

ANNEX B

UNIVERSITY OF MILAN

Public selection for recruiting No._1_ tenure track researcher(s) (RTT) for competition sector _____05/E2 - Molecular Biology_____,
(scientific-disciplinary sector __BIO/11 - Molecular Biology_____) at the Department of
Medical Biotechnology and Translational Medicine _____, (announcement published
in Official Gazette No. ____G.U. 93_____ of __05/12/2023____) -
Competition code 5437 _____

Michaela Fakiola

CURRICULUM VITAE

PERSONAL DATA (DO NOT INCLUDE YOUR PERSONAL ADDRESS AND LANDLINE OR MOBILE PHONE NUMBER)

SURNAME	FAKIOLA
NAME	MICHAELA
DATE OF BIRTH	02 JANUARY 1978

QUALIFICATIONS

DEGREE

(Specify full degree name, University, date, etc.)

10/2001-09/2002

MSc in Environmental Technology (Health and Environment option)

Imperial College London, UK

Thesis Title: Risk of ischaemic heart disease (IHD) associated to aircraft noise and road traffic air pollution near Heathrow airport, Department of Epidemiology and Public Health, Imperial College School of Medicine, St Mary's Hospital (Supervisor: Dr Lars Jarup).

Awarded 01/11/2002

10/1997-09/2001

BSc (Hons) in Biology (4-year course)

Aristotle University of Thessaloniki, Greece.

Thesis title: Characterization of the putative promoter of the 'Immediate-early' gene of human herpesvirus 6 (Supervisor: Prof Minas Arsenakis).

Awarded 22/07/2002

DOCTORAL DEGREE OR EQUIVALENT QUALIFICATION EARNED IN ITALY OR ABROAD / MEDICAL SPECIALISATION DIPLOMA OR EQUIVALENT QUALIFICATION, FOR THE RELEVANT SECTORS, EARNED IN ITALY OR ABROAD

(Specify qualification full name, institution, date, etc.)

2004-2008

PhD in Medicine (Human Genetics)

University of Cambridge, UK

Thesis Title: The genetics of human susceptibility to visceral leishmaniasis in India and Sudan (Supervisor: Prof Jenefer M Blackwell).

Awarded 18/07/2009

RESEARCH CONTRACTS, RESEARCH FELLOWSHIP CONTRACTS, POSTDOCTORAL SCHOLARSHIPS OR SIMILAR CONTRACTS

(Specify, for each contract, university/institution, starting and termination date, etc.)

01/2023 - present

Assegno di Ricerca tipo B

Department of Medical Biotechnology and Translational Medicine, University of Milan, Italy

Principal Investigator (PI): Prof Massimiliano Pagani

01/2022 - 12/2023

Postdoc Bioinformatician

Molecular Oncology and Immunology Lab, IFOM-ETS The AIRC Institute of Molecular Oncology, Milan, Italy

PI: Prof Massimiliano Pagani

01/2021 - 12/2021

Post-doctoral Research Fellowship, Fondazione UMBERTO VERONESI

IFOM-ETS, Milan, Italy

PI: Prof Massimiliano Pagani

01/2020 - 12/2020

Post-doctoral Research Fellowship, Fondazione UMBERTO VERONESI

IFOM-ETS, Milan, Italy

PI: Prof Massimiliano Pagani

01/2018 - 10/2019 (with an interruption of one year maternity leave)

Post-doctoral Research Fellowship, Fondazione UMBERTO VERONESI

National Institute of Molecular Genetics (INGM), Milan, Italy

PI: Prof Massimiliano Pagani

03/2017 - 12/2017

Post-doctoral Research Fellow

INGM, Milan, Italy

PI: Prof Massimiliano Pagani

03/2016 - 02/2017

Post-doctoral Research Fellowship, Fondazione UMBERTO VERONESI

INGM, Milan, Italy

PI: Prof Massimiliano Pagani

07/2014 - 09/2015

Independent Research Fellow

Department of Pathology, University of Cambridge, Cambridge, UK

01/2014 - 06/2014

Research Associate

Department of Pathology, University of Cambridge, Cambridge, UK

PI: Prof Jim Kaufman

02/2013 - 05/2013

Raine International Research Fellow

Telethon Kids Institute, University of Western Australia, Australia

PI: Prof Jenefer M Blackwell

10/2012 - 12/2012

Visiting Scientist

University of Iowa Carver College of Medicine, Iowa City, IA, USA

PI: Prof Mary E Wilson

03/2009 - 12/2013

Research Associate

Cambridge Institute for Medical Research (CIMR), University of Cambridge, Cambridge, UK

PI: Prof Jenefer M Blackwell

01/2003 - 05/2004

Research Assistant

Cambridge Institute for Medical Research (CIMR), University of Cambridge, Cambridge, UK

PI: Prof Jenefer M Blackwell

TEACHING ACTIVITIES AT ITALIAN OR FOREIGN UNIVERSITIES

(Specify academic year, university, degree course, number of hours etc.)

