FOD | OAA
National Metabolic Family Conference

JULY 25 – 26 2014
ARLINGTON VA

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Dr Kim Chapman and Jana Monaco for helping us locate local Speakers and other conference services. Eileen Shank (FOD mom) for helping with the Hotel/Conference planning and Stephanie Harry (LCHAD mom) for helping with food planning with the hotel Chef! And Raymonde DeGrace for another wonderful ending ceremony slideshow!

a big **THANK YOU**
to all that helped make this conference possible!

FRIDAY AGENDA

FATTY OXIDATION DISORDERS GROUP

ORGANIC ACIDEMIA ASSOCIATION GROUP

7:00 - 8:00 am	Continental Breakfast for Conference Registrants
8:15- 8:30 am	Introductions by Deb & Kathy in Salon 4 | Kid's Activity Room - Alexandria

TIME & LOCATION	SPEAKER	TIME & LOCATION	SPEAKER
8:30 - 9:30 am Salon 4	**Brian Kirmse**, MD Children's National Medical Center **TOPIC:** Turning Fat to Water: Energy Metabolism & Fatty Acid Oxidation	8:30 - 9:15 am Salon 5/6	**Hilary Vernon**, MD, PhD Johns Hopkins University School of Medicine **TOPIC:** Introduction to OA's
9:30 - 10:30 am Salon 4	**Matthew Rasberry**, RD, LD, CNSC Children's National Health System **TOPIC:** Passing the Baton of Medical/ Nutritional Management	9:30 - 10:30 am Salon 5/6	**Wadih Zein**, MD, NIH **TOPIC:** Optical Concerns with OA's
10:30 - 10:45 am	Break	10:00 - 10:15 am	Break
10:45 - 11:30 am Salon 4	**FOD Breakout Sessions** Introduce families, then move to groups (ie., MCAD, VLCAD/ LCHAD/TFP, CPT2, GA2, teens/adults, etc) Professionals move between groups.	10:15 - 11:00 am Salon 5/6	**Kim Chapman**, MD, PhD Children's National Medical Center **TOPIC:** Secondary Energy
11:30 - 12:00 pm Salon 4	**Teen and Adult FODer Panel** Discussion and Q&A (dependant on attendance – or merge with breakouts)	11:00- 12:00 pm Salon 5/6	Introductions **Panel of Affected OA's** facilitated by Jen Sloan, NIH Amber Wist and other affected OA adults
12:00 - 1:00 pm	Lunch for Conference Registrants – Food in Foyer, Eat in Salon 4 & 5/6		
1:15 - 2:15 pm Salon 4	**Lynne A. Wolfe**, MS, CRNP, BC NIH/NHGRI **TOPIC:**The Elusive Magic Pill: Supplement Use in FODs	1:15 - 2:00 pm Salon 5/6	**George Diaz,** MD, Mt. Sinai, NY **TOPIC:** What Does It Mean to Have Free Radicals in Your Urine? **Dr. Ah Mew**, Children's National – Carbaglu Study
2:15 - 3:15 pm Salon 4	**Carol Greene**, MD **TOPIC:** With a difference: Exploring variability in management of inborn errors of metabolism	2:00 - 3:00 pm Salon 5/6	**Matthew Rasberry**, RD, LD, CNSC Children's National Medical Center **TOPIC:** Passing the Baton of Medical/ Nutritional Management
3:15 - 3:30 pm	Break		
3:30 - 4:30 pm Salon 4	**Carla Messenger**, PhD, Psychologist **TOPIC:** Staying strong: Building resilience among children/teens with chronic illnesses and their families	3:30 - 4:15 pm Salon 5/6	**Chuck Venditti**, MD, PhD, NIH **TOPIC:** The Translational Research Approach to Study MMA and Cobalamin Disorders
4:30 - 5:15 pm	**Professional Panel** - Q&A with several of our speakers, adult FODers and some 'old timer' parents!	4:15 - 5:00 pm Salon 5/6	**William Zinnanti**, MD Mechanism of metabolic stroke and spontaneous cerebral hemorrhage in Organic Acidemias
5:15 pm	Summary and THANK YOU's!		
6 - 9 pm	RECEPTION FOR FAMILIES, PROFESSIONALS, AND SPEAKERS - SKYVIEW		

