

# Barth Syndrome International Family and Scientific Conference

Baltimore, Maryland  
October 18-21, 2002



Barth Syndrome  
Foundation

[www.barthsyndrome.org](http://www.barthsyndrome.org)

Saving boys' lives through  
education, advances in treatment and pursuit of a cure

## Barth Syndrome International Family and Scientific Conference

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October 18, 2002

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Paterson, NJ

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The Hospital for Sick Children  
Toronto, Ontario, Canada

**Iris L. Gonzalez, Ph.D.**  
Molecular Diagnostics Laboratory  
Alfred I. duPont Hospital for Children  
Wilmington, DE

**Grant M. Hatch, Ph.D.**  
Lipid Lipoprotein Research  
University of Manitoba  
Winnipeg, Canada

**Michael Schlame, M.D.**  
Anesthesiology  
NYU School of Medicine  
New York, NY

**Jeffrey A. Towbin, M.D.**  
Pediatric Cardiology  
Baylor College of Medicine  
Texas Children's Hospital  
Houston, TX

**Ronald J. A. Wanders, Ph.D.**  
Genetic Metabolic Diseases  
Academic Medical Center  
Amsterdam, The Netherlands

**Katherine R. McCurdy** – *ex officio*  
Science and Medicine  
Barth Syndrome Foundation, Inc.  
Larchmont, New York

Dear Friends,

On behalf of the Barth Syndrome Foundation, Inc. (BSF) and our Board, welcome to the 2002 International Barth Syndrome Family and Scientific Conference. We are delighted with the quality of the program being offered during this conference to better educate scientists, clinicians and parents alike.

We at BSF are committed to the expansion of our group not only in numbers but also in quality of programs offered to our members. The BSF conference plays a vital role in our effort of networking families, clinicians and scientists with one another. We realize the need for cooperation between all parties in order to maintain the momentum of positive strides in a better understanding of this life-threatening condition and all those affected by it. Your attendance at this conference validates our viewpoint of the need to research more about this disorder to assist all those affected.

We would like to thank the National Institute of Neurological Disorders and Stroke (NINDS), The Office of Rare Diseases (ORD), The Lied Foundation; Christina Hixson, Trustee and all of the many others who supported us in making this conference a reality. The assistance of these organizations confirms this conference is much needed and through your cooperation we can and will make a difference in the way we understand Barth syndrome in the future.

On a personal note, as a parent of a child with Barth syndrome, I thank you. Your affiliation with our group can and will make a difference in my son's future and all the others who live day-to-day with Barth syndrome. As a parent of a child who has died and one who is living with Barth syndrome, I am sure I speak on behalf of all parents in embracing the ultimate vision of BSF **"...not one more child will suffer or perish from this condition"** and our mission **"To guide the search for a cure, to educate and support physicians and to create a caring community for affected families."** It is my greatest desire for each of us to leave this conference with a renewed commitment to our mission and a personal action plan to help us make our vision a reality.

Respectfully,

**Shelley Bowen**

Shelley Bowen  
President  
The Barth Syndrome Foundation, Inc.

## AGENDA FOR OCTOBER 2002 BARTH SYNDROME SCIENTIFIC MEETING

\*\*\* *All sessions will be held at the Holiday Inn, Inner Harbor unless otherwise noted*\*\*\*

**\*\*Please make reference to the list of Bios located on pages 8-13\*\***

### **FRIDAY, OCTOBER 18, 2002**

- 9:00—17:00 Clinics and consultations for Barth patients with selected physicians in various locations
- 17:00—18:30 *Gerald F. Cox, M.D., Ph.D. — Children's Hospital, Boston, MA and Genzyme Corporation, Cambridge, MA (Fairmont Building, 3rd Floor, Kennedy Krieger Institute)*  
“Multidisciplinary clinical roundtable discussion about treatment of Barth patients”
- 18:00—22:00 Registration and reception (**McHenry II Conference Rm.**)