FRONTAL TEACHING

(2014-present)

2022-2023

Guest lecturer in “Human genome organization and epigenomic regulation” (Prof. Massimiliano Pagani) for the Master’s Degree Programme in Medical Biotechnology and Molecular Medicine. (2 hours)

2021-2022

Lecturing in “Next-generation sequencing: from the bench to the data analysis” for the PhD course of the Department of Medical Biotechnology and Translational Medicine, Università degli Studi di Milano. Introduction of PhD students to RNA-seq, ATAC-seq and scRNA-seq data analyses. (9 hours)

2017-2018

Guest lecturer in “Human Genetic Variation” (Prof Massimiliano Pagani), for the International Medical School and the Master’s Degree Program in Bioinformatics and Functional Genomics, Università degli Studi di Milano. (4 hours)

2014-2015

Lecturing for the Natural Sciences Tripos course, Department of Pathology, University of Cambridge, Cambridge UK

Lecturing (2 lectures), marking annual exam papers, and demonstrating on immunology practicals.

OTHER FORMS OF TEACHING/SUPERVISION

(2005-present)

Co-supervision and hands-on training in next-generation sequencing data analyses (epigenetic, transcriptomic) of PhD and Master students in the Prof Massimiliano Pagani laboratory:

11/2023 - 02/2023

Project supervisor for the Master’s course in Bioinformatics and Functional Genomics, Università degli Studi di Milano (Coordinator: Prof Massimiliano Pagani).

10/2020 - present

Formal assignment of supervision of a student for the PhD Course in Experimental Medicine and Medical Biotechnologies (XXXVII cycle), Department of Medical Biotechnology and Translational Medicine, Università degli Studi di Milano. Tutor: Massimiliano Pagani.

10/2020 - 09/2021

Formal assignment of co-supervision of a student for the Master’s Degree Programme in Medical Biotechnology and Molecular Medicine, Università degli Studi di Milano. Tutor: Massimiliano Pagani

10/2017 - 03/2021

Formal assignment of co-supervision of a student for the PhD Course in Experimental Medicine and Medical Biotechnologies (XXXIII cycle), Department of Medical Biotechnology and Translational Medicine, Università degli Studi di Milano. Tutor: Massimiliano Pagani.

Supervision activities at University of Cambridge:

2014 - 2015

Supervisor of two final year undergraduate research projects, Natural Sciences Tripos course, Department of Pathology, University of Cambridge, Cambridge UK

2005 - 2016

Co-supervision and hands-on training of two PhD students, visiting postdoctoral fellows and research assistants in the Prof JM Blackwell laboratory, University of Cambridge, Cambridge UK.

ATTESTED TRAINING OR RESEARCH ACTIVITIES AT QUALIFIED ITALIAN OR FOREIGN INSTITUTIONS

(Specify academic year, institution, course, period, etc.)

04/2021

Patent Course - IFOM Technology Transfer Office, Milan, Italy (4-session course)

22/02/2018 - 23/02/2018

Software Carpentry Workshop (on Bash, Git, Python coding languages) - Elixir-Italy, University of Milan- Bicocca, Italy (2-day course)

17/07/2014 - 18/07/2014

An introduction to biological problems in R. School of Biological Sciences Bioinformatics Training. University of Cambridge, UK (2-day course)

10/01/2011 - 12/01/2011

Introduction to Programming with Perl. School of Biological Sciences. University of Cambridge, UK (3-day course)

24/02/2010 - 26/02/2010

Microarray data analysis using R and Bioconductor. Cancer Research UK, University of Cambridge Workshop, UK (3-day course)

20/07/2005 - 26/07/2005

Human Genome Analysis: Genetic Analysis of Multifactorial Diseases, Wellcome Trust Advanced Course, Wellcome Genome Campus, Hixton, Cambridge, UK (6-day course)

IMPLEMENTATION OF PROJECTS

(Specify date, project name, etc.)