SATURDAY AGENDA

FATTY OXIDATION DISORDERS AND ORGANIC ACIDEMIA ASSOCIATION JOINT SESSION

Wear your FOD and OAA t-shirts

TIME & LOCATION	SPEAKER
7:00 - 8:00 am	Continental Breakfast for Conference Registrants
8:15 - 8:30 am	Introductions by Deb & Kathy in Salon 4 | Kid's Activity Room - Alexandria
8:30 - 9:30 am Salon 4	**Peter McGuire**, MS, MD, FAAP, NIH/NHGRI **TOPIC:** The NIH MINI Study: Metabolism, Infection and Immunity in Inborn Errors of Metabolism
9:30 - 10:30 am Salon 4	**Andrea Gropman**, MD Neurologist, PhD Children's National Medical Center **TOPIC:** Deciphering the neurology of rare inborn errors of metabolism with a focus on organic acidemias and fatty acid oxidation disorders
10:30 - 10:45 am	Break
10:45 - 11:15 am Salon 4	**Melanie Lockhart** Director, State Affairs, March of Dimes **TOPIC:** Public Policies and Inborn Errors of Metabolism
11:15 - 11:45 am	NBS and Metabolic Formula Panel: **Marshall Summar**, MD, **Jana Monaco**, **Kim Chapman**, MD, PhD, **Melanie Lockhart**
11:45 - 1:00 pm	Lunch for Conference Registrants – Salon 5/6
1:15 - 2:15pm Salon 4	**Suzanne Ripley**, PhD Special Needs Advocate **TOPIC:** Possibilities, Processes, and People: IEPs, 504 Plans and getting what you need
2:15 - 3:15pm Salon 4	**Lynne A. Wolfe**, MS, CRNP, BC NIH/NHGRI **TOPIC:** Transitions: Growing Up with Special Health Needs
3:15 - 3:30 pm	Break - **PICTURE OF BOTH GROUPS WITH YOUR T-SHIRTS!**
3:30 - 4:00 pm Salon 4	**Chuck Hehmeyer** – Attorney, **Raynes McCarty** TOPIC: The Importance of Emergency Protocols for Patients with Inborn Errors
4:00 - 5:00 pm Salon 4	Professional Panel with all conference speakers
5:00 - 5:15 pm Salon 4	Slideshow Ceremony - be sure to stay!

SibSHOP 10 am – Noon in Manassas!

CONFERENCE **SPEAKERS**

The **FOD Family Support Group** and the **Organic Acidemia Association** would like to extend a very sincere appreciation to all of our honored Speakers. Our Families will benefit greatly from having them share their expertise and experiences in the various fields related to metabolic disorders. Thank you!

+ Kimberly A **CHAPMAN** MD, PhD, FACMG

Dr. Kim Chapman is an assistant professor of Pediatrics at the George Washington Medical School and Health Sciences Center. She is also an attending geneticist at the Children's National Medical Center in Washington, D.C.. Dr. Chapman co-hosted the 2011 Consensus conference of PA diagnosis and management at Children's National Medical Center. She received a B.S. in chemistry and a B.A. in biology from Saint Louis University (St. Louis, MO), a Ph.D. in molecular biology and biochemistry from the University of Nebraska, and a M.D. from the University of Nebraska, College of Medicine. She subsequently trained in internal medicine and pediatrics at the University of Pittsburgh Health Sciences Center in Pittsburgh, PA, clinical genetics and clinical biochemical genetics at Children's Hospital of Philadelphia, Philadelphia PA. She is board certified in internal medicine, pediatrics, clinical genetics and clinical biochemical genetics. She divides her time between clinical activities in genetics and metabolism at Children's National Medical Center and basic science research exploring energy metabolism in patients and their cells with Propionic acidemia with the aim of improving therapies and morbidity in organic acidemias and other energy deficiency inborn errors.

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We stand for children.

+ George **DIAZ** MD

Dr Diaz completed residency and fellowship training at the Mount Sinai Medical Center in New York and is currently an Associate Professor of Genetics and Genomic Sciences and Pediatrics at the Icahn School of Medicine at Mount Sinai. Dr. Diaz is involved in multiple aspects of Mount Sinai's mission of providing clinical care and training in Medical Genetics as the Associate Director of the Program for Inherited Metabolic Diseases and the Program Director of Medical Genetics training programs. Dr. Diaz has been very active in the area of inborn errors of metabolism research. He is a site Principal Investigator for the Rare Diseases Clinical Research Network Urea Cycle Disorders Consortium (UCDC) led by Dr. Mark Batshaw and will open a site for the Short and Long-Term Outcome of N-Carbamylglutamate Treatment in Propionic Acidemia and Methylmalonic Acidemia studies led by Dr. Mendel Tuchman. In addition to his work in inborn errors of metabolism, Dr. Diaz's laboratory has identified the genetic basis of several inherited single gene disorders and has characterized the disease pathophysiology of these conditions by using animal models and cell biology approaches. He is also actively involved in the translation of genomic sequencing data to clinical applications as Medical Director of the Mount Sinai Medical Genetics Testing Laboratory.

