CIRM Scientific and Medical Research Funding Working Group
Biographical information of candidates nominated to serve as Scientific Members of the Working Group

Vania Broccoli, PhD

Dr. Broccoli is Head of Research Unit in the Division of Neuroscience at the San Raffaele Scientific Institute in Milan, Italy. He earned his BSc in Biological Sciences at the University of Bologna, his PhD in Neuroscience at the University of Padua, and completed a postdoctoral fellowship in Neurogenetics at the Helmholtz Center in Munich, Germany. Dr. Broccoli was Staff Scientist at the Telethon Institute for Genetics and Medicine before moving to the Stem Cell Research Institute at the San Raffaele Scientific Institute as Staff Scientist. He was promoted to Group Leader at the Stem Cell Research Institute before becoming Head of Research Unit.

Since joining the San Raffaele Scientific Institute as a tenure track scientist, Dr. Broccoli has been working in unraveling the molecular mechanisms governing forebrain development and functionally characterizing the transcription factors Arx, Tbr2, Tbr1, FoxG1, MeCP2 involved in neurodevelopmental pathologies in humans. In a long-term collaboration with Prof. J. Rubenstein's lab at the University of California San Francisco, Dr. Broccoli's group showed that Tbr2 is essential for determining intermediate cortical progenitors, providing the first experimental evidences that these neural progenitors are the founders of most cortical neurons. These data illuminated the biological origins of Tbr2 dependent microcephaly in humans. His own research group aims to translate this knowledge in the stem cell research field contributing to develop protocols and methods for generating sub-type specific telencephalic neurons through in vitro differentiation of embryonic and neural stem cells.

Lately, his group employed lineage specific transcription factors for reprogramming cellular identity and generating therapeutic relevant neuronal subtypes from conversion of skin fibroblasts. Initially, he established genetic cell reprogramming for generating iPS cells with the aim to model human diseases like Alzheimer's and Parkinson's disease and atypical Rett syndrome. Then, his group identified a minimal combination of three transcription factors (Mash1, Nurr1 and Lmx1a) able to directly convert mouse and human fibroblasts into functional dopaminergic neurons (Caiazzo et al., 2011). This discovery allows for the straightforward production of a homogenous source of human functional dopaminergic neurons amenable for a cellular replacement therapy in Parkinson's disease. His lab is currently pursuing new procedures of direct cell reprogramming for generating other neuronal cell types with a therapeutic prospective.

Ulrich Broeckel, MD

Dr. Broeckel is Professor of Pediatrics, Adjunct Professor of Medicine and Physiology, Chief of the Section of Genomics Pediatrics, Associate Director of the Children's Research Institute, and Director of the Individualized Medicine Institute at the Medical College of Wisconsin. He earned his MD from the University of

Heidelberg and completed a residency and internship in the Department of Internal Medicine II: Cardiology, Nephrology and Pulmonary Medicine at the University of Regensburg. Dr. Broeckel was Research Fellow at the University of Ulm before moving to the Medical College of Wisconsin, initially as a postdoctoral fellow in the Laboratory of Genetics Research, where he rose through the ranks to his current position.

Dr. Broeckel's laboratory specializes in the identification and functional evaluation of genes and their variants involved in cardiovascular and other complex diseases. His research interest include: left ventricular hypertrophy, myocardial infarction, coronary artery disease, end-stage renal disease, and hypertension with projects based on large epidemiological studies in clinical cohorts. In addition, his laboratory performs microarray-based diagnostic tests with an emphasis in pharmacogenomics for both Children's Hospital of Wisconsin (CHW) and St. Jude Children's Research Hospital and runs the Nucleic Acid Extraction Core for investigators at Children's Research Institute. Dr. Broeckel's laboratory is also a part of a groundbreaking multicenter National Heart, Lung, and Blood Institute (NHLBI) initiative to generate patient-derived human induced pluripotent stem cells (hiPSC) for the study of complex disease. This collaboration will result in the high-throughput development of hiPSC-derived cardiomyocytes generated from patients participating in a major hypertension epidemiological study.

Dr. Broeckel is a member of several professional and honorary societies, including the American Heart Association (AHA), the American Association for the Advancement of Science (AAAS), and the American Pediatric Society. His research is supported by grants from the National Institutes of Health (NIH) and he is an experienced reviewer for the NHLBI. Dr. Broeckel has published more than 60 articles throughout his career

Daniel A. Doherty, MD, PhD
Dr. Doherty is Associate Professor of Pediatrics and Research Affiliate at the Center on Human Development and Disability at the University of Washington. His research interests focus on hindbrain malformations as a way to understand human brain development and common disorders such as intellectual disability, autism, ataxic cerebral palsy and even mental health disorders such as schizophrenia.

