

Cystinosis Research Network Family Conference
July 19 – 21, 2007
The Hotel Contessa, San Antonio, Texas
Sponsored in part by Sigma Tau Pharmaceuticals
“Searching for a Cure.....Deep in the Heart of Texas”



Scheduled Speakers, Poster Session Participants, and Guests



Corinne Antignac, M.D., Ph.D.

Degrees and titles: Medical Doctor, Paris 6 University, 1982; Certification in Pediatrics, Paris 5 University, 1988; Certification in Nephrology, Paris 5 University, 1988; Ph. D. (Human Genetics), Paris 6 University, 1994

Positions Held: 1976-1983: Resident in Pediatrics and Nephrology, Paris, France; 1983-1986: Assistant - Pediatric Nephrology Unit (Pr M. Broyer), Necker Hospital, Paris, France; 1986-1990: Fellowship at Inserm; 1990-1996: Researcher at Inserm (CR1); 1996-August 2001: Research Director at Inserm (DR2 – DR1 from January 2001); September 2001: Professor (PU-PH) in the Department of Genetics, Necker Hospital, Paris France; January 2003: Director of the Inserm laboratory Unit 574 (Hereditary Nephropathies and Kidney Development)



Lynne Babchuck has been working with the UCSD Pediatric Neurology Research Group, under the direction of Dr. Doris Trauner since July, 2003. Since that time she has coordinated studies examining the neurological implications of cystinosis. She received her BA in Psychology from the University of Vermont in 2002 and her Masters Degree in Clinical Social Work in 2007.



Jodie Badgley has been working with UCSD Pediatric Neurology Research Group, under the direction of Dr. Doris Trauner since September of 2005. Since that time she has been coordinating a study examining cognition and brain development in children and teenagers with a history of pre-natal and neonatal strokes. She received her BA in English from Saint Mary's College in 2004 and previously worked at the United States Department of Education in Washington D.C.



Leticia Belmont, M.D., studied medicine at the National University of Mexico. She did medical pediatrics postgraduate work at the Pediatric Hospital Medical Center SXXI (Mexican Institute of Social Security). She took an advanced course of Internal Medicine at the National Institute of Pediatrics Health Services in Mexico City. She has participated in research studies and the clinical care of patients with metabolic diseases for five years. Currently she is an Investigator Assistant in Medical Sciences in the Nutrition Genetics Unit of the National Institutes of Pediatrics. She is an Internist Pediatrician in the Hospitalization Service of the National Medical Center SXXI, President of the Pediatrics Association of the Mexican Institute of Social Security, and has served as Medical Director of the Cystinosis Foundation Mexico since 2002. She works as a Professor Assistant in a pediatric course, and a coordinator and assessor in different academic activities in Mexican pediatric topics. She has given different talks about Cystinosis and metabolic diseases nationally and internationally. She has shared her experience with Cystinosis in San Diego, Ann Arbor, Colombia, Argentina, Spain, Japan, and Los Angeles. She currently participates in the National Center of Reference, Inborn Inherited Metabolism Control follow up in cystinotic patients.



Mark Benfield, M.D., received his undergraduate and Medical School education at Tulane University. He then completed his internship, residency and chief residency at the University of Alabama at Birmingham, and a fellowship in Pediatric Nephrology at the University of Minnesota. He is now the director of Pediatric Nephrology at the University of Alabama at Birmingham with special clinical and research interests in chronic kidney disease, dialysis and transplantation.



Donald Cairns was born in Glasgow, Scotland and attended the University of Strathclyde where he read Pharmacy. Following qualification, he worked in a number of hospitals then returned to Strathclyde to complete his doctorate researching into long-acting local anaesthetics and tranquillisers. In 1986, Dr Cairns was appointed post-doctoral research fellow for the UK Cancer Research Campaign where he developed computer protocols for the molecular modelling of drug interactions with DNA. In 1987, he received a lectureship in the School of Pharmacy at Leicester and in 1992 was appointed to the post of Senior Lecturer in Medicinal Chemistry at the University of Sunderland. In October 2003, Dr Cairns was appointed Associate Head of the School of Pharmacy in Aberdeen. In addition to his work on cystinosis, Dr Cairns has research interests in the rational design of anticancer drugs and molecular modelling of drug interactions with higher-order DNA. He has published over 50 research papers and is the author of a best-selling undergraduate textbook on Pharmaceutical Chemistry.