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+ Carol **GREEN** MD

Dr. Carol Greene received her M.D. from Albert Einstein College of Medicine, completed her training in pediatrics at the Children's Hospital of Los Angeles, and her training in genetics at Stanford Medical Center. From 1987 to 2000 she directed the Inherited Metabolic Diseases Clinic at The University of Colorado Health Sciences Center and the Children's Hospital in Denver. For seven years chaired the Colorado Newborn Screening Advisory Committee. In 1999 she was the American Society of Human Genetics Fellow through the AAAS, and as a Congressional Fellow worked for the public health subcommittee of the Senate Health, Education, Labor and Pensions Committee. From 2000 to 2004 she divided her time between 1) the Office of the Assistant Secretary for Planning and Evaluation in the Department of Health and Human Services, and 2) clinical work at the Children's National Medical Center in Washington, D.C. Since August 2004 she has been at the University of Maryland where she is a Professor of Pediatrics and of Ob/GYN and directs the Genetics Clinic in the Division of Genetics in the Department of Pediatrics and co-directs the Adult Genetics Clinic, and is Medical Director for the Masters in Genetic Counseling Training Program. She works with the CDC on quality of testing for genetic conditions. She is active in various professional societies and is the immediate Past-President of the Society for Inherited Metabolic Disorders.

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+ Andrea L **GROPMAN** MD, PhD, FACMG

Dr. Andrea Gropman is an associate professor of Neurology and Pediatrics at the George Washington Medical School and Health Sciences Center. She is also an attending neurologist at the Children's National Medical Center in Washington, D.C. as well as a special expert/consultant in the Medical Genetics Branch at the National Human Genome Research Center at the National Institutes of Health, Bethesda, M.D. She received a B.A. in biology and biochemistry from Brandeis University, and an M.D. from the University of Massachusetts Medical School where she received a Hewlett Packard Top Scholar Award. She subsequently trained in pediatrics at the Johns Hopkins Hospital in Baltimore, M.D., Neurology/Child Neurology at George Washington University and the Children's National Medical Center in Washington, D.C., and Genetics at the National Institutes of Health. She is board certified in pediatrics, neurology with special competence in child neurology, developmental disabilities, genetics and board eligible in biochemical genetics. She divides her time between clinical activities in neurology and genetics at both Children's hospital and the NIH where she works on various metabolic protocols, including MMA with Dr. Chuck Venditti, teaching and research and serves on a number of scientific advisory committees and review groups both at the institutional, governmental, as well as national level.

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+ Charles P **HEHMEYER**

Drawing on a wealth of detailed medical knowledge, Charles Hehmeyer has focused his practice on medical negligence, with a special dedication to one group of clients. He is the only lawyer in the U.S. who routinely represents families - all across the country - who have a child suffering from a metabolic disorder. He has become one of the most vocal leaders in the fight against these tragedies. Thanks to his work with support groups and affected families nationwide, Mr. Hehmeyer has prompted profound improvements in newborn medical diagnosis and care.

Genetic metabolic disorders, if diagnosed in infancy, can be successfully treated with special diets or other therapy. If undiagnosed, however, they can lead to severe retardation and even death. For several decades, all infants in the United States have been routinely screened for many of these disorders—PKU (phenyketonuria) is perhaps the best known—but not all of them. The most sensitive test involves a technology called tandem mass

CONFERENCE **SPEAKERS**

spectrometry that can save lives; yet at present, a number of states refuse to send newborn blood for this special testing. Mr. Hehmeyer wrote an article focused on this injustice, which the editors of Exceptional Parent Magazine sent to all U.S. Senators and each state's director of public health. Mr. Hehmeyer has been quoted in national publications discussing newborn screening.