### **SATURDAY, OCTOBER 19, 2002 (Harbor IA Conference Rm.)**

8:00—8:15 MORNING COFFEE AND DANISH available outside **Harbor Foyer**

#### **Introduction to Barth Syndrome**

- 8:15—8:20 *Stephen C. Groft, Pharm. D. — Director, Office of Rare Diseases, National Institutes of Health, Bethesda, MD*  
“Welcome and Opening Remarks”
- 8:20—8:30 *Giovanna Spinella, M.D. — Program Director, Neurogenetics, National Institute of Neurological Disorders and Stroke, Bethesda, MD*  
“Development of a Barth Syndrome Research Agenda”
- 8:30—9:15 *Peter G. Barth, M.D., Ph.D. — University of Amsterdam, Amsterdam, The Netherlands*  
“X-Linked cardiomyopathy and neutropenia - history and natural history of Barth syndrome”
- 9:15—9:45 *Richard I. Kelley, M.D., Ph.D. — Kennedy Krieger Institute, affiliate of Johns Hopkins Medical Institutions, Baltimore, MD*  
“Barth syndrome: a complex metabolic disorder”
- 9:45—10:20 *Iris L. Gonzalez, Ph.D. — Alfred I. duPont Children's Hospital, Wilmington, DE*  
“G4.5, a gene with many mutations”
- 10:20—10:40 **BREAK (Harbor Foyer)**
- 10:40—11:20 *Colin G. Steward, M.R.C.P., Ph.D., F.R.C.P.C.H. — Bristol Royal Hospital for Sick Children, Bristol, UK*  
“The underdiagnosis of Barth syndrome”
- 11:20—12:10 *Fredoen Valianpour, Ph.D. Candidate — University of Amsterdam, Amsterdam, The Netherlands*  
“New insights in cardiolipin deficiency in Barth syndrome; implications for treatment”

12:10—13:00 WORKING LUNCH with physicians and scientists (**Harbor IA Conference Rm.**)

**Clinical Aspects of Barth Syndrome**

13:00—13:40 *Jeffrey A. Towbin, M.D. — Baylor College of Medicine, Houston, TX*  
“Spectrum of cardiac function and pathology in Barth syndrome”

13:40—13:55 *Barry J. Byrne, M.D., Ph.D. — Shands Children’s Hospital, Gainesville, FL*  
“Summary of observations and insights from yesterday’s cardiology clinics with Barth patients”

13:55—14:35 *Mary Ann Bonilla, M.D. — St. Joseph’s Children’s Hospital, Paterson, NJ and Advisory Board member for the Severe Chronic Neutropenia International Registry*  
“Nature and treatment of hematologic disease in Barth syndrome”

14:35—14:50 *Tyler Reimschisel, M.D. — Johns Hopkins Medical Institutions, Baltimore, MD*  
“Summary of observations and insights from yesterday’s neurology clinics with Barth patients”

14:50—15:05 *Gerald F. Cox, M.D., Ph.D. — Children’s Hospital, Boston, MA and Genzyme Corporation, Cambridge, MA*  
“Summary of observations and insights from yesterday’s neurology clinics with Barth patients”

15:05—15:20 BREAK (**Harbor Foyer**)

15:20—15:50 *Richard I. Kelley, M.D., Ph.D. — Kennedy Krieger Institute, affiliate of Johns Hopkins Medical Institutions, Baltimore, MD*  
“Autosomal recessive Barth syndrome: a second disorder with 3-methylglutaconic aciduria and evidence of abnormal phospholipid metabolism”

**Barth Syndrome Foundation, Inc.**

15:50—16:20 *Valerie “Shelley” M. Bowen — President, The Barth Syndrome Foundation, Inc.*  
“The Barth Syndrome Foundation, Inc.”

16:20—18:00 FREE TIME

18:00—21:00 DINNER for Barth families, physicians, and scientists co-sponsored by Starlight Children’s Foundation and A.C. and Rosa Mann  
(**Turner Auditorium Lobby, Johns Hopkins Campus**)

**SUNDAY, OCTOBER 20, 2002 (McHenry I Conference Rm.)**

8:40—9:00 MORNING COFFEE AND DANISH available outside **Harbor Foyer**

### **Biochemistry of Cardiolipin and Other Phospholipids**

- 9:00—9:50 *Miriam Greenberg, Ph.D. — Wayne State University, Detroit, MI*  
“Biosynthesis of cardiolipin and related phospholipids”
- 9:50—10:40 *Grant M. Hatch, Ph.D. — University of Manitoba, Winnipeg, Manitoba*  
“Remodeling of cardiolipin and other related phospholipids”
- 10:40—11:00 **BREAK (Harbor Foyer)**