2022 - present

Epigenomic and gene expression profiling of brain-colon and brain-breast metastases to decipher their distinct and shared epigenomic patterns compared to the primary tumors from which they originate. IFOM ETS - The AIRC Institute of Molecular Oncology, Milan, Italy - Prof Massimiliano Pagani group

2021 - present

Molecular profiling of tumor-infiltrating T cell lymphocytes through the integration of epigenetic (ChIP-seq for histone marks and ATAC-seq), bulk and single-cell RNA-seq data, transcription factor footprinting, and motif discovery analyses. Part of a Fondazione Umberto Veronesi - funded postdoctoral fellowship. IFOM ETS - The AIRC Institute of Molecular Oncology, Milan, Italy - Prof Massimiliano Pagani group

2021 - present

3D genome characterization of patient-derived colorectal cancer (CRC) organoids to decipher the genome-wide chromosomal interactions and target genes of a pan-cancer enhancerome. The project employs chromosome conformation capture techniques (e.g. capture HiC for enhancers and promoters). IFOM ETS - The AIRC Institute of Molecular Oncology, Milan, Italy - Prof Massimiliano Pagani group

2017 - 2021

Characterization of patient-derived CRC organoids through transcriptional (RNA-seq), epigenomic (ChIP-seq), and genetic (WES) analyses. Part of a Fondazione Umberto Veronesi - funded postdoctoral fellowship. IFOM ETS - The AIRC Institute of Molecular Oncology, Milan, Italy - Prof Massimiliano Pagani group. Co-first authorship in the attached publication in Nature Communications.

2020 - 2021

Identification of genetic variants contributing to severe otitis media in an Aboriginal community using exome sequencing and FUnctional and Mapping and Annotation (FUMA) analyses. In collaboration with partners from The University of Western Australia (Australia) and Newcastle University (UK). Co-first authorship in the attached publication in Clinical Infectious Diseases.

2019 - 2020

Genome-wide association study (GWAS) to identify risk loci contributing to cutaneous leishmaniasis. with partners from The University of Western Australia (Australia), Newcastle University (UK), and the National Institute of Science and Technology in Tropical Diseases, Salvador (Brazil). Co-first author in the attached publication in Clinical Infectious Diseases.

2016 - 2017

Identification of lymphocyte-specific long intergenic non-coding RNAs (lncRNAs) within known regions of genetic predisposition to cancer and immune-related diseases. Part of a Fondazione Umberto Veronesi - funded postdoctoral fellowship. INGM - Istituto Nazionale di Genetica Molecolare, Milan, Italy - Prof Massimiliano Pagani group

2015 - 2021

The role of IL-10 as modulator of the transcriptional responses to leishmanial antigens in VL patients. Department of Pathology, University of Cambridge, UK and INGM - Istituto Nazionale di Genetica Molecolare, Milan, Italy. Last author in the attached publication in the Journal of Infectious Diseases.

2014 - 2019

Deciphering the genome-wide transcriptional blood signatures of active and treated visceral leishmaniasis patients and endemic healthy controls. Department of Pathology, University of Cambridge, UK. First author in the attached publication in PLoS Neglected Tropical Diseases.

2014

Differential expression of major histocompatibility (MHC) class II molecules in intestinal tissues. Department of Pathology, University of Cambridge, UK - Prof Jim Kaufman group. Attached publication in Frontiers in Immunology.

2013 - 2017

Understanding the molecular mechanisms underpinning the association of HLA-DRB1 alleles with risk versus protection in visceral leishmaniasis using HLA imputation, epitope capture from dendritic cells, and in silico epitope binding predictions of leishmanial antigens to HLA-DRB1 alleles. Department of Pathology, University of Cambridge, UK - Prof Jenefer M Blackwell group. Co-first author in the attached publication in the Journal of Immunology.

2012 - 2019

Immunological and genetic markers as predictors of progression from asymptomatic to active visceral leishmaniasis. Cambridge Institute for Medical Research (CIMR) and Department of Pathology, University of Cambridge, UK - Prof Jenefer M Blackwell group. Attached publication in PLoS Neglected Tropical Diseases.