One of Mr. Hehmeyer's first encounters with metabolic disorders was when he was called upon to defend an Amish family wrongly accused of murdering their daughter. While the charges were pending, the parents lost custody of all of their children. The family's doctor thought that the little girl might have died from a metabolic disorder. The doctor and the family asked Mr. Hehmeyer to get involved. The investigation revealed that the girl had a bleeding disorder that went undetected and untreated because a midwife failed to give her a vitamin K injection at birth. The parents were exonerated and reunited with their children. Smithsonian Magazine profiled that case in a 2006 article.

An impassioned advocate for the children and their families, Mr. Hehmeyer works also to change hospital practices. One month after Mr. Hehmeyer filed suit against a Philadelphia area hospital for failing to screen a newborn child who had a readily treatable metabolic disorder, the Hospital announced that it would change its practices. Click here to read article. Shortly thereafter, all Philadelphia hospitals changed their policies to include screening. A recent California case filed by Mr. Hehmeyer and co-counsel in San Francisco resulted in a front-page article in the Wall Street Journal and helped change the law in California.

After starting his career at Schnader Harrison, Mr. Hehmeyer established his own firm largely to focus on these cases. Raynes McCarty sought his help in representing a child with a metabolic disorder, and not long after that he accepted an invitation to join the firm full-time. Mr. Hehmeyer has earned the highest rating in legal ability and ethics from Martindale-Hubbell.

EDUCATION: Ohio State, University of Pennsylvania Law School

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+ Brian **KIRMSE** MD

Dr. Kirmse is a pediatric geneticist in the Division of Genetics & Metabolism at Children's National in Washington, DC where he is also an Assistant Professor of Pediatrics at George Washington University. He graduated from the University of Miami School of Medicine in 2001 and completed his Pediatrics training at the University of Florida/Shands Hospital. Dr. Kirmse completed his fellowships in Clinical and Clinical Biochemical Genetics at Mount Sinai School of Medicine and joined Children's National in 2010. Dr. Kirmse's research focuses on the adverse effects of HIV medicines on intermediary metabolism, including fatty acid oxidation, in newborns and children. Part of Dr. Kirmse's clinical role at Children's National is as Director of the Program for Newborn Genetics & Metabolism which is dedicated to ensuring that all newborn screen-positive infants in the Washington Metro Area are cared for in an expert and expeditious way.

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+ Melanie **LOCKHART**

Melanie Lockhart is the Director of State Affairs for the March of Dimes National Office of Government Affairs and is responsible for directing the Foundation's state advocacy program comprising 51 state chapters and up to 40 state lobbyists. She also directs grassroots support for the Foundation's federal lobbying efforts. Prior to joining the Office of Government Affairs, she directed the March of Dimes Central Texas Chapter and state advocacy in Texas. Under her direction at the national level, the Foundation has annually exceeded their legislative and regulatory goals in maternal and child health and non-profit policy.

Lockhart was a founding member of the National Birth Defects Prevention Network and currently serves on the Executive Committee, Parent Advisory Group and Advocacy Committee. She served on the Steering Committee of the National Governors Association Learning Network to Improve Birth Outcomes and has presented at meetings of the National Conference of State Legislatures, Pew Home Visiting Campaign, Independent Sector and the American Academy of Pediatrics.

Lockhart has served as a consultant to the NOW Legal Defense and Education Fund and the Hogg Foundation for Mental Health, and directed the Texas Association Concerned with School Age Parenthood and the Texas Communities in Schools Teen Parent Program. She's co-authored reports to the Governor of Texas on adolescent pregnancy, served as an advisor to the Texas Medical Association on adolescent health issues, managed a private medical practice, and began her career at the University of Texas Medical Branch in the Department of Obstetrics and Gynecology.

Melanie Lockhart
Director, State Affairs
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+ Peter J **McGUIRE** MD, PhD, FAAP

Dr. Peter J. McGuire is a currently Physician-Scientist at the National Human Genome Research Institute of the National Institutes of Health in Bethesda.

Dr. McGuire received his medical degree with honors from the Royal College of Surgeons in Ireland. He completed a combined Pediatrics/Medical Genetics Residency at Mount Sinai Medical Center. During his time at Mount Sinai, he was awarded a fellowship in Biochemical Genetics by Genzyme and the American College of Medical Genetics and became a Children's Health Research Center Scholar. After completing his training, Dr. McGuire remained at Mount Sinai as an Assistant Professor in the Department of Genetics and Genomic Sciences and an Attending Physician in the Program For Inherited Metabolic Disorders. In 2011, he joined the faculty at the National Human Genome Research Institute of the National Institutes of Health (NIH). At the NIH, Dr. McGuire's translational research program focuses on metabolic decompensation due to infection and the function of the immune system in patients with inborn errors of metabolism. He is the Principal Investigator for The NIH MINI Study, a ground-breaking effort at the NIH Clinical Center aimed at understanding immune function in patients with inborn errors of metabolism (www.genome.gov/mini).