### **Biochemistry of Barth Syndrome**

- 11:00—11:50 *Michael Schlame, M.D. — New York University Hospital, New York*  
“Cardiolipin levels and metabolism in Barth syndrome muscle and platelets”
- 11:50—13:00 LUNCH with other physicians, researchers and families (**Chesapeake I Conference Rm.**)
- 13:00—13:30 *Richard I. Kelley, M.D., Ph.D. — Kennedy Krieger Institute, affiliate of Johns Hopkins Medical Institutions, Baltimore, MD*  
“Evaluation of Barth syndromes by in vivo 31P-NMR spectroscopy of heart and skeletal muscle”
- 13:30—14:20 *Troy Phipps, Ph.D. — University of Southern California, Los Angeles, CA*  
“Does a CL remodeling defect adequately describe BTHS genotype/phenotype pathophysiology?”
- 14:20—14:40 **BREAK (Harbor Foyer)**

### **Research and Next Steps**

- 14:40—15:30 *Richard I. Kelley, M.D., Ph.D. — Kennedy Krieger Institute, affiliate of Johns Hopkins Medical Institutions, Baltimore, MD; Chairman of The Barth Syndrome Foundation, Inc. Scientific and Medical Advisory Board*  
Moderates section leaders’ summaries and discussions of future clinical and research initiatives related to Barth syndrome
- 15:30—15:45 *Giovanna Spinella, M.D. — Program Director, Neurogenetics, National Institute of Neurological Disorders and Stroke, Bethesda, MD*  
“Closing remarks”

### **End of General Conference**

- 16:30—18:30 Board Meeting for the BSF Scientific and Medical Advisory Board (**Boardroom B**)
- 18:30—19:00 WORKING DINNER (sandwiches brought in)
- 19:00—20:00 Continuation of Board Meeting for the BSF Scientific and Medical Advisory Board

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## AGENDA FOR OCTOBER 2002 BARTH SYNDROME FAMILY MEETING

\*\*\* *All sessions will be held at the Holiday Inn, Inner Harbor unless otherwise noted*\*\*\*

*\*\*Please make reference to the list of Bios located on pages 8-13\*\**

### **FRIDAY, OCTOBER 18, 2002**

- 8:15—16:35 Clinics and consultations for Barth patients with selected physicians in various locations
- 18:00—22:00 Registration and reception (**McHenry II Conference Rm.**)

### **SATURDAY, OCTOBER 19, 2002 (Harbor IB Conference Rm.)**

- 8:15—8:30 MORNING COFFEE AND DANISH available (**Harbor Foyer**)
- 8:30—8:50 “Welcome and Introduction to the Meeting”  
*Valerie “Shelley” M. Bowen — President, The Barth Syndrome Foundation, Inc.*
- 8:50—9:10 “Overview of Barth Syndrome”  
*Richard I. Kelley, M.D., Ph.D. — Kennedy Krieger Institute, affiliate of Johns Hopkins Medical Institutions, Baltimore, MD*
- 9:10—9:30 “Genetics of Barth Syndrome”  
*Iris L. Gonzalez, Ph.D. — Alfred I. duPont Children’s Hospital, Wilmington, DE*
- 9:30—10:10 “Brief History of Barth Syndrome”  
*Peter G. Barth, M.D., Ph.D. — Emma Children’s Hospital, Amsterdam, The Netherlands*
- 10:10—10:30 BREAK (**Harbor Foyer**)
- 10:30—10:50 “Phospholipid Metabolism — a Parent’s Primer”  
*Michael Schlame, M.D. — New York University Hospital, New York, NY*
- 10:50—12:20 Panel on **Cardiomyopathy** (progression, medications, transplantation)  
*Jeffrey A. Towbin, M.D., moderator — Texas Children’s Hospital, Houston, TX*  
*Barry J. Byrne, M.D., Ph.D. — Shands Children’s Hospital, Gainesville, FL*  
*W. Reid Thompson, M.D. — Johns Hopkins Medical Institutions, Baltimore, MD*
- 12:20—13:20 LUNCH at Holiday Inn (**Chesapeake I Conference Rm.**)
- 13:20—13:40 “Growth and nutrition in Barth syndrome”  
*Richard I. Kelley, M.D., Ph.D. — Kennedy Krieger Institute, affiliate of Johns Hopkins Medical Institutions, Baltimore, MD*