The hindbrain regulates basic functions (level of consciousness, heart rate, respiratory rate), coordinates balance, limb and eye movements, as well as having possible roles in cognition and emotional regulation. Dr. Doherty's group uses a variety of genetic techniques (SNP mapping, array CGH, and high throughput sequencing) to identify the genes responsible for hindbrain malformation disorders such as Disorders of Hindbrain Development. Identifying the genes responsible for a disorder immediately translates into molecular diagnostic testing, and detailed work on genotype-phenotype correlations improves diagnostic, prognostic and medical management information for patients. In addition, Dr. Doherty's group and his collaborators use the disease genes to dissect the molecular mechanisms of

normal and abnormal brain development in vitro and in animal models. The human hindbrain is a fascinating system in which to study the role of basic developmental processes (spatially restricted gene expression to define positional information, organizing centers, morphogenetic movements, cell-cell and long-range signaling, cell migration and axon guidance) in human disease.

Dr. Doherty's clinical interests complement the research interests of his group. He cares for children with all types of central nervous system abnormalities including Disorders of Hindbrain Development, agenesis of the corpus callosum, cortical malformations, hydrocephalus, spina bifida, intellectual disability and cerebral palsy. He also provides prenatal counseling to women carrying fetuses with abnormal CNS imaging findings to provide a pediatric perspective on these conditions.

Meri Firpo, PhD

Dr. Firpo is an Assistant Professor in the Stem Cell Institute and the Department of Medicine at the University of Minnesota, where she works on human pluripotent stem cell biology, and transplantation therapies for diabetes. She received her PhD from the Cornell University Medical College Graduate School of Medical Sciences, and did postdoctoral fellowships at the National Jewish Institute for Immunology and Respiratory Medicine in Denver, Colorado, and the DNAX Research Institute for Molecular and Cellular Biology in Palo Alto, California. She was previously at the University of California San Francisco before joining the UMN Stem Cell Institute and Schulze Diabetes Institute.

The focus of Dr. Firpo's laboratory is the regulation of stem cell growth, both from the perspective of the expansion of stem cells in the lab, and the regulated differentiation to functional tissues. The overall goal of her research is to understand the regulation of human development. The research can be divided into two main directions: the first involves improvements in the derivation and propagation of human embryonic stem cells (hESCs); the second is the development of stem cell-based therapies for diabetes.

Bruce D. Gelb, MD

Dr. Gelb is the Director and Gogel Family Professor of the Child Health and Development Institute at the Mount Sinai School of Medicine. He is Professor of Pediatrics and of Genetics and Genomic Sciences. Dr. Gelb completed a pediatric residency and pediatric cardiology fellowship at Babies Hospital of Columbia-Presbyterian Medical Center and Texas Children's Hospital at the Baylor College of Medicine, respectively. He joined the faculty at Mount Sinai in 1991 and has remained there since.

Dr. Gelb has developed an extensive program in genomics/gene discovery for congenital heart disease. His group is best known for their work on Noonan syndrome and related disorders (now termed Rasopathies). They have ongoing work in gene discovery, animal modeling, and therapy development. The Gelb group

published the first model of cardiovascular disease using human induced pluripotent stem cells, using fibroblasts from patients with LEOPARD syndrome. They continue to use human iPS cells to model cardiovascular disease. In addition to his research, he directs the Cardiovascular Genetics Program at Mount Sinai.

Dr. Gelb has received the E. Mead Johnson Award from the Society for Pediatric Research and the Norman J. Siegel New Member Outstanding Science Award from the American Pediatric Society. He was elected to the American Society of Clinical Investigation and the Institute of Medicine. Dr. Gelb is a member of the Federal Advisory Committee for the National Children's Study and the American Board of Pediatrics, Pediatric Cardiology Subboard.

Derek J. Hei, PhD
Dr. Hei is the Director of Waisman Biomanufacturing. He received his PhD from the University of California Berkeley. Dr. Hei joined the University of Wisconsin-Madison in January of 2000 as the technical director of Waisman Biomanufacturing – a state-of-the-art cleanroom facility designed for the development and production of cutting-edge biotherapeutics for human clinical trials with university and private industry partners. Dr. Hei is currently the PI for the National Institutes of Health (NIH)-funded PACT (Product Assistance for Cellular Therapy) Program. He also previously served as PI for the NIH National Stem-Cell Bank contract with NCRR and the NIAMS contract "Development of a Production Facility for NIH 3T3 Cells".

