

# Cystinosis Research Network Family Conference

July 19 – 21, 2007

The Hotel Contessa, San Antonio, Texas  
Sponsored in part by Sigma Tau Pharmaceuticals

**DRAFT 7/03/07**

“Searching for a Cure.....Deep in the Heart of Texas”

## Family Conference Agenda

<b>Thursday, July 19</b>		<b>Room</b>
<b>Times to be determined</b>	<b>Neurological Testing with Dr. Trauner’s team</b>	<b>Retama and Boardroom</b>
<b>12:00 pm – 8:00 pm</b>	<b>Registration / Information Desk</b>	<b>Contessa Ballroom Foyer</b>
<b>2:00 pm – 5:00 pm</b>	<b>CRN Scientific Review Board Meeting</b> <i>Closed Session</i>	<b>Anaqua</b>
<b>5:00 pm – 8:00 pm</b>	<b>CRN Reception</b> <i>Complimentary</i> <i>Cash bar begins at 5:00 pm, appetizers begin at 5:30 pm</i> <i>John and Kim Shepperd, Hosts</i>	<b>Cypress/Cork Bar Atrium/River Walk Patio</b>
<b>Friday, July 20</b>	<b>Cystinosis Diagnosis, Treatment, and Research</b>	
<b>Times to be determined</b>	<b>Neurological Testing with Dr. Trauner’s team</b>	<b>Retama and Boardroom</b>
<b>Times to be determined</b>	<b>Southwest Foundation for Biomedical Research</b> <b>Blood sample collection</b>	<b>Mesquite</b>
<b>8:00 am – 5:00 pm</b>	<b>Registration / Information Desk</b>	<b>Contessa Ballroom Foyer</b>
<b>8:00 am</b>	<b>Child Care Opens</b> <i>Open to the end of today’s agenda</i>	<b>Laurel</b>
<b>6:30 am – 8:00 am</b>	<b>Breakfast Buffet</b> <i>Complimentary</i>	<b>Las Ramblas</b>
<b>8:15 am – 8:30 am</b>	<b>Morning Session Welcome &amp; Opening Remarks</b> <i>John and Kim Shepperd, Hosts</i>	<b>Contessa Ballroom</b>

<b>8:30 am - 9:00 am</b>	<b>Cystinosis Research Network – Your Advocacy Group</b> <i>José Morales, President</i> Meet the CRN Board of Directors and learn more about how CRN works to achieve its stated vision of the discovery of improved treatments and ultimately a cure for cystinosis and mission of supporting and advocating research, providing family assistance, and educating the public and medical communities about cystinosis.	<b>Contessa Ballroom</b>
<b>9:10 am – 10:00 am</b>	<b>Shad Ireland</b> Shad is an individual who has accepted, and is successfully living with kidney disease and all of the experiences that go along with this diagnosis. On July 25th, 2004 he became the first dialysis patient to ever compete in and complete an Ironman triathlon. You can learn more about Shad on his websites at <a href="http://www.shadirelandfoundation.org">www.shadirelandfoundation.org</a> or <a href="http://www.ironshad.com">www.ironshad.com</a> .	<b>Contessa Ballroom</b>
<b>Session A</b>	<b>Infancy and Childhood Lecture Series</b> <i>Elva Smith, Host</i> Participants should attend the one of the two morning lecture series (A or B) which best meets their current interests and needs.	<b>Contessa Ballroom A</b>
<b>10:15 am – 10:45 am</b>	<b>Cystinosis: A General Overview</b> <i>Paul Goodyer, M.D.</i> This talk will cover basic aspects of cystinosis for families who are relatively new to the problem. The aim is to explain something about the nature of cystinosis and enough about the biology of the kidney to understand why affected children need to take supplements like potassium, phosphorus and citrate. Will describe the basic genetics of cystinosis and explain why the illness is more common in some regions (like Quebec) compared to others.	<b>Contessa Ballroom A</b>
<b>10:55 am – 11:25 am</b>	<b>Fanconi’s Syndrome and Its Treatment</b> <i>Ewa Eleberg, M.D.</i>	<b>Contessa Ballroom A</b>
<b>11:35 am – 12:05 pm</b>	<b>Gastrointestinal Issues and Study Update</b> <i>Ranjan Dohil, M.D.</i> Gastrointestinal (GI) symptoms associated with cystinosis and cysteamine therapy are now accepted to be common occurrences. Although we do not understand the underlying mechanisms for many of the GI symptoms, it is likely that some are associated with gastric acid-hypersecretion. During this presentation I will briefly discuss the nature and management of some of the gastrointestinal problems associated with cystinosis. I will also describe the results of a recently completed study which have helped us to better understand how well cysteamine is absorbed from different regions of the intestinal tract and also the effect that this may have on prolonging white cell cystine depletion. Much of the data acquired from this cysteamine absorption study have helped us to design our ongoing study using a twice daily preparation for cysteamine. I will discuss some of the early data from this study.	<b>Contessa Ballroom A</b>
<b>Session B</b>	<b>Adolescence and Adulthood Lecture Series</b> <i>Karen Gledhill, Host</i>	<b>Contessa Ballroom B</b>

