Saul Pierotti

Computational biologist — PhD fellow at EMBL-EBI Born in 1995 — Italian citizen

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Research experience

European Bioinformatics Institute (EMBL-EBI) University of Cambridge

Cambridge, UK Jan 2021 — Jun 2021

Predoctoral fellow; Supervisor: Prof. Ewan Birney

QTL mapping and exploration of GxE, GxG, and indirect (social) genetic effects in medaka fish.

- Developed and made publicly available 3 Nextflow computational pipelines for genotype imputation, linear mixed model GWAS, and causal variant detection and prioritization (1 published and 1 preprinted)
- Collaborated closely with experimental research groups and delivered a complex GWAS project involving more than 2200 samples that led to the identification of 16 QTLs in medaka fish, 4 of which experimentally validated (manuscript in writing)
- Discovered pervasive gene by environment and genetic interactions in medaka fish, one of which experimentally validated
- Collaborated internationally with 3 research groups from Germany and Japan
- Ran a phenotyping campaign for medaka fish behaviour hosted by a collaborator and collected videos of more than 790 medaka fish pairs
- Helped with data acquisition and coordinated data storage, replication, and transfer from Japan to the UK for a phenotyping campaign of more than 1500 wild medaka fish embryos
- Led the writing of multiple manuscripts and directly handled interactions with scientific journals from submission to publication
- Run analyses on the UK biobank research platform (RAP) on a dataset of almost 500000 samples and millions of genetic variants
- Presented talks and posters at numerous internal and external events
- Delivered training for the Nextflow workflow language and gene-by-environment interactions to an internal and external scientific audience

Science for Life Laboratory (SciLifeLab)

Stockholm, Sweden

Master thesis; Supervisor: Prof. Arne Eloffson

Jan 2021 — Jun 2021

Prediction of protein contact maps from deep mutational scanning data using machine-learning

- Developed a protein contact map prediction method using gradient-boosted trees, neural networks, and other machine-learning approaches
- Achieved equivalent performance to a previously published method but without using protein structural features (which are hard and expensive to obtain)

Max Planck Institute for Evolutionary Biology

Plön, Germany

Intern; Supervisors: Prof. Arne Traulsen, Dr. Jenna Gallie

 $\mathrm{Jun}\ 2019 - \mathrm{Sep}\ 2019$

Experimental evolution of Escherichia coli and Pseudomonas fluorescens in long-term batch cultures

• Cultured bacteria and detected colony shape and number changes following evolution on different culture media

University of Perugia

Perugia, Italy

Bachelor thesis; Supervisor: Dr. Manlio di Cristina

Oct 2018 — Oct 2019

Characterization of vacuolar proteins in Toxoplasma gondii

- Performed PCR, CRISPR, gel electrophoresis, immunofluorescence staining, and other molecular biology techniques
- Obtained and assessed the effect of gene knockouts in Toxoplasma gondii
- Handled infectious Toxoplasma qondii samples and infected human cell lines

EDUCATION

European Bioinformatics Institute (EMBL-EBI)

University of Cambridge

Cambridge, UK Expected Oct 2025

PhD candidate at the University of Cambridge as part of the the EMBL International PhD Program (EIPP) — Applicant acceptance rate 2.7%

University of Bologna

Bologna, Italy

International Master Degree in Bioinformatics (MSc)

Jul 2021

Final mark: 110/110 cum laude

University of Perugia

Perugia, Italy Oct 2019

Bachelor's degree in Biotechnology (BSc)

Final mark: 110/110 cum laude and honorable mention

PUBLICATIONS

S. Pierotti, B. Welz, M. Osuna-López, T. Fitzgerald, J. Wittbrodt, E. Birney; Genotype imputation in F2 crosses of inbred lines, *Bioinformatics Advances*, 2024, https://doi.org/10.1093/bioadv/vbae107

Preprints

- * these authors contributed equally to this work
- **S. Pierotti**, T. Fitzgerald, E. Birney; FlexLMM: a Nextflow linear mixed model framework for GWAS, preprint on arXiv, 2024, https://doi.org/10.48550/arXiv.2410.01533
- S. Pierotti*, I. Brettell*, T. Fitzgerald, C. Herder, N. Aadepu, C. Pylatiuk, J. Wittbrodt, E. Birney, F. Loosli; Measurement and classification of bold-shy behaviours in medaka fish, manuscript ready for preprint pending final co-author approval

PLANNED PUBLICATIONS

Co-first author paper on the detection of 16 QTLs for heart rate in medaka, their CRISPR validation, and the characterization of GxE and GxG effects

Collaboration with Bettina Welz (Wittbrodt group, Heidelberg University, Germany)

Co-first author paper on the effect of direct and indirect (social) genetic effects on medaka behaviour Collaboration with Ian Brettel (Birney group, EMBL-EBI) and the group of Felix Loosli (KIT, Karlsruhe, Germany)

First author application note on a genetic variant prioritization pipeline

First author paper on a GWAS for behavioural phenotypes in medaka Collaboration with the group of Felix Loosli (KIT, Karlsruhe, Germany)

Conference presentations

The Biology of Genomes, Cold Spring Harbor Laboratory (Cold Spring Harbor, USA), May 7th to 11th, 2024 — poster

https://meetings.cshl.edu/abstracts.aspx?meet=GENOME&year=24

NRBP Medaka International Workshop, National Institute for Basic Biology (Okazaki, Japan), April 26th to 27th, 2024 — talk

https://sites.google.com/nibb.ac.jp/nbrp-medaka-workshop/home-program

TEACHING EXPERIENCE

 $\rm EMBL\text{-}EBI$ course "Exploring Gene and Environmental Exposure Interactions to Understand Human Health and Disease", February 2024

https://www.ebi.ac.uk/training/events/exploring-gene-and-environmental-exposure-interactions-understand-human-health-and-disease/

EMBL-EBI course "Using fish models to explore the impact of Gene by Environment (GxE)", October 2022 https://www.ebi.ac.uk/training/events/medaka-workshop

Nextflow practical for the EBI predoc course (online), November 2022 https://saulpierotti.github.io/nextflow_workshop_ebi_predoc_course_2022

LANGUAGES

English: proficient, CEFR level C2. IELTS score 8.0. CAE with grade A awarded by Cambridge ESOL.

Italian: native speaker. Others: currently studying Turkish, German and Dutch at a beginner level.

Referees

Ewan Birney — $current\ PhD\ supervisor\ and\ director\ of\ EMBL-EBI\ (Cambridge,\ UK)$ birney@ebi.ac.uk

Jochen Wittbrodt — close experimental collaborator and group leader at Heidelberg University (Germany) jochen.wittbrodt@cos.uni-heidelberg.de

Virginie Uhlman — member of thesis advisory committee and director of the University of Zurich BioVisionCenter (Switzerland)
virginie.uhlmann@uzh.ch