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+ Carla **MESSENGER** PhD, PLLC

Dr Carla Messenger is a licensed Clinical Psychologist in Virginia. She completed her PhD in Clinical Psychology from The George Washington University in 2005. She was a Postdoctoral Fellow in the Developmental Clinic at Children's National Medical Center. Since 2007, she has practiced independently in Fairfax and now in Arlington. She has also been an adjunct Clinical Assistant Professor at The George Washington University.

Dr Messenger has extensive experience working with clients of all ages who struggle with anxiety, attention deficit disorders, learning disabilities, and chronic medical illnesses. She teaches clients practical coping strategies, so that individuals can manage their symptoms while leading a productive life. She educates family members on how to be most supportive. Her goal in treatment is to empower people through problem-solving, coping, and instilling hope.

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CONFERENCE **SPEAKERS**

+ Jana **MONACO**

Jana Monaco, the mother of four children, has two with an inborn error of metabolism, Isovaleric Acidemia. Stephen has severe neurological impairments and complex health issues due to a late diagnosis. Caroline was tested before and after birth and provided treatment to prevent complications. She is the Advocacy Liason for the Organic Acidemia Association providing support to families with children affected with inborn errors of metabolism.

Ms. Monaco, a former member of the Secretary's Advisory Committee for Heritable Disorders and Genetic Diseases in Newborns and Children, and Co -Chair of its Subcommittee for Education and Training, is currently a member of its Follow Up and Treatment Subcommittee. Jana, a member of the Virginia Genetics Advisory Committee and the NYMAC Regional Collaborative Advisory Council was instrumental in passing state legislation for expanded newborn screening in March of 2005 and legislation in 2014 declaring Rare Disease Day in Virginia every February 28th. Her workgroup involvement includes the NICHD, NYMAC, the SACHDNC workgroup for a "National Policy Regarding the Retention and Use of Residual Dried Blood Spot Specimens Remaining After Newborn Screening" and the MCHB National Medical Home workgroup.

Ms. Monaco worked as a Family Faculty Member of the DC LEND Program at Children's National Medical Center and is currently the chair of the hospital Patient/Family Advisory Council promoting and advancing patient and family centered care.

Jana has presented on newborn screening on Capitol Hill and at various conferences including NORD, Genetic Alliance, the FDA and March of Dimes in addition to the renaming ceremony for the Eunice Kennedy Shriver National Institute for child Health and Human Development. Jana served as a panelist for the NBS 50th Anniversary Celebration sponsored by the APHL in September 2013 and is featured in "The Newborn Screening Story; How One Simple Test Changed Lives, Science and Health in America." She is published in Exceptional Parent Magazine, "My Poster Family" and has given media interviews with Newsweek Magazine, The Wall Street Journal, The Potomac News, Ivanhoe Broadcasting, CBS, NBC and ABC news.

Ms. Monaco, a recipient of the MCHB Director's Award and the Prince William County Board of Supervisors Commendation, received her degree in Therapeutic Recreation from Temple University.

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+ Matthew **RASBERRY** RD, LD, CNSC

Matthew Rasberry is an outpatient metabolic dietitian in Genetics and Metabolism Clinic at Children's National Medical Center (CNMC) in Washington D.C., where he exclusively works with individuals with inborn errors of metabolism. The Genetics and Metabolism Clinic at CNMC has a diverse population of inborn errors of metabolism; including a significant population of organic acidemias and fatty acid oxidation disorders. He received a B.S. in Dietetics from the University of Wisconsin – Madison and underwent his dietitian internship at Johns Hopkins in Baltimore, MD. He received training in the area of nutritional management for inborn errors of metabolism at the Waisman Center in Madison, Wisconsin. He is a registered dietitian through the Academy of Nutrition and Dietetics, a Licensed Dietitian in Washington, D.C., and a Certified Nutrition Support Clinician. He is on the Genetic Metabolic Dietitian International Workgroup to develop nutritional guidelines for Propionic Acidemia.