2012 - 2018

Expression of class II HLA-DR in myeloid and lymphoid cells of VL patients. Cambridge Institute for Medical Research (CIMR) and Department of Pathology, University of Cambridge, UK - Prof Jenefer M Blackwell group. Attached publication in Immunology.

2012 - 2015

Understanding the genetic basis of IgG responses in Streptococcus lung infection using a GWAS. Cambridge Institute for Medical Research (CIMR), Cambridge, UK - Prof Jenefer M Blackwell group. Co-first author in the attached publication in Genes and Immunity.

2010 - 2015

Genome-wide association study of Type 2 Diabetes in an Australian Aboriginal population. Telethon Kids Institute, University of Western Australia, Australia - Prof Jenefer M Blackwell group. Co-first author in the attached publication in PLoS ONE.

2008 - 2012

Genome-wide association study of visceral leishmaniasis in two independent case-control and family cohorts from endemic population as part of the Wellcome Trust Case Control Consortium 2 (WTCCC2). Cambridge Institute for Medical Research (CIMR), Cambridge, UK - Prof Jenefer M Blackwell group. First author in the attached publication in Nature Genetics.

2007 - 2012

Genetic and functional evaluation of the role of DLL1, SLC11A1, CXCR1 and CXCR2 in visceral leishmaniasis. Cambridge Institute for Medical Research (CIMR), Cambridge, UK - Prof Jenefer M Blackwell group. Published in Infection Genetics and Evolution and BMC Medical Genetics.

2005 - 2010

The genetic and functional role of the Notch ligand DLL1 in human susceptibility to visceral leishmaniasis using haplotype association, gene expression analyses and discovery of conserved non-coding regulatory elements. Cambridge Institute for Medical Research (CIMR), Cambridge, UK - Prof Jenefer M Blackwell group. First author in the Journal of Infectious Diseases.

2004 - 2007

Genome-wide linkage scan analysis with refined mapping for human susceptibility to visceral leishmaniasis in India. Cambridge Institute for Medical Research (CIMR), Cambridge, UK - Prof Jenefer M Blackwell group. First author in PLoS ONE.

2003 - 2004

The genetics of infectious diseases in endemic populations using genome wide linkage scan analyses. Cambridge Institute for Medical Research (CIMR), Cambridge, UK - Prof Jenefer M Blackwell group. Published in PLoS Genetics and Genes and Immunity.

ORGANISATION, SUPERVISION AND COORDINATION OF NATIONAL AND INTERNATIONAL RESEARCH GROUPS, OR PARTICIPATION IN THEM

(For each entry, specify year, role, research group, etc.)

2017 - 2021

Participation in the Epigenomics Flagship Project (EPIGEN-Subproject 9), funded by MIUR-CNR and the Italian Ministry of Health. Co-PI: Prof Massimiliano Pagani at IFOM ETS - The AIRC Institute of Molecular Oncology, Milan, Italy.

The study investigated the epigenetic reprogramming in human colorectal cancer using patient-derived organoids with the aim of highlighting the key transcriptional regulators of the aberrant CRC CRC enhancerome. The work involved integration of omics data (ChIP-seq for histone marks and transcription factors, RNA-seq) and experimental studies (transcription factor inhibition in organoids). M. Fakiola's work was funded by an Umberto Veronesi Foundation Fellowship and resulted in co-first authorship in the attached publication in Nature Communications.

2010 - 2021

Collaborated (from UK and Italy) with partners from The University of Western Australia (Australia), and from Newcastle University (Newcastle, UK) to conduct genomic studies, including genome-wide association studies of human susceptibility to type 2 diabetes and leishmaniasis.

1. Identification of genetic variants contributing to severe otitis media using exome sequencing and functional annotation of GWAS results (co-first author in Clinical Infectious Diseases 2021).
2. A genome-wide association study (GWAS) to identify risk loci contributing to cutaneous leishmaniasis (co-first author in Clinical Infectious Diseases 2020).
3. Study of functional variants for chronic renal disease, T2D and hypertension using whole exome sequencing data and pathway enrichment analysis (Scientific Reports 2018).
4. The first genome-wide association study of Type 2 Diabetes in an Australian Aboriginal population (co-first author in PLoS ONE 2015).

5. Understanding the genetic basis of IgG responses in Streptococcus lung infection using a GWAS (co-first author in Genes and Immunity 2015).

