

# SAB/MAB

## Scientific and Medical Advisory Boards

Distinguished professionals to help IRSF guide research from “bench to bedside”

### Overview

In May 2009 IRSF announced the appointment of two advisory boards that will serve to help set strategic goals for the advancement of research towards the development of therapeutics for Rett syndrome. The Scientific Advisory Board (SAB) and Medical Advisory Board (MAB) members consist of leading scientists and clinicians drawn from the Rett syndrome field, augmented by distinguished neuroscientists and prominent individuals with experience in drug discovery and development within academia and industry. Members of the SAB and MAB have been drawn from specific disciplines that are critical to facilitate the rapid translation of basic research findings into therapeutics for the treatment of Rett syndrome.

[Click here](#) to view the official press release

The Scientific Advisory Board (SAB) appointees to date include:

- Jeffrey D. Macklis, MD, D. HST, Harvard University
- Eric J. Nestler MD, PhD, Mount Sinai School of Medicine
- David M. Katz PhD, Case Western Reserve University
- Gail Mandel PhD, Vollum Institute, Oregon Health & Science University
- David Sweatt PhD, University of Alabama at Birmingham
- Jeanine M. LaSalle PhD, University of California Davis
- James H. Eubanks PhD, University of Toronto
- John M. McCall PhD, President and Founder, PharMac LLC
- Emer Leahy PhD, MBA, PsychoGenics Inc.
- Alan Kozikowski PhD, University of Illinois at Chicago
- Rajiv Ratan MD, PhD, Burke/Cornell Medical Research Institute
- Kim Hunter-Schaedle PhD, Children's Tumor Foundation

The Medical Advisory Board (MAB) appointees to date include:

- Alan K. Percy, MD Civitan International Research Center, University of Alabama at Birmingham

- Daniel G. Glaze, MD, Baylor College of Medicine Blue Bird Circle Rett Center
- Walter E. Kaufmann, MD, Kennedy Krieger Institute, Johns Hopkins University
- Shlomo Shinnar, MD, PhD, Montefiore Medical Center
- Bertram H. Lubin, MD, Children's Hospital Oakland Research Institute
- Eric Smeets MD, PhD University of Maastricht, Netherlands
- Jeffrey L. Neul, MD, PhD Texas Children's Hospital, Baylor College of Medicine
- Angus Clarke, DM, FRCP Cardiff University, Wales, UK
- N. Carolyn Schanen, MD, PhD, Alfred I. duPont Hospital for Children/Nemours Children's Clinic at the University of Delaware
- Peter Huppke, MD Georg August University Göttingen, Germany
- Alessandra Renieri MD, PhD University of Siena, Italy
- Roger Albin MD, University of Michigan Health System
- Evdokia Anagnostou, MD Bloorview Children's Rehab and Research Institute, University of Toronto
- Susan E. Swedo MD, National Institute of Mental Health (NIMH)

#### Scientific Advisory Board (SAB)

- Jeffrey D. Macklis, MD, D. HST, Harvard University

Dr. Macklis is Professor of Stem Cell and Regenerative Biology, Harvard University and Director of the newly founded Massachusetts General Hospital-Harvard Medical School Center for Nervous System Repair. He is also Program Head, Neuroscience/Nervous System Diseases at the Harvard Stem Cell Institute and Co-Director for the Regeneration and Repair Program of the Harvard Center for Neurodegeneration and Repair.

[Back to List](#)

- Eric J. Nestler MD, PhD, Mount Sinai School of Medicine

Dr. Nestler is a world renowned neuroscientist, molecular biologist, and psychiatrist who recently joined Mount Sinai School of Medicine as Director of the Mount Sinai Brain Institute and Chairman of the Department of Neuroscience. Dr. Nestler's work spans the broad areas of mind and brain research in both an academic and clinical setting. His research bridges basic biology and molecular neuroscience through to psychology and clinical psychiatry.

[Back to List](#)

- David M. Katz PhD, Case Western Reserve University

Dr. Katz is Professor of Neurosciences at Case Western Reserve University. Dr. Katz's laboratory focuses on understanding the role played by the neurotrophin family of growth factors in nervous system development. These findings may have important implications in terms of therapeutic development for Rett Syndrome.

[Back to List](#)

- Gail Mandel PhD, Vollum Institute, Oregon Health & Science University

Dr. Mandel is a Howard Hughes Medical Institute Investigator and Senior Scientist at the Vollum Institute, Oregon Health & Science University. Her laboratory studies the molecular mechanisms underlying regulation of gene expression in the developing nervous system through study of the DNA-binding protein REST. More recently, Dr. Mandel and her collaborators have uncovered a potential role for glial cells in neuronal dysfunction seen in Rett Syndrome, a process which may contribute to the underlying neuronal pathology observed in the disease.