Waisman Biomanufacturing is designed to manufacture cell and gene therapeutics for early stage (Phase I and Phase II) human clinical trials according to the FDA's cGMP guidelines. Waisman Biomanufacturing is capable of providing development and manufacturing services for a wide range of biotherapeutics. In addition to facilities and equipment for producing plasmid DNA and viral vectors for gene therapy, Waisman Biomanufacturing has facilities for processing cell therapeutics including embryonic and neural stem cells, filling of parenteral therapeutics, and producing recombinant proteins. This range of capabilities allows Waisman Biomanufacturing to provide support to both academic and commercial groups that are involved in the development of complex, multi-component biotherapeutics such as engineered stem cells.

Waisman Biomanufacturing has been selected as one of five facilities nation-wide to serve as a Cell Processing Facility for the PACT program. The PACT program is funded by the National Heart, Lung, and Blood Institute (NHLBI) with a primary mission of supporting translational investigators that are involved in moving promising new cell therapeutics into human clinical trials. The UW PACT team has a primary focus of Mesenchymal Stem Cell (MSC) and human Embryonic/Pluripotent Stem Cell-based therapeutics. Through the PACT program, NHLBI also provides support for translational efforts in other areas outside of the normal scope of NHLBI funding.

Darrell N. Kotton, MD

Dr. Kotton is Co-Director of the Boston University Center for Regenerative Medicine (CReM), and Associate Professor of Medicine, Pathology and Laboratory Medicine at Boston University School of Medicine. He received his BA degree from the University of Pennsylvania, and studied guitar performance at the Berklee College of Music in Boston, before attending medical school at Washington University School of Medicine, where he received his MD in 1994. He completed an internship and residency in Internal Medicine at the University of Pennsylvania from 1994-97 and then spent 1 year working in medical schools in India, Uganda, and Malawi, before returning to the US for fellowships in Pulmonary and Critical Care Medicine at Boston University/Boston Medical Center. Dr. Kotton completed a post-doctoral research fellowship in the laboratory of Richard C. Mulligan, PhD at the Harvard Medical School, Department of Genetics, before returning to Boston University School of Medicine as faculty in 2002.

Dr. Kotton's research focuses on basic stem cell biology with the ultimate goal of developing new therapies for lung diseases. His laboratory and Center have pioneered new technologies for gene transfer and reprogramming in order to derive novel pluripotent stem cell populations for regenerative medicine applications.

In addition to caring for patients with lung disease and critical illness, Dr. Kotton's laboratory continues to train students and young investigators and to publish seminal works advancing our understanding of lung disease and potential new lung gene- and cell-based therapies. In recognition of his accomplishments, he was named Co-Director of Boston University's new Center for Regenerative Medicine in 2009 and was elected to the American Society for Clinical Investigation in 2010. His Center's most well known publications to date include the first derivation of 'lung disease-specific stem cells', the engineering of a novel tool able to reprogram skin cells into stem cells, and the generation of a bioartificial lung.

Joseph Laning, PhD

Dr. Laning is the Senior Director of the Massachusetts Stem Cell Bank and Registry and Research Associate Professor of Molecular Medicine at the University of Massachusetts Medical School. Dr. Laning received his BS degree in Biology from Boston University (1986) and his PhD in Immunology from Harvard University (1995). He is an active member of the International Society for Stem Cell Research and a member of the Coalition for the Advancement of Medical Research.

Dr. Laning has spent the past 17 years seeking to translate concepts into products in the fields of wound care, regenerative medicine, and stem cell therapy. He began his post-doctoral career at Organogenesis, Inc. where he developed and implemented pre-clinical investigations and subsequently managed all patient immunology safety testing leading to the approval of a Premarket Approval Application (PMA) of ApligrafTM with the US Food and Drug Administration (FDA). In 2002, he joined ViaCell, Inc. where he served as Director of Therapeutic Development and subsequently Senior Director of Analytical Biology. In these roles he oversaw strategic and operational scientific plans leading to successful approval of both

Investigational New Drug (IND) applications and Investigational Device Exemption (IDE) FDA filings and completion of the company's cell therapy clinical trial in allogeneic cord blood stem cell transplantation. He subsequently brought his extensive experience in scientific management and product development to scientific and business consulting opportunities in industry and academics as Founder and Principal of JCLaning BioConsulting.

Dr. Laning's current research interests include developing enhanced methods for human induced pluripotent stem (iPS) cell derivation and characterization standards and studying the differentiation utility of adult stem cells as potential universal components for disease modeling and pharmaceutical screening.