2012 - 2017

Co-coordination in a multi-project international collaborative network (partners from UK, USA, Belgium, Australia, India, Brazil), funded by the National Institutes of Health (NIH), USA (PI: Prof Shyam Sundar). Co-Leader in Sub-Project 4 (co-PI: Jenefer M Blackwell): Molecular and cellular action of HLA class II molecules, the major genetic risk factors for visceral leishmaniasis (VL).

1. Investigating the role of IL-10 as modulator of the transcriptional responses to leishmanial antigens in VL patients (last author in the Journal of Infectious Diseases 2021).
2. Deciphering the transcriptional blood signatures of active and amphotericin B treated visceral leishmaniasis patients (first author in PLoS Neglected Tropical Diseases 2019).
3. Identifying immunological and genetic markers as predictors of progression from asymptomatic to active visceral leishmaniasis (PLoS Neglected Tropical Diseases 2019).
4. Assessing the expression of class II HLA-DR in myeloid and lymphoid cells of VL patients (Immunology 2019).
5. Understanding the molecular mechanisms underpinning the association of HLA-DRB1 alleles with risk versus protection in visceral leishmaniasis. The study involved HLA alleles imputation, epitope capture from dendritic cells, and in silico epitope binding predictions of leishmanial antigens to HLA-DRB1 alleles (co-first author in Journal of Immunology 2018).

2012 - 2012

Associate Investigator in a University of Western Australia (UWA) Research Collaborative Award. PI: Prof Jenefer M Blackwell, collaborating institution University of Iowa College of Medicine, USA (Prof Mary E Wilson).

Title: Capitalising on UWA/University of Iowa connections to maximise the output of experimental studies of leishmaniasis using HLA-DRB1-humanised mouse models. Visiting Scientist at Prof Mary E Wilson group, Iowa University, USA.

2008 - 2012

Participation as postdoc in phase 2 of the Wellcome Trust Case Control Consortium (WTCCC2) and LeishGEN Consortium (PI: Prof Jenefer M Blackwell) to conduct the first GWAS of visceral leishmaniasis (VL) in collaboration with researchers from the University of Cambridge (UK), the Wellcome Trust Centre for Human Genetics (UK), University of Oxford (UK), the Newcastle University (UK), the Institute of Medical Sciences, Varanasi (India), and the Federal University of Rio Grande do Norte, Natal (Brazil). The study provided strong evidence for specific risk and protective MHC class II HLA-DRB1 molecules determining VL susceptibility and resulted in first authorship in the attached publication in Nature Genetics.

The work paved the way to a five-year NIH-TMRC (Tropical Medicine Research Centre program) multi-project grant renewal to understand the host-parasite interactions and the associated immune responses under the influence of the risk/protective HLA proteins in the context of *Leishmania* infection.

2004 - 2010

Participation in the Blackwell group at the Cambridge Institute for Medical Research, University of Cambridge, UK in collaborative projects with researchers from the Institute of Medical Sciences, Varanasi (India), the Federal University of Rio Grande do Norte, Natal (Brazil), the Institute of Endemic Diseases, University of Khartoum (Sudan), the Medical University of Gdansk (Poland) to perform genetic studies of human susceptibility to infectious diseases.

1. Understanding the role of the Notch ligand DLL1 in human susceptibility to visceral leishmaniasis by analysing resequencing and gene expression data and identifying conserved non-coding regulatory elements associated with phenotypic variability (first author in the Journal of Infectious Diseases 2011).
2. The first genome-wide linkage scan analysis with refined mapping for human susceptibility to visceral leishmaniasis in India. Using classification and spatial interpolation analyses the study showed geographical heterogeneity in the influence of novel loci on VL susceptibility (first author in PLoS ONE 2010).
3. Investigating the association between SLC11A1 and the risk of sarcoidosis (European Journal of Human Genetics 2005).
4. Deciphering the genetics of visceral leishmaniasis in an ethnic group in Sudan using genome wide linkage scan and Y chromosome haplotype analysis (PLoS Genetics 2004).

PRESENTATIONS AT NATIONAL AND INTERNATIONAL CONFERENCES AND INVITED SEMINARS

(Specify conference/convention title, date, etc.)

20/09/2023 - 23/09/2023

CICON23 - CRI-ENCI-AACR 7th International Cancer Immunotherapy Conference: Translating Science into survival, Milan, Italy.