Ranjan Dohil, M.D. is an Associate Professor in Pediatric Gastroenterology at the University of California, San Diego. Dr. Dohil graduated in medicine in the United Kingdom, and trained in Pediatrics and Pediatric Gastroenterology in Cardiff and

London in the United Kingdom and in Vancouver, Canada. Dr. Dohil has been interested in acid related disorders of the stomach in children.

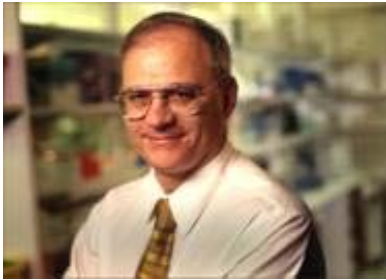


Dr. Ewa Elenberg completed Pediatric Residency training at Miami Children’s Hospital and Pediatric Nephrology Fellowship training at Massachusetts General Hospital, Harvard Medical School in Boston. She is Board Certified in Pediatrics and Pediatric Nephrology. Since 1997, she has been living in Texas. Currently, she is an Assistant Professor in Pediatrics, Renal Section, at Texas Children’s Hospital, Baylor College of Medicine, in Houston, TX. In 1994 during her Fellowship training in Boston, she became interested in Cystinosis after meeting a patient who had severe feeding dysfunction and received nutrition via total parenteral nutrition. This inability to eat inspired her to develop the survey that helped her identify the spectrum of feeding problems in cystinotic patients. The article, “Feeding Problems in Cystinosis”, was published in *Pediatric Nephrology*, vol.12, No.5, pp. 365-370, June, 1998. She is also an author of the Cystinosis chapter in *E-Medicine*. In 1995, she received an award from the Cystinosis Foundation “In grateful recognition of special concern for and outstanding service to children with Cystinosis”. Since 2003, she has been a member of the Medical Advisory Board of the Cystinosis Research Network. Dr Elenberg is a passionate educator in Cystinosis, presenting at national and international meetings. She currently treats 4 patients with Cystinosis.



Katy Freed, Ph.D., was born in Melbourne, Australia and has lived there for most of her life. She received her Bachelor of Science (Honours) degree (Biochemistry major) from the University of Melbourne in 1985. For the next few years she undertook research in Melbourne, followed by a year working at the Hopital des Enfants Malades, in Paris, France. For the last 13 years she was based at the Royal Women’s Hospital, Melbourne, Australia, where her primary research focus was trying to identify genes that contribute to human pregnancy disorders, such as preeclampsia. To that end she completed her PhD at the University of Melbourne in 2004, focusing on the genetics of preeclampsia. In December 2006, she moved to San Antonio, Texas to work at the Southwest Foundation for Biomedical Research. In January 2007 she received a 2 year Post-Doctoral Fellowship from the Cystinosis Research Foundation to

work on a project entitled “Complex genetic approaches to monogenetic disease: Genomic and transcriptomic dissection of normal expression of *CTNS*, the gene involved in nephropathic cystinosis”.



Dr. William Gahl graduated from MIT in 1972 and received his M.D. and Ph.D. from the University of Wisconsin. He is board-certified in Pediatrics, Clinical Genetics, and Clinical Biochemical Genetics and is past president of the Society of Inherited Metabolic Disorders. He is a member of the American Society for Clinical Investigation and the Association of American Physicians. Dr. Gahl is an international expert in cystinosis, Hermansky-Pudlak syndrome, disorders of sialic acid metabolism, and alkaptonuria. He has trained 19 fellows certified in Clinical Biochemical Genetics. All of Dr. Gahl's work is accomplished with the support of his wife, Mary, with whom he has 4 children and 34 years of marriage.



