Oxford Course Bios

Nichola Cooper, MD

Nichola Cooper is a consultant haematologist and honorary senior lecturer at Hammersmith Hospital, Imperial Health Care NHS Trust, London, UK. She completed her undergraduate degrees at Cambridge University and the London Hospital Medical School. During her haematology training, Dr Cooper undertook a research fellowship at Cornell Medical School, New York Presbyterian Hospital, USA, with Jim Bussel where she was involved with research into existing and experimental treatments in patients with ITP. She has subsequently studied at the Institute of Child Health, Great Ormond Street Hospital where she gained further experience in both paediatric and adult immunodeficiency disorders. Dr Cooper currently runs the centre for immune haematology at the Hammersmith hospital and has a research group investigating the pathology of ITP.

Martina Daly, PhD

After graduating from University College Dublin with a degree in Biochemistry, I completed a PhD at the Children's Research Centre, Dublin, where I first became interested in haemostasis, particularly in genetic variants of antithrombin. This was followed by a postdoctoral research post in Dublin, before I joined the Department of Haematology, Cambridge UK in 1988 where I worked on the investigation of the molecular basis of antithrombin deficiency. I came to Sheffield in 1991, to take up a postdoctoral research post working with Professor Ian Peake on the investigation of genotype-phenotype relationships in von Willebrand disease. I was appointed lecturer at the University of Sheffield in 1992 and became a senior lecturer in 2001. My research is aimed at improving understanding of the pathogenesis of inherited haemostatic disorders. Current areas of interest include the identification and characterisation of novel genetic defects underlying inherited platelet bleeding disorders and inherited thrombocytopenia. I was one of the chief investigators in a study which identified and characterised the underlying genetic defects in a UK cohort of patients with Glanzmann's thrombasthenia, and I was one of the original investigators on the British Heart Foundation funded Genotyping and Phenotyping of Platelets (GAPP) study which is combining extensive phenotype analysis with the power of whole exome sequencing to identify underlying genetic defects in patients with platelet bleeding disorders. This has led to the identification of a subgroup of patients with defects in the transcription factors FLI1 and RUNX1 whose platelets display abnormal secretion. I am also interested in the contribution of platelet gene variation to the bleeding tendency in patients with type 1 von Willebrand factor.

Cedric Ghevaert, PhD

Dr Ghevaert is a senior Lecturer in Transfusion Medicine at the University of Cambridge and Consultant Haematologist for the NHS Blood and Transplant. He did his PhD in Cambridge to develop recombinant antibodies for the treatment of fetomaternal alloimmune thrombocytopenia which culminated in a first-in-man study. Upon obtaining his PhD, Dr Ghevaert obtained a personal fellowship from the British Heart Foundation to work on megakaryopoiesis in the context of myeloproliferative diseases. His current group focuses its work on the production of blood cells from pluripotent stem cells with the declared aim to produce novel cellular therapies for transfusion to patients. His approach combines the manipulation of key transcriptional regulators to efficiently forward programme stem cells into mature blood cells and 3D bioengineered scaffolds to recreate the bone marrow niche and increase the production efficiency and purity of the manufactured blood cells. He still has a keen interest in inherited platelet disorders (such as Thrombocytopenia with Absent Radii) using the pluripotent stem cell technology for disease modelling. His expertise lies at the hinge of basic bench-based science and translational studies and as such will be supervising a clinical trial of in vitro-produced red cells in human volunteers in 2017.

Anne Goodeve, PhD

Having started her scientific career in microbiology, Anne undertook a PhD on influenza virus genetics. This was followed by a five-year postdoc at the Paterson Institute in Manchester examining the role of Epstein-Barr virus in nasopharyngeal carcinoma, which provided an excellent introduction into molecular genetics.

This provided useful expertise to enable Anne to secure a post as an NHS Clinical Scientist in Sheffield from 1990. This has taken Anne from a laboratory of 2 people undertaking genetic analysis on haemophilia and protein C/S to one of 110 staff examining many aspects of inherited and acquired genetic disorders at Sheffield Diagnostic Genetics Service. Anne spends half her time leading “The Haems” service that provides genetic analysis for inherited coagulation, thalassaemia and sickle cell disease, haemochromatosis, Fanconi anaemia and bone marrow failure syndromes. Next Generation DNA sequencing panels are used in many aspects of service provision.