Jacob L. McCauley, PhD
Dr. McCauley is Assistant Professor at the University of Miami Miller School of Medicine in the Dr. John T. Macdonald Foundation Department of Human Genetics. He is also the Director of the Biorepository facility at the John P. Hussman Institute for Human Genomics. Dr. McCauley is a graduate of Bethany College in Bethany, WV (BS, 2000) and Vanderbilt University (PhD, 2005) in Nashville, TN. Following his PhD training in molecular genetics, he completed a postdoctoral fellowship in genetic epidemiology and then took a faculty position at the University of Miami. Dr. McCauley's PhD dissertation focused on understanding the genetic mechanisms involved in autism, with his postdoctoral training focused on the genetic aspects of both Alzheimer's disease and multiple sclerosis.

Dr. McCauley is a genetic epidemiologist with a background comprised of training in both molecular and statistical genetic techniques. His primary interest is to improve the understanding of human disease through disease gene discovery, genomics, and in-depth examination of environmental factors that influence disease outcome. His current research focuses on the use of molecular techniques, bioinformatics, and statistical methods to identify genetic variation and to characterize its role in disease susceptibility within a variety of diseases including multiple sclerosis and inflammatory bowel disease (IBD). Dr. McCauley's role as Director of a Biorepository facility has multiple administrative responsibilities (e.g. research and development, staffing, budget management). This role has also provided him the opportunity to consult and direct biological sample collections, oversee biological sample tracking, quality control procedures, genotyping, and genetic analyses involved in large-scale consortia projects. Combined these efforts have aided in a number of discoveries within the genetics of complex disease phenotypes.

Dr. McCauley has been an active member of the International Multiple Sclerosis Genetics Consortium (IMSGC) for the last six years, the Biorepository Core Director of the Ethnic/Racial Variations of Intracerebral Hemorrhage (ERICH) study led by Dr. Daniel Woo at the University of Cincinnati over the last two years, an Investigator of a previous Formative Research study for the National Children's Study (NCS), and a Co-Investigator for the BioResource Component of a recently funded University of Miami Clinical and Translational Science Institute (CTSI)

application. Dr. McCauley has been the recipient of a National Alliance for Autism Research predoctoral fellowship, a National Multiple Sclerosis Society postdoctoral fellowship, and the Stanley J. Glaser Foundation Research Award at the University of Miami.

Aarno Palotie, MD, PhD

Dr. Palotie has been a member of the Human Genetics Faculty at the Wellcome Trust Sanger Institute since 2007. He also holds a position at the Finnish Institute for Molecular Medicine (FIMM) in Helsinki and has been a visiting faculty member at the Broad Institute of MIT and Harvard since 2004. Dr. Palotie is the chair of the International Headache Genetics Consortium and the co-chair of the neurodevelopmental arm of the UK10K project, which aims to sequence the exome of 3000 schizophrenia and autism cases. He received his MD and PhD degrees at the University of Oulu, Finland. He served his residency in laboratory medicine and gained his specialty in Clinical Pathology at the University of Helsinki. After his residency, as Professor of Cell and Molecular Biology at the University of Helsinki, he founded and ran the diagnostic laboratory for molecular genetics. From 1998 to 2002 he was Professor of Pathology at the University of California School Of Medicine, Los Angeles. He then became the Director of the Finnish Genome Centre at the University of Helsinki from 2002 until 2008.

Dr. Palotie has a long history in research in the genetics of Mendelian and complex traits. Currently his group is interested in the genetics of neurological and neurodevelopmental traits, especially migraine, schizophrenia, autism and epilepsy. Projects are based on the latest high throughput genotyping and sequencing strategies and analysis methods. The wealth of multiple large study samples has also enabled Aarno's group to use different study designs for genome variant identification, verification and effect size estimation. The group recently identified the first gene variant associated with common forms of migraine.

One of the other aims of the group is to build towards a more comprehensive understanding of the genomic landscape of such common diseases using the unique opportunities provided by the Finnish population, the Finnish health care infrastructure and large national sample collections. These unique resources have stimulated several large whole exome and whole genome sequencing projects organized as part of a collaborative initiative, SISu (Sequencing Initiative Suomi) which includes researchers from FIMM, THL, Lund University, the Broad Institute of MIT and Harvard, Michigan University, UCLA, NIH, Oxford University and the Wellcome Trust Sanger Institute. During 2012 the SISu project will produce the complete genome or exome sequence of thousands of Finns. The combination of these data with the existing genome wide association study (GWAS) data from more than 47 000 Finns provide a rich resource to facilitate a more comprehensive understanding of the genome landscape associated with diseases that are major health burdens in the population. This improved knowledge will provide new tools for the development of more individualized health care.