Completing a doctorate in Counselor Education from the University of Florida in 1982, **David Glaize** currently serves as Director of Assessment in the Division of Student Success Services at Seminole Community College, Sanford, Florida. With cognate study in assessment, and career development, Dr. Glaize has worked professionally as a middle school, high school, adult education, and community college counselor; he has served as a high school Director of Guidance, a community college administrator, and has taught courses in assessment, psychology, and sociology at both the college and university levels. In his present position, he provides consultation and testing for the college's career assessment program, supervises all college placement entry and exit testing, GED and state licensing/certification programs, and provides leadership and supervision for Seminole Community College's services for students with disabilities. Dr. Glaize is a member of the Cystinosis Research Network's Professional Advisory Committee. Dave and his wife, Sandy, have been married for 31 years and have two daughters, Whitney, who is completing her Masters degree from the University of Central Florida in Speech/Language Pathology, and Leslie, who is graduating from the University of Florida with a Bachelors degree in Finance, both completing their studies in the summer of 2003. They live in Orlando, Florida.



Whitney Glaize has been practicing in the field of Speech-Language Pathology for the last 5 years. She is employed by Princeton House Charter School which provides educational services for autistic children aged 3-18 years old. She also does private speech therapy in a private practice, and works as an independent contractor, providing speech language services to children of all ages. Whitney graduated with a Bachelors degree in Speech Pathology from University of South Florida and a Masters degree in Communication Disorders from University of Central Florida. She is a Certified Clinical Speech Language Pathologist (CCC-SLP), has a Florida state license, a Florida's teacher certificate, a Medicaid provider license, and is a national provider of Speech Language Pathology . After entering the field and remembering her own struggles with eating as a child, she is pursuing this area of research and has been attending workshops on feeding and swallowing disorders in infants, toddlers, and children. She hopes to one day improve the quality of life for children who have feeding and swallowing difficulties as it relates to Cystinosis. Whitney was diagnosed with Cystinosis in 1981 at age 2 years old.



Dr. Paul Goodyer is James McGill Professor of Pediatrics and Director of Pediatric Nephrology at the Montreal Children's Hospital. He was born in New Haven, Connecticut, attended Harvard University and obtained his MD at the University of Rochester in Rochester, New York. He trained in Pediatrics at McGill and Harvard and then completed his clinical training in Pediatric Nephrology and postdoctoral research training in Biochemical Genetics at McGill. He joined the staff of the Montreal Children's Hospital as a Pediatric Nephrologist in 1980 and, soon afterwards, initiated a specialized Renal-Genetics Clinic where he has cared for about 30 children with cystinosis; many of these children were involved in the North American trial of cysteamine. The Renal Genetics Program at the Montreal Children's Hospital serves as the Canadian reference centre for leukocyte cystine monitoring. Dr. Goodyer directs a basic research laboratory focused on the molecular basis of hereditary renal diseases. He has written over 100 original research articles, book chapters and reviews; these include articles on the molecular genetics of cystinosis in Quebec and the outcome of renal transplantation in cystinotic children.



Dr. David H. Ivey, II graduated from Sherman Chiropractic College of Spartanburg, South Carolina in March 2001. He opened his multi-disciplinary chiropractic office in the Greenville area in September 2002. This year Dr. Ivey became 1 of 900 doctors in the world to earn the title of Certified Chiropractic Extremity Practitioner. Dr. Ivey owns a family-oriented chiropractic practice. It is through his work with children that Dr. Ivey had the pleasure of meeting Laura and her family. After a thorough examination and briefing Dr. Ivey decided that Laura would be a great candidate for customized orthotics. Laura has since had improvement in the areas of leg pain, fatigue and muscle spasms.



Vasiliki Kalatzis, Ph.D., received her Bachelor of Science degree from the University of Adelaide in Australia. She moved to Paris, France, in 1993 and performed her Ph.D. training at the Pasteur Institute where she was involved in cloning the gene responsible for branchio-oto-renal syndrome. She began working on cystinosis in 1998 when she joined Pr Corinne Antignac's laboratory as a post-doctoral fellow. Pr Antignac's team had just cloned the gene responsible for cystinosis and Dr Kalatzis was involved in determining the function of the defective protein and correlating the molecular defect with disease severity. She also participated in the characterisation of disease mutations and generation of a cystinosis animal model. In 2001, Dr Kalatzis moved to Montpellier, France, where her research is centred on the further characterisation of cystinosis mice and studying the feasibility of gene therapy as a complementary treatment.