From late 1990, when Prof Ian Peake moved to Sheffield, Anne became part of a growing Haemostasis Research Group at Sheffield University. The group’s main interests have been in inherited bleeding and thrombotic disorders and Anne’s main interest for many years has been in gaining better understanding of the genetics of von Willebrand disease. Along with Ian Peake and Francesco Rodeghiero (Vicenza), Anne had a major contribution to the EU funded project; Molecular and Clinical Markers for the Diagnosis and Management of Type 1 von Willebrand Disease. This subsequently led to a ten-year collaboration with Bob Montgomery and colleagues in Milwaukee and David Lillicrap and colleagues in Kingston, Ontario, through the

Zimmerman Program on the Molecular and Clinical Biology of von Willebrand Disease. She also leads the genetic analysis of von Willebrand factor in the 3WINTERS-IPS study examining genetics and treatment required for patients with type 3 von Willebrand disease (PI Augusto Federici, Milan).

Paul Harrison, PhD

Paul Harrison attained his PhD in Biochemistry from the University of London in 1988. Since then, his scientific research has centred on haemostasis and thrombosis with particular emphasis on the study of platelets, platelet function and extracellular vesicles. In the past he has worked at St. Thomas's Hospital Haemophilia Centre, The University of Oklahoma, the Haemostasis Research Unit at University College London and at the Oxford Haemophilia and Thrombosis Centre. He is currently the Healing Foundation Senior Lecturer within the School of Immunity and Infection at the University of Birmingham in the UK. Paul has authored many original scientific papers (h index =45) and recent state of the art book chapters, reviews and is a co-editor of a book entitled "Extracellular Vesicles in Health and Disease". He is a Fellow of the Royal Society of Pathologists, past-President/Secretary of the British Society of Haemostasis and Thrombosis and past-chairman of the Platelet Physiology scientific subcommittee (SSC) of the ISTH. Paul is also the current co-editor in chief of Platelets and on the editorial board of the Journal of Extracellular Vesicles, Journal of Thrombosis and Haemostasis and the International Journal of Laboratory Haematology. Paul was the recipient of the Sysmex Outstanding Science Award in 2009 and in 2011 was featured as one of the top 100 healthcare scientists in the UK by the Department of Health.

J.W.M. Heemskerk, PhD

Prof. Dr. Johan Heemskerk studied chemistry and biology at the Radbout University of Nijmegen. He carried out postdoctoral studies in Hamburg (Germany), Cambridge (United Kingdom) and Helsinki (Finland). In 1986, joined the Department of Biochemistry of Maastricht University to study effects of dietary fats on blood platelet activation. Prof. Heemskerk takes the chair of Cell Biochemistry in Thrombosis and Haemostasis, and is a Principal Investigator at CARIM. His laboratory with affiliated microscopic imaging facilities researches in the fields of blood cell function, cell signaling and coagulation in relation to health and disease. Prof. Heemskerk is a member of several Scientific Advisory Boards of Foundations and Institutes in the field of thrombosis and hemostasis. He is associate editor of the Journal of Thrombosis and Haemostasis, and a recipient of the BACH award of the International Society on Thrombosis and Haemostasis. He authored or co-authored over 250 full scientific papers in the areas of cell signalling, experimental thrombosis and haemostasis. His team participates in various European networks and consortia on genetics, platelet function and structure. His current research focuses are: (1) unravelling the interaction mechanisms between platelet activation and blood coagulation; (2)

the short- and long-term processes underlying thrombus buildup under flow conditions; and (3) the development and employment of multiparameter assays of platelet function to characterize the haemostatic state of patients and mice as a function of the genetic constitution.

Paula James, MD, FRCPC

A graduate of the University of Saskatchewan (MD, 1996), Dr. James joined the Queen's University faculty as an Assistant Professor in the Department of Medicine in 2004. She was promoted to Associate Professor in 2008 and full Professor in 2015. She holds cross-appointments to the Department of Pathology and Molecular Medicine and Pediatrics at Queen's.