Poster presentation: "Characterization of the epigenomic profile shaping human CD4⁺ FOXP3⁺ regulatory T cell identity in the tumor microenvironment".

05/04/2022 - 09/04/2022

Keystone Symposium - Single Cell Biology: Pushing New Frontiers in the Life Sciences, Florence, Italy.

Poster presentation: "Charting the epigenome of intratumoral CD4⁺ Type 1 regulatory T-cells through an integrative omics approach". Co-supervision of a Master's degree student (Tutor: Prof Massimiliano Pagani).

19/03/2014 - 20/03/2014

Invited Speaker at the Visceral Leishmaniasis workshop, National Institutes of Health (NIH), Bill & Melinda Gates Foundation Conference Center, Seattle, WA, USA.

Oral presentation title: "Technical and ethical feasibility of genetic fingerprinting across a study population".

The purpose of the meeting was to discuss specific aspects of the NIH and Bill & Melinda GATES FOUNDATION co-funded project in Epidemiology of Visceral Leishmaniasis (VL).

13/11/2013

Invited Speaker at the Cambridge Infectious Diseases 2013 Meeting of Minds, University of Cambridge, UK.

Oral presentation title: "Major genetic risk factor for visceral leishmaniasis lies at the heart of eliciting CD4 T cell immunity".

13/05/2013 - 17/05/2013

Oral presentation at the 5th World Congress on Leishmaniasis (2013), Porto de Galinhas, Pernambuco, Brazil.

Presentation title: "HLA class II association with visceral leishmaniasis: implications for vaccine development".

I communicated findings arising from the first genome-wide association study (GWAS) of visceral leishmaniasis (Nature Genetics, 2013, first authorship), conducted as part of phase 2 of the Wellcome Trust Case-Control Consortium (WTCCC).

01/05/2013

Invited Keynote Speaker at the Telethon Kids Institute, Perth, Australia.

Seminar title: "HLA Class II association with visceral leishmaniasis: a potential role in vaccine development?".

The lecture was given as part of the Raine International Visiting Research Fellowship awarded to me in 2013 by the Raine Medical Research Foundation, University of Western Australia (UWA).

15/08/2010 - 20/08/2010

Oral presentation at the XIth International Congress of Parasitology (ICOPA) hosted in 2010 in Melbourne, Australia.

Oral presentation title: "Genome wide linkage study for visceral leishmaniasis in India".

Communicated the major findings from my PhD studies at the University of Cambridge.

03/02/2009 - 07/02/2009

4th World Congress on Leishmaniasis, Lucknow, India

Poster presentation: "The genetics of susceptibility to visceral leishmaniasis in India".

28/05/2008 - 30/05/2008

Invited Speaker at the DMID International Research on Infectious Diseases (IRID) Meeting, National Institute of Allergy and Infectious Diseases, National Institutes of Health (NIH), Bethesda, Maryland, USA.

Oral Presentation title: "The Genetics of Susceptibility to Visceral Leishmaniasis in India".

NATIONAL AND INTERNATIONAL AWARDS AND ACCOLADES FOR RESEARCH ACTIVITY

(Specify award, date, issuing organisation, etc.)

2021 - Post-doctoral Research Fellowship, Fondazione UMBERTO VERONESI, Italy

2020 - Post-doctoral Research Fellowship, Fondazione UMBERTO VERONESI, Italy

2018 - Post-doctoral Research Fellowship, Fondazione UMBERTO VERONESI, Italy

2016 - Post-doctoral Research Fellowship, Fondazione UMBERTO VERONESI, Italy

2013 - Raine International Visiting Award, Raine Medical Research Foundation, University of Western Australia, Australia

OTHER RELEVANT INFORMATION

Description of research activities

During my PhD and postdoctoral studies in UK, Australia and USA, I developed expertise in human genetic susceptibility and participated in international collaborative projects (funded by the Wellcome Trust, UK and the National Institutes of Health, USA) that identified clinically relevant genes, and common and rare genetic risk variants for infectious diseases, metabolic disorders and immune-related diseases. These studies were complemented by transcriptional, cellular, and computational approaches to provide further leads on disease pathogenesis. Moving from Cambridge to Milan, I obtained funding from Fondazione Umberto Veronesi to apply my knowhow in genetic studies towards investigating the role of long non-coding RNAs in genetic predisposition to cancer and immune-related diseases. I subsequently reoriented my interests towards cancer research seeking to understand how the transcriptional and epigenetic reprogramming of lymphocyte populations and cancer cells lead to the impairment of immune responses and tumor growth. These studies expanded my expertise in omics-based data, including analyses of RNA-seq, ChIP-seq, ATAC-seq, whole-exome sequencing, single-cell RNA-seq, and transcription factor footprinting. My current and past research has focused on the following main areas:

- Tracing the roots of T lymphocyte-mediated immune suppression and dysfunction in cancer through epigenomics (ChIP-seq, ATAC-seq) and transcriptomics (bulk and single-cell RNA-seq).
- Dissecting the molecular complexity of colorectal cancer by analyzing the transcriptional and epigenetic landscape of genetically heterogeneous patient-derived tumor organoids and identifying the key regulators of the deregulated tumor-specific enhancerome.
- Molecular/Experimental studies to understand the host-parasite interactions and the associated immune responses under the influence of the risk/protective HLA proteins, including transcriptional profiling, epitope capture from dendritic cells, and imputation of classical HLA alleles and amino acids.
- A genome-wide association study of visceral leishmaniasis conducted as part of the Wellcome Trust Case Control Consortium (WTCCC2), providing novel insights into the role of the MHC class II HLA-DRB1 locus as the most important genetic risk factor for this infectious disease.

Abilitazione Scientifica Nazionale di Seconda Fascia nel Settore Concorsuale (2022-2032)

05/E2 - BIOLOGIA MOLECOLARE

05/I1 - GENETICA

06/A1 - GENETICA MEDICA

Bioinformatics experience

Experienced in the management and integration of next-generation sequencing (NGS) and microarray data:

- Analysis of transcriptomic data deriving from RNA sequencing and microarrays, and single cell transcriptomic data (scRNA-seq).
- Experienced in omics-based data for epigenomic analyses, including ChIP-seq data (chromatin immunoprecipitation followed by sequencing) for histone modification profiling and transcription factor occupancy; ATAC-seq data (assay for transposable accessible chromatin) for chromatin accessibility; data for 3D chromatin organisation derived from chromosome capture conformation approaches (enhancer capture HiC).
- De novo discovery of chromatin states from epigenomic features (ChromHMM).
- Use of scRNA-seq for identification of enhancer RNAs (eRNAs) and copy number variations (inferCNV).
- Application of motif enrichment and footprinting analyses to epigenomic data.
- Data analysis and management of whole-exome sequencing studies, genome-wide association studies, and HLA-imputation methods.

Experienced with bioinformatics tools for omics analyses, including deepTools, DESeq2, MACS2, HOMER, SCANPY, GATK (genome analysis tool kit), the Galaxy platform, PLINK, SNPTEST, fastPHASE, IMPUTE, NetMHCIIpan.

Knowledge of scripting and data analysis in common programming languages, such as Bash, R and Python. Use of the Nextflow workflow management system.

Editorial Activity

Receiving Editor for the ELSEVIER Journal *Infection, Genetics, and Evolution* (IF 4.393) (2015-2017).

Reviewer for scientific journals, including *Human Genetics*, *PLoS ONE*, *Human Immunology*, *PLoS Neglected Tropical Diseases*, *Journal of Infectious Diseases*.

Book Chapter

Fakiola M, Lu W, Jamieson SE, Peacock CS. Genomics and infectious diseases: susceptibility, resistance, response and anti-microbial therapy. In *Genomic Medicine: Principles and Practice* (2nd edition), 2014, Oxford University Press.

Edited by Dhavendra Kumar and Charis Eng.

Memberships to Scientific Societies (active)

British Society for Immunology and British Society for Parasitology.

Complete list of Publications

34 peer-reviewed publications; **H-index=18; FIRST, co-FIRST or LAST author in 12 publications** in the field of genomics, transcriptomics, population genetics, and bioinformatics for human diseases (Nature Communications; Nature Genetics; Frontiers in Immunology; Journal of Immunology; Immunology; Journal of Infectious Diseases; Clinical Infectious Diseases; Scientific Reports)

Scopus ID: 8601763300; ORCID: 0000-0001-9071-4709

Citations: Scopus 902; Google Scholar 1364

Della Chiara G*, Gervasoni F*, **Fakiola M***, Godano C*, [...], Piccolo S, Pagani M. Epigenomic landscape of human colorectal cancer unveils an aberrant core of pan-cancer enhancers orchestrated by YAP/TAZ. *Nature Communications*, 2021 Apr 20;12(1):2340. doi: 10.1038/s41467-021-22544-y. *Equal first authorship.