Robert Kleta, M.D., Ph.D., received his clinical training in Pediatrics (Board certified) and Pediatric Nephrology in Hamburg, Luebeck, Berlin, and Muenster, Germany; clinical training in Biochemical Genetics (Board certified) in Bethesda, MD, USA; research training in Renal Physiology in Freiburg, and Muenster, Germany; and research training in Genetics in Bethesda, MD, USA. He is presently, Potter Chair of Nephrology at University College London / UK. Main interests: Clinical and molecular findings in children and adults with rare inherited disorders of the kidney and other metabolic disorders; e.g., Hartnup disorder, Iminoglycinuria, renal Fanconi syndromes, Bartter syndromes, Gitelman syndrome, Hypomagnesemia, and Cystinosis.



Craig B. Langman, MD is the Isaac A. Abt, MD Professor of Kidney Diseases at the Feinberg School of Medicine, Northwestern University, and Head, Kidney Diseases, Children's Memorial Hospital, Chicago IL. Additionally, Dr. Langman holds the Mary Weston visiting professorship at the University of Natal, Durban South Africa. Dr. Langman has authored over 175 original publications, reviews, chapters, web-materials, and commentaries, principally in the area of bone and mineral metabolism. His clinical and research interests involve genetic and acquired forms of metabolic bone disease, nephrolithiasis, and genetic kidney diseases in infants, children, adolescents, and transitioning young adults. Dr. Langman serves as the Senior Associate Editor for the American Journal of Nephrology; Associate Editor for each of the six editions of the Primer on the Metabolic Bone Diseases from the American Society of Bone and Mineral Metabolism; Editorial Board for Journal of Bone and Mineral Research; Journal of Renal Nutrition; Section on Genetic and Tubular Diseases, Editorial Board for Pediatric Nephrology, and is the only North American on the Editorial Board of the European Journal of Pediatrics. Dr. Langman serves as Medical Editor and Board Member for the Oxalosis and Hyperoxaluria Foundation; and is on the Scientific Boards of the Cystinosis Foundation and the Cystinosis Research Network. He helped found 'Cystinosis Mexico', for which he continues ongoing support. He serves on the KDIGO Initiative for CKD-MBD. He serves on the K/DOQI Advisory Board, was the co-chair of the K/DOQI guidelines for the care of pediatric renal osteodystrophy, and served on the K/DOQI committees for adult renal osteodystrophy and nutrition in chronic renal failure guidelines. Dr. Langman is the Chairperson of the 4th International Conference on Children's Bone Health in Montreal, June 21-24, 2007 (www.iccbh.org).



Bola Lawal, Ph.D., graduated from The School of Pharmacy, University of London in 1998. After a year's internship at The Hillingdon Hospitals NHS trust, she registered with The Royal Pharmaceutical Society of Great Britain. She has worked as a clinical Pharmacist since registration. Her last role was at a leading private hospital in the UK in the capacity of a Pharmacy Manager.

Kelly Martin is Director, Prescription Products & Healthcare Policy. Before joining Sigma-Tau in February 2003, she spent 15 years in the pharmaceutical / biotech industry; working for Merck, Amgen, GAMBRO and Bone Care International. Kelly's experience includes sales, clinical support, training, managed care, and governmental affairs. Martin holds a BS in Biology/Medical Technology from Siena Heights University in Adrian, Michigan. She is currently a Global Executive MBA student at Duke University – The Fuqua School of Business in Durham, North Carolina. Kelly will graduate with her MBA in November 2007 and is continuing her education at Duke by completing the Health Sector Management Program. For the last ten years Kelly has been a member of City Mission, servicing the needs of the Brightmoor Community of inner city Detroit. She is also a member of the Healthcare Business Women's Association and a past board member for the Michigan State Youth Soccer Association. Kelly is still active in a variety of leadership and volunteer programs at Kensington Community Church.