Dr. James is a Clinician Scientist with an internationally recognized research program focused on the molecular genetic basis of von Willebrand disease and the clinical impact of this common, inherited bleeding disorder. She has published many peer-reviewed manuscripts in her field, and has held multiple national and international leadership roles including Co-Chair of the VWF (von Willebrand Factor) Scientific and Standardization Committee of the International Society on Thrombosis and Haemostasis and the Chair of the Research Advisory Committee of the Canadian Hemophilia Society.

Current areas of focus include evaluating the role of VWF in angiodysplasia, understanding abnormal bleeding in hemophilia carriers and translating previous work on quantitative assessments of bleeding into the general community to increase the understanding about normal and abnormal bleeding. A major objective is to accurately diagnose and manage patients with undiagnosed mild bleeding disorders.

Additionally, Dr. James is a well-respected clinician and the Medical Director of the Inherited Bleeding Disorders Clinic of Southeastern Ontario and its aligned Women and Bleeding Disorders Clinic.

Will Lester, MBChB(hons), BSc, FRCP, FRCPath, PhD

Haematology Consultant at University Hospital Birmingham, Queen Elizabeth and Birmingham Women's Hospital and Honorary Senior Clinical Lecturer in the School of Clinical and Experimental Medicine, University of Birmingham. Special Interests include thrombosis and haemostasis, obstetric and general haematology. Lecturer at the National Centre for Anticoagulant Training and has received two thrombus innovation awards. Member of BSH haemostasis and thrombosis task force and NEQAS blood coagulation steering committee

Gill Lowe, MBBS, PhD

Dr Gill Lowe is a Hematology Consultant at University Hospitals Birmingham. She specializes in disorders of haemostasis and thrombosis.

She studied at the University of Cambridge then undertook general medical placements in Norfolk, Newcastle and Manchester before moving to Birmingham in 2006 to start hematology specialist training.

She was a clinical research fellow at the University of Birmingham between 2010 and 2013 when she was the recipient of a Wellcome Trust Combined Training Fellowship, working in the laboratory of Professor Steve Watson. She worked on a project that looked at patients with excessive bleeding and whether they had an underlying inherited platelet defect, leading to several publications and to the award of a PhD in 2015.

She is the non-malignant hematology specialty group lead for the West Midlands Local Comprehensive Research Network, and has an interest in clinical research within the field of haemostasis and thrombosis. She has organized several conferences and educational meetings, including events targeted specifically at doctors in training who are interested in haemostasis and thrombosis.

Michael Makris, MD

Michael Makris is Director of the Sheffield Haemophilia and Thrombosis Centre, Sheffield, UK. He trained in Medicine and Haematology at the Universities of Oxford, London and Sheffield in the UK. He is Professor of Haemostasis and Thrombosis at the University of Sheffield and since 1994 he has been an honorary consultant haematologist at The Sheffield Teaching Hospitals NHS Trust in Sheffield, UK.

Professor Makris's research interests include the monitoring of adverse events in haemophilia, hepatitis C in haemophilia, thrombin generation assays in bleeding and thrombotic disorders and the genetics of inherited platelet disorders. He is one of two editors of the journal *Haemophilia and* is on the Editorial Board of the journals *British Journal of Haematology, Journal of Thrombosis and Haemostasis and Blood Transfusion.*

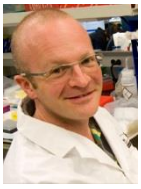
Neil Morgan, PhD

Dr Neil Morgan is a non-clinical lecturer in Cardiovascular Genetics at the University of Birmingham, UK.

He has published over 75 research papers in high impact scientific journals in the field of human genetics. His research has primarily involved the identification and characterisation of novel genes for inherited diseases in the haematopoietic system.
His current research is focusing on gene identification in patients with inherited bleeding disorders and low platelet counts (thrombocytopenia). He utilises next generation sequencing technologies followed by investigation into the platelet and megakaryocyte biology of novel disease genes mutated in disorders with inherited bleeding.

He hopes that the identification of novel gene defects will provide clues to genes and proteins involved in inherited platelet disorders which will ultimately lead to devising new treatments to minimise the risk of bleeding in such patients.