13:40—15:10 Panel on **Skeletal Muscle Weakness, Lack of Stamina, Fatigue, Headache, Small Stature**

*Peter G. Barth, M.D., Ph.D., moderator — Emma Children's Hospital, Amsterdam, The Netherlands*

*Salvatore DiMauro, M.D. — New York Presbyterian Hospital, New York, NY*

*Annette Feigenbaum, M.B., Ch.B., F.R.C.P.C. — Hospital for Sick Children, Toronto, Ontario, Canada*

*Richard I. Kelley, M.D., Ph.D. — Kennedy Krieger Institute, affiliate of Johns Hopkins Medical Institutions, Baltimore, MD (Until 14:35)*

15:10—15:30 **BREAK (Harbor Foyer)**

15:30—17:00 Panel of **Neutropenia and Infection** (how to calculate ANC, SCNIR, G-CSF; preventing infection, and how to deal with infection)

*Mary Ann Bonilla, M.D., moderator — St. Joseph's Children's Hospital, Paterson, NJ*

*Colin G. Steward, MRCP, Ph.D., F.R.C.P.C.H. — Bristol Royal Hospital for Sick Children, Bristol, UK*

*Gerald F. Cox, M.D., Ph.D. — Medical Director of Clinical Research, Genzyme Corporation, Cambridge, MA*

17:00—18:00 **FREE TIME**

18:00—21:00 **DINNER** for Barth families, physicians, and scientists co-sponsored by Starlight Children's Foundation and A.C. and Rosa Mann  
**(Turner Auditorium Lobby, Johns Hopkins Campus)**

**Sunday, October 20, 2002**

8:40—9:00 **MORNING COFFEE AND DANISH** available in the **Harbor Foyer** throughout the morning for breaks whenever appropriate

9:00—11:00 **Four simultaneous sessions**

**Session A**— Parent Discussion about **Social, Educational and Nutritional** issues of Barth boys aged 0-11 years (**Harbor IB Conference Rm.**)

*Lynda M. Sedefian, parent moderator*

*Michele M. M. Mazzocco, Ph.D. — Kennedy Krieger Institute, Baltimore, MD*

*Eileen McMahon, R.D. — Kennedy Krieger Institute, Baltimore, MD*

*Jules Spotts, Ph.D., P.C. — New Canaan, CT (9:00 – 10:00)*



**Session B**— Parent Discussion about **Social, Educational and Nutritional** issues of Barth boys aged 12 years old plus (**Harbor IA Conference Rm.**)

*Susan V. Wilkins, parent moderator*

*Joan C. Stoner, Ed.D. — Capella University, Minneapolis, Minnesota*

*Eileen Q. Juico, M.A., M.Ed., Learning Specialist —Rye Country Day School, Rye, NY*

*Jules Spotts, Ph.D., P.C. — New Canaan, CT (10:00 – 11:00)*

**Session C**— **Barth Youth Forum (McHenry II Conference Rm.)**

*Peter G. Barth, M.D., Ph.D., moderator — Emma Children’s Hospital, Amsterdam, The Netherlands*

*Mary Ann Bonilla, M.D. — St. Joseph’s Children’s Hospital, Paterson, NJ*

*Jeffrey A. Towbin, M.D. — Texas Children’s Hospital, Houston, TX*

*Richard I. Kelley, M.D., Ph.D. — Kennedy Krieger Institute, Baltimore, MD*

*Jaclyn M. Butera, M.S.W., C.S.W, M.S.Ed., Counselor and Dean — Rye Country Day School, Rye, NY*

**Session D**— **Grandparents’ Forum (Boardroom B)**

*Judith M. Levy, M.S.W., M.A. — Kennedy Krieger Institute, Baltimore, MD*

11:00—12:00 FREE TIME

12:00—13:00 LUNCH at Holiday Inn with Physicians and Scientists attending Scientific Meeting  
(**Chesapeake I Conference Rm.**)