Ludovic Vallier, PhD
Dr. Vallier is a Medical Research Council (MRC) senior-non clinical fellow and Reader in Stem Cells and Regenerative Medicine affiliated with the Department of Surgery Cambridge University, Principal Investigator at the Cambridge Stem Cell Institute and Director of the Cambridge Biomedical Research Centre hiPSCs core facility. He also recently obtained a joint appointment as Senior Group Leader at the Wellcome Trust Sanger Institute where he intends to lead a program on pancreas genetics. Finally, he is also the co-funder of the biotechnology company DefiniGEN, which is producing hepatocytes and pancreatic cells for drug and toxicology screening. Dr. Vallier received his Magister in Cellular and Molecular Biology and PhD in Cell Biology from ENS Lyon.

Dr. Vallier has developed a strong expertise in human embryonic stem cells and human induced pluripotent stem cells by discovering key mechanisms controlling their differentiation and pluripotency. The current objective of Dr. Vallier's research is to define the molecular mechanisms controlling the specification of the endoderm germ layer from which key organs such as the pancreas and the liver are derived. For that, his group uses human pluripotent stem cells (hESCs and hiPSCs) as in vitro model of development. The resulting knowledge allows the development of new culture systems to drive differentiation of pluripotent stem cells into hepatocytes and pancreatic islet cells. The resulting cells can then be used to model in vitro metabolic disorders for basic studies and also drug screening. Furthermore, Dr. Vallier's group explores the potential of hepatic and pancreatic cells for cell based therapy approach. Overall, his objective is not only to differentiate human pluripotent stem cells (hESCs/hiPSCs) into cell types relevant for clinical applications but also to acquire the knowledge necessary to differentiate any cell types into pancreatic and hepatic progenitors.

Dr. Vallier's contributions to the stem cell field have generated over 30 original papers, 3 book chapters and 5 reviews on basic aspects of stem cell biology and resulted in numerous invitations to internationally recognized meetings such as the European association for the study of the liver, the European society for gene therapy and Keystones Symposia. Furthermore, he has been awarded the NC3Rs research price in 2011 for his work on disease modeling using hiPSC. Finally, he is recognized as a world wide expert in hiPSC derivation and differentiation through his activity in the BRC hiPSC core facility and more recently at the Wellcome Trust Sanger Institute.

Richard Wade-Martins, PhD
Dr. Wade-Martins heads the Laboratory of Molecular Neurodegeneration at the University of Oxford, focused on better understanding the molecular and genetic mechanisms of neurodegenerative diseases. He is also Director of the Oxford Parkinson's Disease Centre. The Centre joins together ten laboratories in Oxford and supports a full time staff of fourteen posts focused on understanding the very earliest molecular pathways to pathology in Parkinson's disease. Dr. Wade-Martins studied Natural Sciences (Genetics) at the University of Cambridge, followed by a

PhD at the Wellcome Trust Centre for Human Genetics, University of Oxford. He then held consecutive Wellcome Trust Research Fellowships at the University of Oxford, Harvard University and then again at Oxford. In 2008, he was appointed to a faculty position at the Department of Physiology, Anatomy and Genetics, University of Oxford.

Dr. Wade-Martins' career is focused on improving our understanding of the molecular and genetic mechanisms of neurodegenerative diseases with the aim one day of developing novel protective therapies for neurological disorders. In 2004, he returned from Harvard Medical School to the Wellcome Trust Centre for Human Genetics in Oxford and was awarded a Wellcome Trust Research Career Development Fellowship to study the molecular genetics of gene expression from the microtubule associated protein tau (MAPT or tau) genomic locus. His range of interests has expanded and his group now studies the molecular mechanisms of degeneration associated with tau, alpha-synuclein, leucine rich repeat kinase 2 (LRRK2), TDP-43, FUS and C9ORF72 using human post-mortem brain tissue, BAC transgenic mouse lines and in vitro neuronal culture systems. His research includes a major effort to develop iPS cell-derived dopaminergic neuronal culture models to study Parkinson's disease. Dr. Wade-Martins heads the "CNS: Neurodegenerative and neurodysfunctional diseases" theme in the new StemBANCC consortium, a large European Union-funded collaborative study between academia and industry to develop iPS cell models for common diseases.

Dr. Wade-Martins has held numerous highly competitive Wellcome Trust Fellowships and is a previous Research Into Ageing UK Young Investigator. He currently sits on the Scientific Advisory Board of Alzheimer's Research UK and the Research Advisory Panel for Parkinson's UK. He is Director of the Oxford Parkinson's Disease Centre and a Fellow of Christ Church College, Oxford.