Singh OP, Syn G, Nylén S, Engwerda C, Sacks D, Wilson ME, Kumar R, Chakravarty J, Sundar S, Blackwell JM*, **Fakiola M***. Anti-Interleukin-10 unleashes transcriptional response to leishmanial antigens in visceral leishmaniasis patients. *Journal of Infectious Diseases*, 2021 Feb 13;223(3):517-521. doi: 10.1093/infdis/jiaa381. *Joint senior authors

Jamieson SE*, **Fakiola M***, Tang D, Scaman E, Syn G, Francis RW, Coates HL, Anderson D, Lassmann T, Cordell HJ, Blackwell JM. Common and rare genetic variants that could contribute to severe otitis media in an Australian Aboriginal population. *Clinical Infectious Diseases*, 2021 Mar 9:ciab216. doi: 10.1093/cid/ciab216. *Equal first authorship

Blackwell JM, **Fakiola M**, Singh OP. Genetics, Transcriptomics and Meta-Taxonomics in Visceral Leishmaniasis. *Frontiers in Cellular and Infection Microbiology*, 2020 Nov 25;10:590888. doi: 10.3389/fcimb.2020.590888. eCollection 2020.

Castellucci LC*, Almeida L*, Cherlin S*, **Fakiola M***, Francis RW, Carvalho E, Santos da Hora A, Souza do Lago T, Figueiredo AB, Cavalcanti CM, Alves NS, Morais KLP, Teixeira-Carvalho A, Dutra WO, Gollob KJ, Cordell HJ, Blackwell JM. A Genome-Wide Association Study Identifies SERPINB10, CRLF3, STX7, LAMP3, IFNG-AS1 and KRT80 As Risk Loci Contributing to Cutaneous Leishmaniasis In Brazil. *Clinical Infectious Diseases*, 2021 Aug 23;ciaa1230. Online ahead of print. doi: 10.1093/cid/ciaa1230. *Equal first authorship

Blackwell JM, **Fakiola M**, Castellucci LC. Human genetics of leishmania infections. *Human Genetics*, 2020 Jun;139(6-7):813-819. Review.

Fakiola M*, Singh OP*, Syn G, Singh T, Singh B, Chakravarty J, Sundar S, Blackwell JM. Transcriptional blood signatures for active and amphotericin B treated visceral leishmaniasis in India. *PLoS Neglected Tropical Diseases*, 2019 Aug 16 ;13(8):e0007673. doi: 10.1371/journal.pntd.0007673. eCollection. *Equal first authorship.

Chakravarty J, Hasker E, Kansal S, Singh OP, Malaviya P, Singh AK, Chourasia A, Singh T, Sudarshan M, Singh AP, Singh B, Singh RP, Ostyn B, **Fakiola M**, Picado A, Menten J, Blackwell JM, Wilson ME, Sacks D, Boelaert M, Sundar S. Determinants for progression from asymptomatic infection to symptomatic visceral leishmaniasis: A cohort study. *PLoS Neglected Tropical Diseases*, 2019 Mar 27;13(3):e0007216. doi: 10.1371/journal.pntd.0007216. eCollection

Singh B, **Fakiola M**, Sudarshan M, Oommen J, Singh SS, Sundar S, Blackwell JM. HLA-DR Class II expression on myeloid and lymphoid cells in relation to HLA-DRB1 as a genetic risk factor for visceral leishmaniasis. *Immunology*, 2019 Feb;156(2):174-186. doi: 10.1111/imm.13018.

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SCIENTIFIC PRODUCTION

SCIENTIFIC PUBLICATIONS

(For each publication, specify the following: authors' names, full title, publisher, date and place of publication, ISBN/ISSN/DOI or equivalent code)

1. Della Chiara G*, Gervasoni F*, **Fakiola M***, Godano C*, [...], Piccolo S, Pagani M. Epigenomic landscape of human colorectal cancer unveils an aberrant core of pan-cancer enhancers orchestrated by YAP/TAZ. *Nature Communications*, 2021 Apr 20;12(1):2340. doi: 10.1038/s41467-021-22544-y. *Equal first authorship. IF: 17.694
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