Andrew Paterson, MD



Andrew is a Senior Scientist in the Program in Genetics and Genome Biology at The Hospital for Sick Children Research Institute in Toronto. and Professor in the Epidemiology and Biostatistics Divisions at the Dalla Lana School of Public Health and Institute of Medical Sciences at the University of Toronto. From 2002-2012 he held a Tier II Canada Research Chair in Genetics of Complex Diseases.

His scientific interest is concentrated on the genetics of human diseases. Specifically, he is the lead investigator on large study to investigate the genetic determinants of risk for long-term complications of type 1 diabetes, including retinopathy and nephropathy, as well as their risk factors. He has also worked on a number of other traits, including bleeding disorders, autistic traits, polycystic kidney disease, as well as measures of eye sight, heart rate, breast density and blood pressure. In the field of hemostasis and thrombosis he led the identification of the mutation and gene that is responsible for Quebec Platelet Disorder, and has contributed to work on the genetics of von Willebrand factor/disease. Since 2001 he has been director and co-director of the Statistical Analysis Facility of The Centre for Applied Genomics, a Canadian Genome Centre based at The Hospital for Sick Children where he has directed genome-wide linkage, association, sequencing, expression and epigenetic studies of both rare simple diseases and complex diseases. In particular he is focused in combining traditional genome-wide linkage approaches with next-generation sequencing (exome, genome) to identify causal variants for rare Mendelian diseases.

He has published over 210 papers in various scientific journals. He dreams of using genetic information to improve public health.

David Rabbolini, BSc MBBCh, FRACP, FRCPA

Dr. Rabbolini is a haematologist at The Royal North Shore Hospital in Sydney, Australia. His interest is in thrombosis and haemostasis. He is currently completing a PhD at the Northern Blood Research Centre, University of Sydney. His research focus is on the utilization of induced pluripotent cells for the investigation of inherited thrombocytopenia caused by mutations in hematopoietic transcription factors, as well as the use of next generation sequencing platforms for the investigation of these disorders. He is spearheading a Sydney platelet co-operative group whose aim is to improve the diagnosis of inherited platelet disorders in the region.

Dr. Rabbolini completed medical school at the University of the Witwatersrand, South Africa. His post-graduate training was undertaken in Australia where he was awarded fellowships with the Royal Australasian College of Physicians and Royal College of Pathologists of Australasia. He holds memberships with professional societies including the International Society of Thrombosis and Haemostasis and the Australasian Society of Thrombosis and Haemostasis.

Dr. Rabbolini has a keen interest in medical education. He is a clinical lecturer at the University of Sydney and mentors junior physician trainees and medical students.

Suthesh Sivapalaratnam, MD

Dr. Suthesh Sivapalaratnam studied Medicine at the University of Amsterdam. Hereafter he ventured into a combined PhD fellowship between the University of Cambridge (supervisor: Prof. W. H. Ouwehand) and the University of Amsterdam (Supervisor: Prof. M. Levi). In 2012 he successfully defended his thesis “The Molecular Basis of Early onset Cardiovascular Disease”. He subsequently core medical training at the Academic Medical Center in Amsterdam. During this period he completed a Post-doc at the Center for Human Genetic Research at the Massachusetts General Hospital Boston and the Broad in Cambridge. In 2015 he rejoined the Ouwehand Group, where he still works as a Post Doctoral fellow in Rare Bleeding and Platelet Disorders. Since February 2016 he combines this with Specialist Registrar training in Haematology at the Barts Rotation in London as an Academic Clinical Fellow. This year he is one of the recipients of the TRTH EHA ASH Fellows. Next to work he has a passion for good food and aspires to one day do a (mini) triathlon.

Julie Vogt

I was born and grew up in London. After studying medicine at St Thomas’ Hospital Medical School I trained and was accredited in paediatrics. During my registrar rotation at Great Ormond Street Hospital caring for patients undergoing bone

marrow transplantation for primary immunodeficiencies and haematological malignancies I developed a special interest in genetics. I moved to the West Midlands to gain experience and train in clinical genetics and became a Consultant Clinical Geneticist in the West Midlands Regional Genetics Service in 2009.