Susie McCalla, is a high school counselor at Center Point High School in Center Point, Texas. She also teaches senior English, sponsors the Dance Team, serves as the competitive prose/poetry coach, and the Travel Club Sponsor, taking groups of high school students to Ireland, England, France and Italy over the last three years. She received her Bachelor of Arts degree in History from the University of Texas in 1976 and was commissioned as a 2nd Lieutenant in the USMC. She also received her Master of Arts degree in Psychology, Guidance and Counseling from the University of Northern Colorado in 1979 while on active duty with the USMC. She received her teaching certificate from San Diego State University in 1983, a Master of Arts degree in English from the University of Hawaii in 1995, and a school counseling certificate from the University of Texas at San Antonio in 1999. Susie has been married to John for 27 years and they have three children – Michael (28), Kevin (22 with cystinosis and married

to Liza), and Kelly (21). All three children have attended the University of Texas at Austin or are currently students there.



Wendy Perrow, MBA is Vice President of Marketing for Sigma-Tau Pharmaceuticals, Inc. In this role, Perrow leads the team responsible for all marketing, managed care, government relations, and public affairs functions. Perrow joined Sigma-Tau in 2004 as Vice President of Marketing. Prior to joining Sigma-Tau, Perrow spent 14 years at Merck and Co., Inc. in the areas of global marketing, US marketing, promotion, international business research analysis, training, and sales. Perrow brings a strong leadership and management dimension to Sigma-Tau's marketing and sales division. A skilled marketer, she has led US and global initiatives for leading products that have significantly increased brand awareness and have resulted in multi-billion dollar sales. Perrow holds a BA from Eastern Illinois University in Charleston, Illinois and an MBA in finance and marketing from Duke University—The Fuqua School of Business in Durham, North Carolina. Perrow is a member of the National Association for Female Executives and a member of the Healthcare Business Women's Association. She is also active in a charitable organization that raises money to provide education for women. She has been a trustee with A Dinner of Hope, Inc. for the past 3 years and serves as the chairperson for A Light of Day Award. She is also active in a variety of community volunteer programs.



Magdalena Pudlo has been working with UCSD Pediatric Neurology Research Group, under the direction of Dr. Doris Trauner since August of 2006. Since then she has been researching cognition, focusing on language development in infants and children with focal lesions. She received her BS in Psychology, with an emphasis in Cognitive Neuroscience from the University of Denver in 2004.



Dr. Poonam Sansanwal is a post-doc scholar working with Dr. Minnie Sarwal at Stanford University. Dr. Sansanwal earned her Ph.D. degree in Biochemistry from the All India Institute of Medical Sciences, India in 2004. During her doctoral research she studied a lysosomal protease, Cathepsin L, having implications in cancer metastasis which involved extensive work in the field of molecular and cell biology. Her research interests include metabolic renal disorders and as a part of the Sarwal group, she is interested in unraveling underlying molecular mechanisms for the pathogenesis of Nephropathic Cystinosis, an inherited lysosomal storage disorder.



Jerry Schneider, M.D. began to study cystinosis in 1965 as a Clinical Associate at the NIH. He joined the Department of Pediatrics at the University of California, San Diego (UCSD) in 1970. In 1995, he became Dean for Academic Affairs at the UCSD School of Medicine, but continued to study cystinosis. Dr. Schneider officially retired last year (2004), but has been recalled by the University as Research Professor of Pediatrics. He continues to direct UCSD's Cystine Determination Laboratory and is spending more time studying cystinosis than before his "retirement." In the last ten years, the Cystine Determination laboratory has diagnosed 144 new cases of cystinosis.



Nicholas W. Smith, M. A., LMHC is a Licensed Child Psychotherapist in the State of Florida who specializes in child/adolescent psychotherapy. He is the Director of New Hope Counseling Centers and Educational Services. He has 17 years experience working with families and children who have neurological, emotional, psychological, behavioral and physical challenges. He earned a Masters degree in Counseling from Rollins College and a Bachelors degree in Religion from Mt. Vernon Nazarene College. He is married 27 years to Linda who is also a Licensed Psychotherapist and Critical Care RN. He has two daughters, Jennifer 26 and Alison 20.