[Back to List](#)

- David Sweatt PhD, University of Alabama at Birmingham

Dr. Sweatt is a highly accomplished researcher and an internationally recognized expert on the biological mechanisms underlying learning and memory. He is currently, Chair of the UAB Department of Neurobiology. Dr. Sweatt's research focuses on signal transduction mechanisms in learning and memory. He has received numerous honors and awards for his research and is on the editorial board of the Journal of Neuroscience and several other scientific journals.

[Back to List](#)

- Jeanine M. LaSalle PhD, University of California Davis

Dr. LaSalle is Professor of Microbiology and Immunology at the UC Davis School of Medicine and a member of the Graduate Groups of Genetics, Biophysics, Neuroscience, Biochemistry and Molecular Biology and a Member of the Rowe Program in Human Genetics at UC Davis. Dr. LaSalle's work focuses on the role of epigenetics in human neurodevelopmental disorders such as Rett syndrome, Prader-Willi syndrome, Angelman syndrome and autism-spectrum disorders.

[Back to List](#)

- James H. Eubanks PhD, University of Toronto

Dr. Eubanks is a Senior Scientist within the Division of Genetics and Development, at Toronto Western Research Institute and holds a concurrent Associate Professorship within the Department of Surgery at the University of Toronto. His laboratory is currently working to define how the loss of MeCP2 affects brain development in animal models of Rett Syndrome, and testing whether gene replacement strategies and/or targeted drug interventions ameliorate Rett-like symptoms in these animals.

[Back to List](#)

- John M. McCall PhD, President and Founder, PharMac LLC

Dr. McCall has substantial expertise in drug discovery and development, risk minimization, and medicinal chemistry. John holds 53 US patents and has over 60 refereed publications. He has worked in a number of disease areas including different acute and chronic neurologic disorders and has previously held senior positions within the Pharmaceutical Industry at Pharmacia, Pharmacia & Upjohn and was Vice President of Research with Pfizer. John currently chairs an NINDS development team in the spinal muscular atrophy area, and has co-founded a company that is targeting new therapies for muscular dystrophy.

[Back to List](#)

- Emer Leahy PhD, MBA, PsychoGenics Inc.

Dr. Emer Leahy is President and CEO of PsychoGenics, Inc. She received her Ph.D. in Neuropharmacology at University College Dublin, Ireland and earned her MBA from Columbia University. She has more than 15 years of behavioral research experience, as well as substantial experience in technology assessment business development, licensing, and strategic planning. Within the pharmaceutical and biotech industries she spearheaded contract negotiations and licensing agreements for therapies to treat various neurological disorders.

[Back to List](#)

- Alan Kozikowski PhD, University of Illinois at Chicago

Dr. Kozikowski holds a Professorship within the Department of Medicinal Chemistry and Pharmacognosy at the University of Illinois at Chicago, and is Director of the International Drug Discovery Institute. He has published more than 400 peer reviewed research papers in a variety of prestigious scientific journals and also holds more than 100 patents. Dr. Kozikowski's group focuses on discovery and development of drugs that primarily target neurologic disease. His lab is currently developing unique classes of pharmacologic compounds that may be useful for the treatment of a wide range of neurologic disorders.

[Back to List](#)

- Rajiv Ratan MD, PhD, Burke/Cornell Medical Research Institute

Dr. Ratan is the Burke Professor of Neurology, Neuroscience and Rehab Medicine, Weill Medical College of Cornell University and Director of the prestigious Burke/Cornell Medical Research Institute. His lab focuses on understanding the role of transcription in survival and death in the nervous system. His group has a longstanding interest in oxidative stress, epigenetics, and repair in the brain and the Institute he directs is a major translational center for neurodegenerative and neurodevelopmental disorders.

[Back to List](#)

- Kim Hunter-Schaedle PhD, Children's Tumor Foundation

Dr. Kim Hunter-Schaedle is Chief Scientific Officer of the Children's Tumor Foundation (CTF), which seeks treatments for neurofibromatosis. She has held positions with the technology transfer & research commercialization office of Columbia University and the Juvenile Diabetes Research Foundation, where as Director of Industry Relations she established its successful Industry Discovery and Development Partnership program. She has previously conducted scientific research within academia and industry at the University of London, The Rockefeller University, and the biotechnology company, Curis Inc.