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<b>10:15 am – 10:45 am</b>	<b>Transplantation Issues</b> <i>Mark Benfield, M.D.</i>	<b>Contessa Ballroom B</b>
<b>10:55 am – 11:25 am</b>	<b>Ocular Effects and Treatments of Cystinosis</b> <i>William A. Gahl, MD, PhD, NHGRI, NIH, Bethesda, MD</i> Ocular complications of nephropathic cystinosis involve both the posterior and the anterior segments of the eye. Posterior segment involvement can be responsive to oral cysteamine therapy. Specifically, chronic oral cystine depletion is associated with a reduced frequency of retinopathy. Anterior segment involvement responds to cysteamine eyedrops. In particular, the corneal crystals of cystinosis can be dissolved with chronic topical cysteamine therapy.	<b>Contessa Ballroom B</b>
<b>11:35 am – 12:05 pm</b>	<b>Adult Medical Issues in Cystinosis</b> <i>William A. Gahl, MD, PhD, NHGRI, NIH, Bethesda, MD</i> Nephropathic cystinosis is an autosomal recessive storage disease due to defective transport of cystine out of lysosomes. Early findings, at 6-12 months of age, include polyuria, polydipsia, dehydration, acidosis, hypophosphatemic rickets, hypokalemia, hypocalcemic tetany, hypocarnitinemia, and growth retardation. Renal glomerular damage results in end-stage renal disease by 9-10 years of age unless cystine-depleting therapy is initiated early in life. Oral cysteamine therapy has delayed renal deterioration and enhanced growth when taken diligently. Renal replacement therapy has transformed cystinosis from a pediatric disease to one that affects adults as well. Complications include retinal blindness, a vacuolar myopathy, swallowing dysfunction, diabetes mellitus, pancreatic exocrine insufficiency, central nervous system involvement, pulmonary dysfunction, male hypogonadism, benign intracranial hypertension, vascular calcifications, and nodular regenerating hyperplasia of the liver. We evaluated 100 individuals (58 males; 42 females) with nephropathic cystinosis age 18-45 years between January 1985 and May 2006. Ninety-two had a renal allograft, and 33 were deceased. Half or more had hypothyroidism, hypergonadotropic hypogonadism (males), pulmonary insufficiency, swallowing abnormalities, or myopathy. One-third had retinopathy or vascular calcifications, and 24% had diabetes. Homozygosity for the 57-kb <i>CTNS</i> deletion was associated with increased risk of death and morbidity. The 39 patients who received long-term ( $\geq 8$ years) oral cysteamine were taller and heavier, underwent a renal allograft later in life, had lower cholesterol levels, and suffered fewer complications and deaths. The frequencies of diabetes mellitus, myopathy, pulmonary dysfunction, hypothyroidism, and death increased with time off cysteamine treatment, and decreased with time on cysteamine. We conclude that untreated nephropathic cystinosis has extensive morbidity and mortality in adulthood. Long-term oral cysteamine therapy mitigates these effects.	<b>Contessa Ballroom B</b>
<b>12:15 pm – 1:15 pm</b>	<b>CRN Luncheon</b> <i>Complimentary</i>	<b>Las Ramblas/Cork Bar Atrium/River Walk Patio</b>