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+ Suzanne **RIPLEY** PhD

Suzanne is the founder of Accessing Disability Services and brings an authentic perspective on the lives, services and needs of individuals with disabilities, their families, and those who work for and with them. She has worked in the field for many years, both professionally and as a volunteer, and is the parent of two sons with a severe form of Glutathione synthetase deficiency resulting in multiple and complex disabilities. The guys are now adults living in the community -- with a lot of excellent supports.

Suzanne knows the needs, the questions, the frustrations and the joy of finding out what works best. She's been through the ups and downs and learned a lot along the way. She has met wonderful people and survived the ones who weren't all that wonderful.

On the professional side, Suzanne served as Director of NICHCY, the National Dissemination Center for Children with Disabilities, and later as Director of KIN, the national Family Support Center on Disabilities: Knowledge & Involvement Network. She is an instructor with DC Advocacy Partners, a program based on the Partners in Policy Making model. She was also a high school English teacher and taught at the Community College level. She has served on many local, state and national committees and advisory boards and has benefited from learning a great deal about many disability resources across the nation.

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+ Marshall **SUMMAR** MD

Dr. Marshall Summar is the Chief of the Division of Genetics and Metabolism and the Margaret O'Malley Chair of Molecular Genetics. He joined Children's National from Vanderbilt University School of Medicine, where he directed the Program in Translational Genetics, the DNA Core program, and started the inborn errors of metabolism program. Dr. Summar is board-certified in Pediatrics, Clinical Genetics, and Biochemical Genetics.

Dr. Summar is an expert in translational studies, taking basic molecular genetics research and developing direct clinical applications. His work has piloted treatments from the rare disease field to common conditions, especially in the intensive care and emergency room setting. His work in the urea cycle has involved the development of treatment protocols, translational research, and basic molecular research into these rare defects in nitrogen metabolism. This involves clinical trials to improve the outcomes of patients with congenital heart defects, acute lung injury, asthma, and premature infants using compounds from metabolic pathways he studies.

Currently, the focus of Dr. Summar's research is the study of the interactions between common genetic variations and the environment. This work involves research in heart disease, asthma, pulmonary hypertension, oxidant injury and aging, Down syndrome, and liver disease.

Dr. Summar serves on the editorial board of The Journal of Pediatrics and is the president-elect for the Society of Inherited Metabolic Disease. He serves on the NIH study section for the CETT program, the National Human Genetic Research Institute Fellowship Training Program Board, and the NASA radiation research review panel.

Marshall Summar, MD
Children's National Medical Center
Division Chief, Genetics and Metabolism

CONFERENCE **SPEAKERS**

+ Jennifer **SLOAN** PhD, MS, CGC

Dr Sloan is currently working at the National Human Genome Research Institute (NHGRI) as the genetic counselor and protocol coordinator for the methylmalonic acidemia (MMA) and cobalamin disorders study with Dr. Venditti. She received her bachelor of science in Biology from Penn State University and went on to complete a PhD in Neurobiology. Dr. Sloan was interested in pursuing more clinically relevant work, which led her to pursue a Master's program in genetic counseling shortly after finishing her doctoral dissertation. She received a M.S. in Genetic Counseling from Northwestern University in 2004 and has been at the NHGRI since that time. Dr. Sloan became a board-certified genetic counselor in 2005. She is interested in genetic counseling issues in metabolic disorders and identifying new disease genes.

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+ Charles **VENDITTI** MD, PhD

Dr. Charles P. Venditti is an Investigator in the National Human Genome Research Institute and the Director of the Organic Acid Research Section at the National Institutes of Health in Bethesda, MD. He received his BS from and was an MD, PhD scholarship recipient at Penn State University. After graduation, he completed a pediatrics residency at Massachusetts General Hospital/Harvard Medical School and a combined clinical and biochemical genetics fellowship at the Children's Hospital of Philadelphia/University of Pennsylvania School of Medicine. He is board-certified in pediatrics, clinical and biochemical genetics and is an Attending Physician at the Mark O. Hatfield Clinical Center at the NIH, where he has initiated a translational research program to study the natural history and clinical phenotype(s) of the hereditary methylmalonic acidemias (MMA) and cobalamin metabolic disorders. The clinical research studies are complemented by laboratory investigations that have focused on the development of experimental systems to study genetic, genomic and biochemical aspects of organic acid metabolism in model organisms, including roundworms, mice and zebrafish. Using a translational research approach, Dr Venditti and his colleagues have published a number of papers that connect disease pathophysiology in MMA to mitochondrial dysfunction and prove the efficacy of gene therapy as a treatment for both methylmalonic acidemia and propionic acidemia. Dr Venditti is a member the American Society of Clinical Investigation (ASCI) as well as numerous professional organizations in the fields of inborn errors of metabolism, genetics and gene therapy. In 2009, he was the recipient of a Presidential Early Career Award for Scientists and Engineers (PECASE), the US Government's highest honor for early-career scientists. He serves on the medical advisory board of the Organic Acid Association (OAA) and as a member of the Scientific Review Committee for the NHGRI IRB. He has authored and co-authored more than 70 research articles, clinical reports, patent applications and textbook chapters.