13:00—15:00 The Barth Syndrome Foundation, Inc. Meeting (Detailed Agenda To Be Determined)  
(**Harbor 1AB Conference Rm.**)

15:00—15:30 BREAK (**Harbor Foyer**)

15:30—16:30 Activities Fair (Booths with information (**Harbor Foyer**))

16:30—18:00 International Meeting Regarding Creation of BSF Affiliate Organizations  
(**Harbor IAB Conference Rm.**)

## **SPEAKERS INVOLVED IN FAMILY AND SCIENTIFIC SESSIONS AND CLINICIANS INVOLVED IN CLINICAL STUDIES AND CONSULTATIONS**

**Peter. G. Barth, M.D.** — Pediatric Neurology, Emma Children’s Hospital / AMC; Professor, Pediatric Neurology, University of Amsterdam, Amsterdam, The Netherlands; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. Barth is the pediatric neurologist who first described the condition that now bears his name, Barth syndrome. He is Co-founder of the Dutch Pediatric Neurology Society (1980). His present research activities, in cooperation with Laboratories of Genetic Metabolic Diseases and Neurogenetics, include Barth syndrome, Peroxisome Biogenesis Disorders, and Neurodegenerative disorders with prenatal onset.*

**Mary Ann Bonilla, M.D.** — Division of Pediatric Hematology Oncology, St. Joseph’s Children’s Hospital; Assistant Professor, Department of Pediatrics, Columbia University; Advisory Board, Severe Chronic Neutropenia International Registry; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. Bonilla was involved in conducting the initial clinical trials for G-CSF as a treatment for congenital neutropenia while at Memorial Sloan-Kettering Hospital. She is a pediatric hematologist oncologist who maintains an active interest in the treatment of white cell disorders, including Barth syndrome. As an Advisory Board member for the Severe Chronic Neutropenia International Registry, she has helped treat at least one Barth patient and has been involved with data concerning many more patients with Barth syndrome.*

**Valerie “Shelley” M. Bowen** — President, The Barth Syndrome Foundation, Inc.

*Shelley Bowen has had two sons who have been diagnosed with Barth syndrome. As a result of losing her first son to this life-threatening disorder and having another son diagnosed, she realized the need for an organization to support her family and others alike. Shelley has been passionate about networking scientists, clinicians and families since 1998. She firmly believes that any parent of a child with Barth syndrome is capable of becoming an effective advocate in the care of a child with Barth syndrome. "Because of the existence of the Barth Syndrome Foundation, Inc. there is no longer any need for isolation in caring for a child with Barth syndrome."*

**Jaclyn M. Butera, M.S.W., C.S.W., M.Ed.** — Counselor and Dean, Rye Country Day School, Rye, NY

*Ms. Butera has spent her career working with youth in various capacities – from case worker to youth advocate to school dean. She currently serves as an Upper School administrator, the Academic Dean for the tenth grade, the Upper School Counselor and the Dean of Student Life at the Rye Country Day School in New York.*

**Barry J. Byrne, M.D., Ph.D.** — Cardiology Director, Department of Pediatrics, Shands Children’s Hospital, University School of Medicine, Gainesville, FL; Professor and Associate Chair of Pediatrics, Molecular Genetics & Microbiology; Director, Powell Gene Therapy Center, University of Florida, Gainesville, FL; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. Byrne has both clinical and research interests in Barth syndrome. As a pediatric cardiologist, his main emphases include cardiomyopathy and transplantation; and he currently treats two boys with Barth syndrome. His research focus is on developing new gene therapies for cardiovascular disease. In the area of cardiomyopathy, his laboratory is studying gene replacement in an autosomal recessive form of fatal cardiomyopathy in children. The disease is the prototype of lysosomal storage disorders leading to skeletal and cardiac muscle weakness.*

**Gerald F. Cox, M.D., Ph.D.** — Medical Director, Department of Clinical Research, Genzyme Corporation Cambridge, MA; Assistant in Medicine, Division of Genetics, Children's Hospital, Boston, MA; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*As a clinician in genetics, Dr. Cox has been involved in the care of several Barth patients. His particular interests include the genetic basis of cardiomyopathy and treatment of inborn errors of metabolism. In addition, he oversees clinical trials for a well-known biotechnology corporation.*