[Back to List](#)

#### Medical Advisory Board (MAB)

- Alan K. Percy, MD Civitan International Research Center, University of Alabama at Birmingham

Dr. Percy is Professor of Pediatric Neurology at UAB and has over 30 years experience working on neurodevelopmental and related disorders. He has been recognized both nationally and internationally for his achievements in the Rett syndrome field. Dr. Percy currently directs the UAB Sparks Clinics Rett Syndrome Research program and heads the NIH funded Angelman, Rett and Prader-Willi syndromes consortium.

[Back to List](#)

- Daniel G. Glaze, MD, Baylor College of Medicine Blue Bird Circle Rett Center

Dr. Glaze is a Professor of Pediatrics in the Section of Neurology at Baylor College of Medicine, Houston, Texas and

Medical Director of the Blue Bird Circle Rett Center and the Children's Hospital Sleep Center clinic. His primary interests are in epilepsy, sleep disorders, and clinical studies on Rett Syndrome. Dr. Glaze also participates in the Rett, Angelman and Prader-Willi syndromes consortium which is conducting longitudinal assessments of patients with Rett syndrome enrolled in a natural history study charting progression of the disease over time.

[Back to List](#)

- Walter E. Kaufmann, MD, Kennedy Krieger Institute, Johns Hopkins University

Dr. Kaufmann is the Director of the Center for Genetic Disorders of Cognition and Behavior and of the Fragile X Clinic at Kennedy Krieger Institute. He is also a Professor of Pathology, Neurology, Pediatrics, Psychiatry and Radiology at the Johns Hopkins University School of Medicine. Dr. Kaufmann's research activities focus on Down syndrome, Rett syndrome, Fragile X syndrome, and disorders of cholesterol metabolism linked to autism. At the international level, Dr. Kaufmann serves as the Chair and Coordinator of RettSearch, an IRSF supported international consortium of Rett syndrome clinical researchers.

[Back to List](#)

- Shlomo Shinnar, MD, PhD, Montefiore Medical Center

Dr. Shinnar is Professor of Neurology and Pediatrics, Hyman Climenko Professor of Neuroscience Research, and Director of the Comprehensive Epilepsy Management Center at Montefiore Medical Center and the Albert Einstein College of Medicine in the Bronx, New York. Dr. Shinnar is highly regarded for his research on topics relating to childhood seizures, including when to initiate and discontinue antiepileptic drug therapy, Dr. Shinnar is a recipient of the Research Recognition Award of the American Epilepsy Society and has authored over 100 peer reviewed papers and over 60 review articles and chapters.

[Back to List](#)

- Bertram H. Lubin, MD, Children's Hospital Oakland Research Institute

Dr. Lubin is President and Director of Medical Research of Children's Hospital Oakland Research Institute. He has considerable experience directing both clinical and basic research programs. His primary research interest has been in pediatric diseases with a focus on sickle cell disease. Dr. Lubin's research interests span a range of pediatric diseases including asthma, obesity, diabetes, nutrition and studies of other pediatric disorders. His research seeks to understand how environmental factors interact with genetics to cause certain diseases.

[Back to List](#)

- Eric Smeets MD, PhD University of Maastricht, Netherlands

Dr. Smeets, is a pediatrician and child neurologist at the Department of Clinical Genetics of the Academic Hospital Maastricht, Maastricht University, the Netherlands and a consultant pediatrician at the Center for Human Genetics, University Hospital Gasthuisberg, Leuven, Belgium. He has long experience with developmental disorders and genetic syndromes in children and adults with intellectual disabilities. Dr. Smeets has published articles on the molecular genetics of Rett syndrome and MECP2 related disorders and other clinical issues in Rett syndrome including cardiorespiratory dysfunction and the aging process.

[Back to List](#)

- Jeffrey L. Neul, MD, PhD Texas Children's Hospital, Baylor College of Medicine

Dr. Neul is Anthony and Cynthia Petrello Scholar at the Jan and Duncan Neurological Research Institute at Texas Children's Hospital and Assistant Professor in Pediatrics in the Section of Neurology, Baylor College of Medicine, Houston, Texas. Dr. Neul's research focus is in both clinical and basic research on Rett syndrome. As such, he is interested in translating knowledge acquired from basic science research into potential therapies for Rett syndrome. His laboratory uses animal models of Rett syndrome to understand mechanisms of disease and to conduct preclinical trials of potential therapies.