Within the West Midlands, Birmingham continues to have a consistently high rate of stillbirth and infant mortality. Pakistani families have significantly higher mortality rates from metabolic disorders. Currently I am the clinical genetics paediatric neurometabolic /neuromuscular service lead. I work together with clinicians from the Birmingham Children's Hospital to provide a multi-disciplinary model of care for children with these often serious life-limiting conditions.

I also work with surgical colleagues to provide an Upper Limb Service and a multi-disciplinary service for families with arthrogryposis (multiple joint contractures). The arthogryposis service links in well with my research study into the molecular basis of the multiple pterygium syndromes and fetal akinesia deformation sequence. We have identified and published novel causative genes and have validated the clinical exome methodology for use in the clinical laboratory which has applications for a wider spectrum of joint contracture conditions.

The clinical service has seen translation of the recent advances in genetic testing into the diagnostic pathway for patients. I have been recruiting families into the national exome sequencing study, Deciphering Developmental Disorders, and the 100, 000 Genomes Project.

My role also involves teaching and training of undergraduate and post graduate students and other allied health professions, and I have taught on the Birmingham University MSc in Genomics module.

Tim Warner, PhD

Tim Warner studied for his BSc in Pharmacology at Chelsea College, later King's College London, from 1982 to 1986. This course included an extramural year, during which time Tim worked in the cardiovascular research group at Smith Kline French in Welwyn, conducting experiments on combined alpha- and beta-receptor blockers. After graduating with a first class degree in 1986, Tim went to study for his Ph.D. under the supervision of Sir John Vane at the newly opened William Harvey Research Institute. Tim's PhD project was on the processes controlling the release of mediators from endothelial cells. For his post-doctoral research Tim moved to work with Prof. Ferid Murad, who at that time was well known as a leading biochemical pharmacologist who had made fundamental observations on the regulation of guanylyl cyclase; notably in the 1970s that nitric oxide stimulated guanylyl cyclase to cause the production of cyclic GMP. After gaining invaluable biochemical experience Tim returned in 1992 to the William Harvey Research Institute. Tim has been there ever since and after being a British Heart Foundation lecturer (1995-2000) is now Professor of Vascular Inflammation, Deputy Dean for Research for the School of Medicine and Dentistry, and Deputy Director (Research) QMUL Life Sciences Initiative.

Tim has acted as an editor for the British Journal of Pharmacology and Platelets, and in 2000 was the recipient of the Novartis Prize of the British Pharmacological Society. In his research career Tim has published more than 300 research papers, reviews, articles and abstracts. The impact of these articles places Tim within the top 0.5% of cited researchers over the last 20 years (Institute for Scientific Information). Tim is a fellow of the British Pharmacological Society as well as the American Heart Association and the Royal Society of Biology. He is also a member of the International Society for Thrombosis and Haemostasis, the American Society for Pharmacology and Experimental Therapeutics, the Working Group on Thrombosis of the European Society of Cardiology, and the British Society for Cardiovascular Research.

Stephen Watson, PhD

Dr. Stephen Watson is a Professor at the University of Birmingham in Birmingham, England. He holds a PhD in Neurochemical Pharmacology from the University of Cambridge and a degree in Pharmacology from the University of Leeds where he graduated with first class honors. Dr. Watson's research currently focuses on the study of surface receptors and their signaling pathways in platelets and megakaryocytes, and the study of patients with platelet bleeding disorders. He has published over 220 primary research papers in over 40 different journals. He has also served as a Senior Editor on several scientific journals including Trends Pharmacological Sciences (2001); Platelets (2005); JTH (2007-13); Thrombosis and Haemostasis (2008-12); JBC (2009-13) and Biochemical Journal (2010-13). Dr. Watson was elected in 2014 to the International Society on Haemostasis' (ISTH) Council for a six year term. He also serves on ISTH's Nominating and Education and Outreach Committees. Awards and recognitions received by Dr. Watson include Fellow of the Academy of Medical Sciences, Nature/NESTA mid-career award for creative mentoring in science and ISTH Investigator Recognition Award for Contributions to Haemostasis.