**Salvatore DiMauro, M.D.** — Lucy G. Moses Professor of Neurology, Columbia University College of Physicians and Surgeons; Director, Columbia University Myopathy DNA Diagnostics Laboratory, New York, NY; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. DiMauro is a world-renown authority on mitochondrial diseases. His laboratory conducts biochemical and DNA investigations of human metabolic myopathies. Throughout his career, Dr. DiMauro has kept a focused interest on inborn errors of energy metabolism, recognizing unusual patients through clinical observation, and using both biochemical and molecular approaches to define disease entities.*

**Annette Feigenbaum, M.D., Ch.B., F.R.C.P.C.** — Division of Clinical and Metabolic Genetics, The Hospital for Sick Children; Assistant Professor, Department of Pediatrics, University of Toronto, Toronto, Ontario, Canada; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. Feigenbaum is a clinician who has helped treat a number of Barth patients in Canada. She also conducts research, with her primary interests including issues of mitochondrial diseases.*

**Iris L. Gonzalez, Ph.D.** — Molecular Diagnostics Laboratory, Alfred I. duPont Hospital for Children, Wilmington, DE; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. Gonzalez works in the only molecular diagnostics laboratory that has been approved by the CLIA (the Department of Health and Human Services' Clinical Laboratory Improvement Amendments) in the United States to conduct the genetic analysis necessary to confirm a diagnosis of Barth syndrome. She has written a layman's guide to genetics that has been extremely valuable to BSF families and others.*

**Miriam L. Greenberg, Ph.D.** — Professor, Department of Biological Sciences, Wayne State University; Detroit, MI

*Dr. Greenberg's well-known research interests focus on genetic control of mitochondrial membrane biogenesis in yeast, in particular. How newly synthesized lipids and proteins are integrated into the membranes of specific organelles is a central question in organelle biogenesis.*

**Stephen C. Groft, Pharm. D.** — Director, Office of Rare Diseases, National Institutes of Health, Bethesda, MD

*Dr. Groft began his focus on rare disorders in 1982 when he became one of the first six staff people in the Office of Orphan Drug Products. He has been the Director of the Office of Rare Diseases at the National Institutes of Health (NIH) since 1993 when it was established. With his expertise in pharmacology, he also recently has headed the White House Commission on Complementary and Alternative Medicine Policy. He has long been an important champion of rare disorders.*

**Grant M. Hatch, Ph.D.** — Director of the Lipid Lipoprotein and Atherosclerosis Research Group, University of Manitoba; Professor, Department of Pharmacology and Therapeutics; Department of Biochemistry and Medical Genetics, University of Manitoba, Winnipeg, Canada; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. Hatch's research interests focus on metabolism and pharmacological modulation of phospholipids (including cardiolipin) in the mammalian heart and cells in culture. He has published numerous papers on these topics.*

**Eileen Q. Juico, M.A., M.Ed.** — Learning Specialist, Rye County Day School, Rye, NY

*Ms. Juico is an educator with a particular interest in learning differences. She holds an M.Ed. in Curriculum & Instruction and an M.A. in special education. Having worked in several different public and private school settings, she currently is the Upper School Learning Specialist at the Rye Country Day School where she works closely with one Barth boy.*

**Richard I. Kelley, M.D., Ph.D.** — Director, Division of Metabolism, Kennedy Krieger Institute; Associate Professor, Department of Pediatrics, Johns Hopkins University, Baltimore, MD; Chair, The Barth Syndrome Foundation Scientific and Medical Advisory Board

*Dr. Kelley is an expert in metabolic diseases and has been involved in the treatment of more cases of Barth syndrome than any other individual in the US. He hosted the first International Scientific and Family Conference on Barth Syndrome in June 2000 at Johns Hopkins' Kennedy Krieger Institute.*

**Rebecca Kern, M.G.C.** — Kennedy Krieger Institute, Baltimore, MD

*Rebecca Kern joined Dr. Kelley's team at Kennedy Krieger Institute as a genetic counselor in July of 2002. One of her primary roles is to assist with both clinical care and research involving families with Barth syndrome. Other interests include new technologies and education relevant to newborn screening, pre-implantation genetic diagnosis for single gene disorders, and neuropsychiatric genetics.*