Dr. Jess Thoene is a native of California and attended undergraduate studies at Stanford University, majoring in chemistry. He attended medical school at Johns Hopkins and completed a 3 year fellowship in metabolic genetic disease at the University of California, San Diego. His research interests include investigation of the pathophysiology of cystinosis, and understanding the molecular biology of lysosomal transport. Clinically, he is interested in the diagnosis and treatment of inborn errors of metabolism with particular emphasis on urea cycle disorders, organic acidemias, and cystinosis. He is a member of the American Society for Clinical Investigation, the American Society of Human Genetics, the American Pediatrics Society, the American Society of Biochemistry and Molecular Biology and the American Society of Gene Therapy. He was chairman of the National Commission of Orphan Diseases 1988-1989. He is past chairman of the Board of the National Organization for Rare Disorders. He was a Joseph P. Kennedy, Jr. fellow in Washington, D.C. and is the editor of the Physician's Guide to Rare Diseases. Currently, he is the Karen Gore Professor of Pediatrics, Director of the Hayward Genetics Center/Human Genetics Program at Tulane University Health Sciences Center. Dr. Thoene is President-Elect of the Southeast Regional Genetics Group, an eight state coalition of geneticists, and CoPI of a HRSA grant to study genetic health care delivery and enhanced newborn screening in the region.



Dr. Doris Trauner is Professor of Neurosciences and Pediatrics at the University of California, San Diego School of Medicine, Chief of Pediatric Neurology at UCSD, and a Senior Staff physician at Children's Hospital of San Diego. She received her medical degree from the Medical College of Virginia, completed post-graduate training in Pediatrics at UCSD Medical Center in San Diego, and a Pediatric Neurology fellowship at the University of Chicago. Dr. Trauner is board-certified in Pediatrics; in Psychiatry and Neurology with Special Competence in Child Neurology; and in Neuro-Developmental Disabilities. Her research and clinical interests relate primarily to neurodevelopmental and neurobehavioral disorders. Her specific research interest is in effects of cystinosis on the brain, with special focus on learning and cognition. She has described a specific visual processing deficit in cystinosis that is associated with a certain type of learning disability. She is also interested in the late complication of muscle wasting and weakness that can occur in young adults with cystinosis. Her goal is to develop effective interventions and treatments for these problems. She has over 120 publications.



Jenny Williams started working for Dr. Doris Trauner in the UCSD Pediatric Neurology Research Group in July, 2005. Throughout her time in the lab she has been the study coordinator for studies of cognition and brain development in individuals with cystinosis of all ages. She graduated from San Diego State University in 2005 with a B.A. in Psychology and will return to school next fall to obtain her Doctor of Philosophy degree in the field of Clinical Psychology.



Martijn Wilmer was born on May 19, 1978 in Tegelen, The Netherlands. After graduation from high school in 1996, he started an education at the Fontys School for Higher Laboratory Education, with biochemistry and cell biology as a main topic. For his graduation thesis, he has spent 9 months in the Laboratory of Dr. W.S. Argraves in Charleston, SC, USA. There he worked on the endocytosis of high density lipo-proteins, mediated by megalin-cubilin, two multi-ligand receptors expressed in proximal tubule cells of the kidney. After graduation in 2001, he started working as a research technician in the Laboratory of Pediatrics and Neurology of the St. Radboud University Medical Centre, Nijmegen, The Netherlands. Together with Dr. E.N. Levtchenko and Dr. L.P. van den Heuvel he studied the pathogenesis of cystinosis. During this period, Martijn performed a working visit to Prof. P. Mathieson and Dr. M. Saleem of the Academical Renal Unit of the Southmead Hospital, Bristol UK. Since April 2006, Martijn has been working as a PhD-student at the Laboratory of Pediatrics and Neurology of the St. Radboud University Medical Centre, Nijmegen, The Netherlands. For six months, he has worked in the group of Prof. E.I. Christensen of the Anatomic Institute, Aarhus University, Denmark. Currently, he is working on the development of a cell model for cystinosis for investigating the pathways involved in cystinotic cell damage.

Rosimar Zavarse, LMSW, is a licensed master social worker at Texas Children's Hospital in the Texas Medical Center. As the primary social worker for renal transplant patients and their families, her role is to help them successfully navigate the psychosocial complexities of a chronic diagnosis. Over the past three years she has worked with cystinosis patients in the renal clinic and has been exposed to the unique challenges they face. Rosimar is a member of the National Association of Social Workers and a member of the National Kidney Foundation's Council of Nephrology Social Work.