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+ Hilary **VERNON** MD, PhD

Dr. Vernon received her MD from Robert Wood Johnson Medical School and her PhD from Rutgers University. She trained in Pediatrics, Medical Genetics, and Biochemical Genetics at Johns Hopkins University. She is a former Chief Resident of Medical Genetics at Johns Hopkins University, and a past recipient of the James Sidbury Fellowship in Biochemical Genetics. Dr. Vernon has been on the faculty at the McKusick-Nathans Institute of Genetic Medicine at Johns Hopkins University and at the Kennedy Krieger Institute in Baltimore, MD since 2011. She is the co-director of the Barth Syndrome Interdisciplinary Clinic at the Kennedy Krieger Institute and is also currently serving on the newborn

screening advisory council in Maryland. Dr. Vernon is a clinician-researcher whose main areas of research are in organic acidemias including propionic acidemia, methylmalonic acidemia and Barth Syndrome (3-methylglutaconic aciduria type II). Her research employs both metabolic and molecular approaches towards understanding how a single enzymatic defect causes a biochemical "ripple-effect" throughout the cell.

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+ Lynne A. **WOLFE** MS, PNP, BC

Lynne has been a Nurse for over 30 years and a Nurse Practitioner working with children and adults who have all types of Inborn Errors of Metabolism and Mitochondrial diseases for 15 years. Currently, Lynne is a Nurse Practitioner & Co-Investigator in the Undiagnosed Diseases program, a Co-Investigator for the Epi-743 trial, and a Co-Investigator in the Congential Disorders of Glycosylation Natural History study at National Institutes of Health. Her areas of interest include: Mitochondrial Disease/dysfunction, Nutrition and Supplement support for metabolic and mitochondrial diseases, and Transitional Care all areas she has also published in. She is speaks frequently to professional and family support groups.

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+ Wadih **ZEIN** MD

Wadih Zein, M.D. is a staff clinician in the Ophthalmic Genetics and Visual Function Branch at the National Eye Institute (NEI). He obtained his M.D. from the American University of Beirut and completed his ophthalmology residency at the American University of Beirut Medical Center. He completed a fellowship in pediatric ophthalmology and strabismus at the Children's National Medical Center, Washington DC, and a fellowship on ophthalmic genetics and visual function NEI.
Dr. Zein became an NEI employee in 2008 and specializes in the care of patients with pediatric and hereditary eye diseases. He sees patients-both children and adults-with a broad array of ocular and systemic disorders such as achromatopsia, albinism, aniridia, Best disease, Cohen syndrome, gyrate atrophy, Joubert syndrome, Leber congenital amaurosis, methylmalonic acidemia, mucolipidosis, retinitis pigmentosa, Stargardt disease, and Usher syndrome.

Dr. Zein recognizes the effect that many inherited eye diseases can have on the lives of the patients and the lack of adequate treatment options. He is interested in studying the natural history of hereditary eye diseases as well as developing outcome measures to be used in future treatments.

+ William **ZINNANTI** MD, PhD

Dr. Zinnanti graduated from UCLA with a degree in molecular cell and developmental biology. He then completed the Physician Scientist Training Program at Pennsylvania State College of Medicine where he earned both doctorates in Medicine and Neuroscience. Dr. Zinnanti participated in post-doctoral studies in neurobiology at Cal Tech, resulting in the discovery of potential treatments for two rare metabolic disorders. He then completed an intership in pediatrics at State University of New York, Brooklyn and fellow ship in neurology at Stanford University, Lucile Packard Children's Hospital. Dr Zinnanti is a licenced physician and neurometabolic expert.

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