**Judith M. Levy, M.S.W., M.A.** — Director, Department of Social Work Chair, Ethics Committee, Kennedy Krieger Institute, Baltimore, MD

*Judith M. Levy is the Director of Social Work, and Director for the Therapeutic Foster Care and Adoptions Program at the Kennedy Krieger Institute. Ms. Levy has over 30 years of experience in the social work field ranging from direct clinical practice to administrative roles. Her major areas of interest include: ethical issues pertaining to people with disabilities, and psychosocial issues related to individuals with developmental and neurological disabilities.*

**Michele M. M. Mazzocco, Ph.D.** — Kennedy Krieger Institute; Associate Professor, Psychiatry and Behavioral Sciences, Johns Hopkins University School of Medicine, Baltimore, MD

*Dr. Mazzocco's primary research interests pertain to general cognitive development in young children. In that context, she has maintained research programs that fall under two general categories which involve studies of cognitive development and cognitive phenotypes in children with specific disorders.*

**Eileen McMahon, M.S., R.D., L.D., C.N.S.D.** — Pediatric Clinical Nutritionist; Clinical Dietician, Kennedy Krieger Institute, Baltimore, MD

*Eileen McMahon conducts comprehensive clinical evaluations and treatments with inpatient and outpatient populations. As such, she works closely with an interdisciplinary team, including pediatric gastroenterologists, occupational therapists, speech and language pathologists, behavior psychologists, and social workers.*

**Troy Phipps, Doctoral Student** — University of Southern California, Institute for Genetic Medicine, Department of Biochemistry and Molecular Biology, Los Angeles, CA

*Troy Phipps is a Ph.D Candidate in Biochemistry and Molecular Biology at the University of Southern California in Los Angeles. His main areas of interest include inborn errors of metabolism, human nutritional biochemistry, and understanding how single nucleotide polymorphisms may alter androgen metabolism in relation to complex human diseases.*

**Tyler Reimschisel, M.D.** — Resident in Pediatric Neurology, The Johns Hopkins Hospital, Baltimore, MD

*Dr. Reimschisel's research and clinical interests involve Genetic diseases that affect the nervous system.*

**Michael Schlame, M.D.** — Department of Anesthesiology, NYU School of Medicine, New York, NY; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. Schlame's clinical focus includes Adult and Pediatric Critical Care, Cardiothoracic Anesthesia, and Pediatric Anesthesia. His research interests include Mitochondrial Energy Metabolism, Pulmonary Surfactant, Cardiolipin, Mechanisms of Multiple Organ Failure, and Cardiomyopathy.*

**Carolyn T. Spencer, M.D.** — Assistant Professor, Division of Pediatric Cardiology, University of Florida College of Medicine, Department of Pediatrics (Cardiology), Gainesville, FL; Fellow, Non-invasive Laboratory, Department of Cardiology, Children's Hospital Boston,

*Dr. Spencer is the Director of Pediatric Echocardiography at the University of Florida. Her particular interests include echocardiography and management of heart failure as well as adults with congenital heart disease.*

**Philip J. Spevak, M.D.** — Pediatric Cardiology, The Johns Hopkins University, Baltimore, MD

*Dr. Spevak is the Director of Pediatric Echocardiography at Johns Hopkins. His particular interests include echocardiography of the fetus, infant and child as well as that of adults with congenital heart disease.*

**Giovanna Spinella, M.D.** — Program Director, Neurogenetics, National Institute of Neurological Disorders and Stroke, Bethesda, MD

*As a NINDS Program Director, Dr. Spinella's specialties include pediatric and neonatal brain injury, neuro-development, pediatric neurodegenerative disease and hereditary ataxias, neurobehavior, developmental neuroimaging, movement disorders and tics, choreas, myoclonic disorders, dystonias, clinical ADHD. She has been a wonderful early advocate for the work of The Barth Syndrome Foundation, Inc.*

**Jules Spotts, Ph.D., P.C.** — Child Guidance Center, New Canaan, CT

*Dr. Spotts is a Clinical Psychologist who has a private practice in Connecticut and who also provides a broad range of mental health services at several private schools located in the New York and Connecticut areas.*

