Segata N., Spector T.D., Berry S.E., Falchi M., Menni C., Vitamin A carotenoids, but not retinoids, mediate the impact of a healthy diet on gut microbial diversity, BMC Medicine, 2024, doi:10.1186/s12916-024-03543-4
- [J55] Rossi N., Syed N., **Visconti A.**, Aliyev E., Berry S., Bourbon M., Spector T.D., Hysi P.G.<sup>‡</sup>, Fakhro K.A.<sup>‡</sup>, Falchi M.<sup>‡</sup>, Rare variants at KCNJ2 are associated with LDL-cholesterol levels in a cross-population study, npj genomic medicine, 2024, doi:10.1038/s41525-024-00417-9
- [J54] Attaye I.<sup>†</sup>, Beynon-Cobb B.<sup>†</sup>, Louca P., Nogal A., **Visconti A.**, Tettamanzi F., Wong K., Michellotti G., Spector T.D., Falchi M., Bell J.T., and Menni C., *Cross-sectional analyses of metabolites across biological samples mediating dietary acid load and chronic kidney disease*, iScience, 2024, doi:10.1016/j.isci.2024.109132
- [J53] Danckert N.P.†, Freidin M.B.†, Granville Smith I., Wells P.M., Naeini M.K., Visconti A., Compte R., MacGregor A., and Williams F.M.K, Treatment response in rheumatoid arthritis is predicted by the microbiome: a large observational study in UK DMARD-naïve patients, Rheumatology, 2024, doi:10.1093/rheumatology/keae045
- [J52] Kirk D., Costeira R., Visconti A., Mirzaei M.K., Deng L., Valdes A.M., and Menni C., Bacterio-phages, gut bacteria, and microbial pathways interplay in cardiometabolic health, Cell Reports, 2024, doi:10.1016/j.celrep.2024.113728
- [J51] Budu-Aggrey A., Kilanowski A., Sobczyk M.K., Shringarpure S.S., Mitchell R., ..., Visconti A., ..., Holloway J.W., Min J., Brown S.J., Standl M., Paternoster L., European and multi-ancestry genome-wide association meta-analysis of atopic dermatitis highlights importance of systemic immune regulation, Nature Communications, 2023, 10.1038/s41467-023-41180-2
- [J50] Nogal, A.<sup>†</sup>, Tettamanzi F.<sup>†</sup>, Dong Q., Louca P., **Visconti, A.**, ..., Spector T.D., Bell J.T., Gieger C., Valdes A.M.<sup>‡</sup>, and Menni C.<sup>‡</sup>, A faecal metabolite signature of impaired fasting glucose: results from two independent population-based cohorts, Diabetes, 2023, doi:/10.2337/db23-0170
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- [J48] Louca P., Meijnikman A., Nogal MacHo A., Asnicar F., Attaye I., Vijay A., Kouraki A., Visconti A., ..., Bulsiewicz W., Nieuwdorp M., Valdes A., Menni C., The secondary bile acid isoursodeoxy-cholate is associated with postprandial lipaemia inflammation and appetite and changes post bariatric surgery, Cell Reports Medicine, 2023, doi:10.1016/j.xcrm.2023.100993
- [J47] Zito A., Roberts A.L.<sup>†</sup>, **Visconti A.**<sup>†</sup>, Rossi N., Andres-Ejarque R., Nardone S., El-Sayed Moustafa J.S., Falchi M., and Small K.S., *Escape from X-inactivation in twins exhibits intra- and inter-individual variability across tissues and is heritable*, PLoS Genetics, 2023, doi:10.1371/journal.pgen.1010556