[Back to List](#)

- Angus Clarke, DM, FRCP Cardiff University, Wales, UK

Dr. Clarke is currently the Clinical Professor in Medical Genetics at Cardiff University in the UK. He leads the Rett syndrome research team at Cardiff University, which acts as custodian for the British Isles Rett Syndrome Survey and undertakes clinical and molecular genetics research into Rett syndrome and related disorders (such as the CDKL5-associated disorder). His research interests focus on Rett syndrome and ectodermal dysplasia, clinical genetics, genetic counseling, ethical and social issues and newborn screening. Dr. Clarke currently serves as a representative of the Chief Medical Officer for Wales on the UK Commission on Human Genetics, as Medical Advisor to the Rett Syndrome Association UK and the Scottish Rett Syndrome Association and participates in the RettSearch Clinical Research Consortium.

[Back to List](#)

- N. Carolyn Schanen, MD, PhD, Alfred I. duPont Hospital for Children/Nemours Children's Clinic at the University of Delaware

Dr. Schanen is currently Head of Human Genetics Research at the Alfred I. duPont Hospital for Children and holds an Adjunct Assistant Professorship in Human Genetics at UCLA School of Medicine. Dr. Schanen's laboratory has a long-standing interest in Rett and other forms of inherited autism spectrum disorders. Her work has focused on the molecular genetics of Rett syndrome and her laboratory is developing new cell-based assays that can be used to screen possible

drugs to treat Rett syndrome by restoring function to the protein product of the MECP2 gene.

[Back to List](#)

- Peter Huppke, MD Georg August University Göttingen, Germany

Peter Huppke is a Professor of Pediatrics and Pediatric Neurology within the Faculty of Human Genetics, at the Georg August University, Göttingen in Germany. Dr. Huppke's research has focused on the molecular genetics of Rett syndrome. He has conducted research on endocrine function in patients with Rett syndrome and contributed to our understanding of the spectrum of clinical features of the disorder leading to improvements in clinical diagnosis of Rett syndrome. Dr. Huppke's research now focuses on respiratory dysfunction and the testing of potentially disease modifying therapeutics for Rett syndrome.

[Back to List](#)

- Alessandra Renieri MD, PhD University of Siena, Italy

Dr. Renieri is a Professor of Medical Genetics at the University of Siena, Italy. Her specialty is medical genetics with a specific focus on the underlying causes of Rett syndrome and Alport syndrome, a hereditary form of kidney disease. Her laboratory previously identified several novel mutations in the MECP2 gene in patients with Rett syndrome and established that a benign variant form of Rett phenotype most frequently results from either miss-sense mutations or late truncated forms of the MECP2 gene.

[Back to List](#)

- Roger Albin MD, University of Michigan Health System

Dr. Albin is a Professor in the Department of Neurology, Associate Chair for Research Co-Director of the Movement Disorders Clinic and currently serves as Chief of Neuroscience Research within the Veterans Affairs Ann Arbor Healthcare System Geriatric Research, and Clinical Center. Dr. Albin's clinical research focuses on neurologic diseases associated with movement disorders such as Parkinson's and Huntington's disease, Tourette Syndrome, Dystonias, Tremors, disorders of Basal ganglia structure and function and associated Cognitive disorders.

[Back to List](#)

- Evdokia Anagnostou, MD Bloorview Children's Rehab and Research Institute, University of Toronto



Dr. Anagnostou is a clinician-scientist at Bloorview Children's Rehab and Research Institute. She is an Assistant Professor, Department of Pediatrics, University of Toronto and an Adjunct Assistant Professor, Mount Sinai School of Medicine, New York. Her program of research focuses on the psychopharmacology and neuroimaging of autism, and on the development of clinical trials to test novel, traditional, and alternative compounds for the treatment of autism and related disorders. Her research focuses on the development of clinical trials to test novel traditional and alternative compounds for the treatment of autism and related disorders.

[Back to List](#)

- Susan E. Swedo MD, National Institute of Mental Health (NIMH)

Dr. Swedo is a Senior Investigator within the Child Psychiatry intramural branch of the National Institute of Mental Health. She has previously held positions as Head of the Section on Behavioral Pediatrics, Chief of Pediatrics and Developmental Neuropsychiatry and also served as the Acting Scientific Director for NIMH from 1995 through 1998. Dr. Swedo recently received the Joel Elkes International Research Award from the American College of Neuropsychopharmacology. Her laboratory studies childhood-onset obsessive compulsive disorder and related disorders, including Tourette syndrome and Sydenham chorea and she is a specialist with expertise in clinical trial design and implementation specifically in pediatric populations.

[Back to List](#)