PI profile

## Pier Palamara

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| A person in a blue shirt  Description automatically generated with medium confidence | **Prof. Pier Palamara**  **Titles**: Associate Professor of Statistical and Population Genetics, Group Leader  **Location**: Department of statistics / Wellcome Centre for Human Genetics  **Department**: Department of statistics / Wellcome Centre for Human Genetics  **Group**: Palamara  **Webpage**: <http://www.stats.ox.ac.uk/~palamara>  **Email**: [palamara@stats.ox.ac.uk](mailto:palamara@stats.ox.ac.uk) |

### GMS themes:

* Genomic analysis (bioinformatics and statistical genetics)
* Genomics of disease

### Research Overview

Our work is at the intersection of computer science, statistics, and genetics. We develop statistical and machine learning algorithms to enable new types of analyses in large human genomic data sets. Specific areas of research include:

* Developing algorithms for simulation, reconstruction, and analysis of large-scale genealogical data (gene genealogies, haplotype sharing, phasing, imputation).
* Studying demographic events and evolutionary parameters (migration, expansion/contraction of populations, natural selection, mutation/recombination rates).
* Studying the genetic architecture of complex traits and detecting disease-causing variation in the human genome (heritability, polygenic prediction, association).

Project areas:

During the past few years our group has been developing several new methods to infer the presence of hidden relatedness and to reconstruct deep genealogical structures from large genotyping and sequencing data sets [Palamara et al. Nature Genetics 2018; Nait Saada et al. Nature Communications 2020; Zhang et al. Biorxiv 2021]. We have applied these algorithms to the UK Biobank data set, studying fine-scale population structure ([website](https://ukancestrymap.github.io/)), recent evolutionary history, and association between heritable traits and unobserved genomic variation. We are working on extending this framework in several directions, which will create new opportunities for the study of human evolutionary history and disease.

We have also been developing new machine learning methodology for analyses in statistical and population genetics. Examples include efficient stochastic variational inference approaches to perform statistically powerful and robust genome-wide association, and methods that use deep neural networks to infer genealogical relationships across samples.

Several DPhil projects are available within these research areas, please get in touch to discuss these directions.

### Representative publications:

* B. Zhang, A. Biddanda, P. Palamara. Biobank-scale inference of ancestral recombination graphs enables genealogy-based mixed model association of complex traits. Biorxiv.
* R. Fournier, D. Reich, P. Palamara. Haplotype-based inference of recent effective population size in modern and ancient DNA samples. Biorxiv.
* J. Nait Saada, G. Kalantzis, D. Shyr, F. Cooper, M. Robinson, A. Gusev, P. Palamara. Identity-by-descent detection across 487,409 British samples reveals fine-scale population structure, evolutionary history, and trait associations. Nature Communications, 2020.
* P. Palamara, J. Terhorst, Y. Song, A. Price. High-throughput inference of pairwise coalescence times identifies signals of selection and enriched disease heritability. Nature Genetics, 2018.
* S. Gazal, H. Finucane, N. Furlotte, P. Loh, P. Palamara, X. Liu, A. Schoech, B. Bulik-Sullivan, B. Neale, A. Gusev, A. Price. Linkage disequilibrium–dependent architecture of human complex traits shows action of negative selection. Nature Genetics, 2017.
* P. Palamara. ARGON: Fast, whole-genome simulation of the discrete time Wright-Fisher process. Bioinformatics, 2016.
* P. Loh, P. Palamara, A. Price. Fast and accurate long-range phasing in a UK Biobank cohort. Nature Genetics, 2016
* P. Palamara, et al.. Leveraging distant relatedness to quantify human mutation and gene conversion rates. The American Journal of Human Genetics, 2015.
* P. Palamara, T. Lencz, A. Darvasi, I. Pe'er. Length distributions of identity by descent reveal fine-scale demographic history. The American Journal of Human Genetics. 2012.

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