<sup>†</sup> indicates that the authors contributed equally to the work

<sup>&</sup>lt;sup>‡</sup> means that the authors jointly supervised the work

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- [J45] Valles-Colomer M., Blanco-Míguez A., Manghi P., Asnicar F., ..., Visconti A., ..., Spector T.D., Domenici E., Collado M.C., and Segata N., The person-to-person transmission landscape of the gut and oral microbiomes, Nature, 2023, doi:10.1038/s41586-022-05620-1
- [J44] Zhang X., Adebayo A.S., Wang D., Raza Y., Tomlinson M., Dooley H., Bowyer R.C.E., Small K., Steves C.J., Spector T.D., Duncan E.L., **Visconti A.**<sup>†</sup>, and Falchi M.<sup>†</sup>, *PPI-induced changes in plasma metabolite levels influence total hip bone mineral density in a UK cohort*, Journal of Bone and Mineral Research, 2022, doi:10.1002/jbmr.4754
- [J43] Rossi N.<sup>†</sup>, Lee K. A.<sup>†</sup>, Bermudez M.V., **Visconti A.**, Thomas A.M., Bolte L.A., ..., Weersma R.K., Hospers G.A.P., Fehrmann R.S.N, Bataille V.<sup>‡</sup>, and Falchi M.<sup>‡</sup>, Circulating inflammatory proteins associate with response to immune checkpoint inhibition therapy in patients with advanced melanoma, EBioMedicine, 2022, doi:10.1016/j.ebiom.2022.104235
- [J42] Visconti A.<sup>†</sup>, Murray B.<sup>†</sup>, Rossi N., Wolf J., Ourselin S., Spector T.D., Freeman E.E., Bataille V.<sup>‡</sup>, and Falchi M.<sup>‡</sup> Cutaneous Manifestations of SARS-CoV-2 infection during the Delta and Omicron waves in 348,691 UK users of the UK ZOE COVID Study App, British Journal of Dermatology, 2022, doi:10.1111/bjd.21784
- [J41] Le Roy C.I., Kurilshikov A., Leeming E.R., Visconti A., Bowyer R.C.E, Menni C., Falchi M., Koutnikova H., Veiga P., Zhernakova A., Derrien M., and Spector T.D. Yoghurt consumption is associated with changes in the composition of the human gut microbiome and metabolome, BMC Microbiology, 2022, doi:10.1186/s12866-021-02364-2
- [J40] Murray B., Kerfoot E., Chen L., Deng J., Graham M.S., Sudre C.H., Molteni E., Canas L.S., Antonelli M., Klaser K., Visconti A., Hammers A., Chan A.T., Franks P.W., Davies R., Wolf J., Spector T.D., Steves C.J., Modat M., and Ourselin S. Accessible data curation and analytics for international-scale citizen science datasets, Scientific Data, 2021, doi:10.1038/s41597-021-01071-x
- [J39] Grosche S.<sup>†</sup>, Marenholz I.<sup>†</sup>, Esparza-Gordillo J.<sup>†</sup>, Arnau-Sole A.<sup>†</sup>, ..., **Visconti A.**, ..., Worth CL, Xu CJ, and Lee YA, Rare variant analysis in eczema identifies exonic variants in DUSP1, NOTCH4 and SLC9A4, Nature Communications, 2021, doi:10.1038/s41467-021-26783-x
- [J38] Dotz V.<sup>†</sup>, **Visconti A.**<sup>†</sup>, Lomax-Browne H.<sup>†</sup>, Florent C.<sup>†</sup>, Ederveen A.H., Medjeral-Thomas N., Cook H.T., Pickering M., Wuhrer M.<sup>‡</sup>, and Falchi M.<sup>‡</sup>, O- and N-Glycosylation of Serum Immunoglobulin A is Associated with IgA Nephropathy and Glomerular Function, Journal of the American Society of Nephrology, 2021, doi:10.1681/ASN.2020081208
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- [J33] Visconti A.<sup>†</sup>, Bataille V.<sup>†</sup>, Rossi N., Kluk J., Murphy R., Puig S., Nambi R., Bowyer R.C.E., Murray B., Bournot A., Wolf J., Ourselin S., Steves C., Spector T.D.<sup>‡</sup>, and Falchi M.<sup>‡</sup>, *Diagnostic value of cutaneous manifestation of SARS-CoV-2 infection*, British Journal of Dermatology, 2021, doi:10.1111/bjd.19807

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