<b>1:30 pm – 3:30 pm</b>	<b>Poster Sessions</b> This session will showcase a mix of science, medicine, industry and advocacy group, and patient experiences to provide an interactive experience for both family and professional attendees. Researchers, clinicians, industry, advocacy representatives, students, patients, and caregivers will be invited to exhibit their latest research findings, treatment breakthroughs, advocacy group updates, and real patient and family experiences. This will be an interactive session where exhibitors will be available to discuss their work or experiences with those attending. We invite you to browse the posters and take this opportunity to ask the authors and presenters questions.	<b>Contessa Ballroom</b>
<b>4:00 pm – 5:30 pm</b>	<b>Medical Panel</b> <i>Christy Greeley, Host</i> <i>Panelists: Don Cairns, Ph.D., Craig Langman, M.D., Robert Kleta, M.D., Ph.D., Jerry Schneider, M.D., Bill Gahl, M.D., Ph.D., Ranjan Dohil, M.D., Jess Thoene, M.D., Ewa Elenberg, M.D., Corinne Antignac, M.D., Ph.D., Mark Benfield, M.D., Paul Goodyer, M.D., Doris Trauner, M.D.</i> Please join the entire group for the unique and informative opportunity to have your questions and concerns addressed by the leading physicians and researchers in cystinosis. All of the doctors who have presented at the Family Conference, all attending Medical Advisory Board and Scientific Review Board members, as well as other health care professionals involved in treating and researching cystinosis are scheduled to participate. Questions for the panel will be collected during the proceedings today.	<b>Contessa Ballroom</b>
<b>Saturday, July 21</b>	<b>Education and Family Support Issues</b>	
<b>Times to be determined</b>	<b>Southwest Foundation for Biomedical Research Blood sample collection</b>	<b>Mesquite</b>
<b>Times to be determined</b>	<b>Neurological Testing with Dr. Trauner’s team</b>	<b>Retama and Boardroom</b>
<b>8:00 am – 4:30 pm</b>	<b>Registration / Information Desk</b>	<b>Contessa Ballroom Foyer</b>
<b>8:00 am</b>	<b>Child Care Opens</b> <i>Open to the end of today’s agenda</i>	<b>Laurel</b>
<b>6:30 am – 8:00 am</b>	<b>Breakfast Buffet</b> <i>Complimentary</i>	<b>Las Ramblas</b>
<b>8:15 am – 8:30 am</b>	<b>Opening Comments</b> <i>Marybeth Kruppenacker, Host</i>	<b>Contessa Ballroom</b>
<b>8:40 am – 9:30 am</b>	<b>Brain Structure and Function in Cystinosis</b> <i>Doris Trauner, M.D.</i>	<b>Contessa Ballroom</b>

	Topics to be covered will include brain development, neurological, cognitive and academic functioning in children and adults with cystinosis.	
<b>9:40 am – 10:20 am</b>	<p><b>Education Issues</b>  <i>Susie McCalla</i>  This session will focus on issues relating to elementary and secondary education and children or young adults with cystinosis. It will begin with an overview of issues that are important to students and parents, such as medications in school, communication between schools and families, possible interventions under the special education or 504 umbrella, bullying and teasing, and others. It will end with a question and answer session in which parents' and students' questions can be discussed and answered.</p>	<b>Contessa Ballroom</b>
<b>10:30 am – 11:10 am</b>	<p><b>Bridging the Gaps: Transitioning from Pediatric to Adult Care</b>  <i>Rosimar Zavarse, LMSW</i>  Objective: to provide a brief review of the concepts of transitioning and the medical home, and to prepare parents and young adults with rare disease to consider and plan their own medical transitions.</p>	<b>Contessa Ballroom</b>
<b>11:20 am – 12:00 pm</b>	<p><b>Managing the Psychosocial Effects of Cystinosis: A Family Systems Approach</b>  <i>Nick Smith, M.A., LMHC</i>  Objective: To provide a brief review of research concerning the psychosocial effects of chronic illnesses on families and to provide practical team building information to better equip our cystinosis families.</p>	<b>Contessa Ballroom</b>
<b>12:00 pm – 1:15 pm</b>	<b>Lunch on your own</b>	
<b>1:15 pm – 2:45 pm</b>	<p><b>Workshop Session A</b>  These age and topic specific workshops will allow families to tailor their conference experience to meet their specific needs. Families will have the opportunity to discuss and share their own experiences regarding the latest research, management techniques, and therapies with leading experts as well as with other families with similar issues and concerns. Sessions will be repeated so that participants may attend more than one workshop.</p>	
	<p><b>A1 – Infancy/Preschool</b>  <i>Brittney LeBeau, Host</i>  Panelists: <i>Jerry Schneider, M.D., Ranjan Dohil, M.D., Craig Langman, M.D., LMHC, Whitney Glaize, MA, CCC-SLP</i></p>	<b>Contessa Ballroom A</b>
	<p><b>A2 – Childhood</b>  <i>Dave Greeley, Host</i>  Panelists: <i>Jess Thoene, M.D., Ewa Elenberg, M.D., Doris Trauner, M.D., Don Cairns, Ph.D., Paul Goodyer, M.D.</i></p>	<b>Contessa Ballroom B</b>
	<p><b>A3 – Adolescence</b>  <i>Pam Woodward, Host</i></p>	<b>Magnolia</b>