**Colin G. Steward, B.M., B.Ch., M.R.C.P., F.R.C.Pc.H., Ph.D.** — Bristol Royal Hospital for Sick Children, Bristol, UK

*Dr. Steward is a hematologist whose special interests include bone marrow transplantation, genetic diseases, metabolic diseases, osteopetrosis, norenoleukodystrophy, and Fanconi anaemia.*

**Joan C. Stoner, Ed. D.** — Capella University, an all internet university that is North Central Accredited with administrative offices located in Minneapolis, Minnesota

*Dr. Stoner was a Learning Disabilities Specialist at the Menninger Center for Learning Disabilities. She developed The Center for Learning Disabilities to prepare teachers to implement a mandated program for students with dyslexia in the state of Louisiana. She has established two college programs for dyslexic students and taught dyslexic students at all levels. Dr. Stoner earned the Ed.D. in Administration, Curriculum and Supervision from the University of Nebraska-Lincoln.*

**W. Reid Thompson, M.D.** — Assistant Professor, Division of Pediatric Cardiology, The Johns Hopkins University, Baltimore, MD

*Dr. Thompson has been a full time faculty member in the Department of Pediatrics, Division of Pediatric Cardiology, at the Johns Hopkins University School of Medicine since 1992. His special interests are echocardiography, the evaluation of ventricular function, and exploring new methods of teaching cardiac auscultation.*

**Jeffrey Towbin, M.D.** — Associate Chief, Pediatric Cardiology; Professor, Department of Pediatrics and Molecular and Human Genetics, Baylor College of Medicine; Director, Phoebe Willingham Muzzy Pediatric Molecular Cardiology Laboratory; Texas Children's Hospital Foundation Chair of Pediatric Cardiac Research; Director, Heart Failure and Transplant Program, Texas Children's Hospital, Houston, TX; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Dr. Towbin is a pediatric cardiologist whose major interests include cardiomyopathy, cardiovascular genetics and cardiac transplantation. In addition to his clinical practice, he also conducts research in related areas.*

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**Fredoan Valianpour, Ph.D. Candidate** — University of Amsterdam, Amsterdam, The Netherlands

*Fredoan Valianpour holds a master degree in analytical chemistry and is now working at the Laboratory Genetic Metabolic Diseases as a Ph.D. candidate. Together with Dr. Peter Vreken, he first described the CL deficiency in BTHS patients. His research concerns the use of mass spectrometry in diagnostics and research on metabolic disorders, and his major field of interest is phospholipids and their fatty acid composition.*

**Ronald J. A. Wanders, Ph.D.** — Laboratory of Genetic Metabolic Diseases, Academic Medical Center; Professor Dr., Department of Pediatrics, Emma Children's Hospital and Department of Clinical Chemistry, University of Amsterdam, Amsterdam, The Netherlands; Scientific and Medical Advisory Board, The Barth Syndrome Foundation, Inc.

*Professor Wanders heads the laboratory in which the initial work concerning the underlying biochemical causes of Barth syndrome was conducted. This laboratory works very closely with Dr. Peter Barth and continues to make very important contributions to the understanding of Barth syndrome.*

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### **CLINICS / RESEARCH STUDIES**

**Cardiology:** Barry J. Byrne, M.D.; Carolyn T. Spencer, M.D.; Phillip J. Spevak, M.D.; and W. Reid Thompson, M.D.

**Math Skills Development Project:** Michele Mazzocco, Ph.D.

**Neurology:** Tyler Reimschisel, M.D.

### **CONSULTATIONS**

**Cardiology:** Jeffrey Towbin, Ph.D., M.D.

**Neurology:** Peter G. Barth, Ph.D., M.D.

**Metabolics:** Richard I. Kelley, Ph.D., M.D.

**Hematology:** Colin G. Steward, Ph.D., M.D.

**Genetics:** Iris Gonzalez, Ph.D.; Rebecca Kern, Ph.D.

**Education:** Joan C. Stoner, Ph.D.; Eileen Q. Juico, M.A., M.Ed.

**Nutrition:** Eileen McMahon, R.D.

**Coordinators:** Jean Smoot, R.N.; Elaine Stashinko, R.N., Ph.D.

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