	<i>Panelists: Robert Kleta, M.D., Ph.D., David Glaize, Ed.D., Susie McCalla, Corinne Antignac, M.D., Ph.D, Mark Benfield, M.D., Rosimar Zavarase</i>	
	<b>A4 – Adult</b> <i>Paula Shal, Host</i> <i>Panelist: Bill Gahl, M.D.</i>	<b>Cedar</b>
	<b>A5 – Orthotics/Spinal Stabilizers Clinic</b> <i>Dr. David Ivey</i> Dr. Ivey believes that the strength of the musculoskeletal system starts with a strong foundation. The foot has approximately ¼ of the bones in the body and is the body’s primary source of support. Excessive pronation is one of the major causes of knee, hip and lower back pain. Laura McGinnis came into my office with leg pain and with the use of chiropractic adjustments and orthotics, her leg pain has been resolved. AMFit, Dr. Ivey’s choice for customized orthotics, has generously donated 25 sets for the benefit of the cystinosis community. Make your appointment at the registration table, it will be first come first serve basis.	<b>Palm</b>
	<b>A6 – Siblings Only</b> <i>Nick Smith, M.A., LMHC</i>	<b>Anaqua</b>
<b>3:00 pm – 4:30 pm</b>	<b>Workshop Session B</b>	
	<b>B1 – Infancy/Preschool</b> <i>Sandy Glaize, Host</i> <i>Panelists: Jerry Schneider, M.D., Ranjan Dohil, Craig Langman, M.D., Whitney Glaize, MA, CCC-SLP</i>	<b>Contessa Ballroom A</b>
	<b>B2 – Childhood</b> <i>Lynn Thomas, Host</i> <i>Panelists: Jess Thoene, M.D., Ewa Elenberg, M.D., Doris Trauner, M.D., Don Cairns, Ph.D., Paul Goodyear, M.D.</i>	<b>Contessa Ballroom B</b>
	<b>B3 – Adolescence</b> <i>Carol Hughes, Host</i> <i>Panelists: Robert Kleta, M.D., Ph.D., David Glaize, Ed.D., Susie McCalla, Corinne Antignac, M.D., Ph.D., Mark Benfield, M.D., Rosimar Zavarase</i>	<b>Magnolia</b>
	<b>B4 – Adult</b> <i>Mack Maxwell, Host</i> <i>Panelist: Bill Gahl, M.D., Ph.D.</i>	<b>Cedar</b>
	<b>B5 – Orthotics/Spinal Stabilizers Clinic</b> <i>Dr. David Ivey</i> Dr. Ivey believes that the strength of the musculoskeletal system starts with a strong foundation. The foot has approximately ¼ of the bones in the body and is the body’s primary source of support. Excessive pronation is one of the major causes of knee, hip and lower back pain.	<b>Palm</b>

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	<b>B6– Siblings Only (continued from Session A)</b> <i>Nick Smith, M.A., LMHC</i>	<b>Anaqua</b>
<b>4:45 pm – 5:15 pm</b>	<b>Conference Wrap Up</b> <i>Jose Morales, President</i>	<b>Contessa Ballroom</b>
<b>6:00 pm</b>	<b>CRN Dinner</b> <i>Complimentary</i> <i>Cash bar opens at 6:00, dinner begins at 6:30 pm</i>	<b>Las Ramblas/ Cypress/River Walk Patio</b>