



Family Conference Agenda

Wednesday, July 13		Room
4:00 pm - 8:00 pm	CRN Board of Directors Meeting <i>Closed Session</i>	Bayview
Thursday, July 14		Room
12:00 pm – 8:00 pm	Registration/Information Desk	Second Floor Landing
1:00 pm – 4:00 pm	CRN Scientific Review Board Meeting <i>Closed Session</i>	Bayview
1:45 pm – 4:00 pm	Transitioning Project Interviews/Focus Group – Parents Group <i>Maya Doyle, LCSW-R</i> The Cystinosis in Emerging Adulthood study is a focus group and interview based study being undertaken by Maya Doyle, LCSW, ABD. Focus groups for young adults (age 18-30) and their parents will be held during the CRN Family Conference on July 14 and 15. Follow-up interview will occur in the Fall 2011 and Winter 2012 at locations near participants' homes. Patients and families interested in participating in the study can find out more and register at https://www.surveymonkey.com/s/yacys or in person at the CRN Family Conference Registration Desk, or just prior to the Focus Groups in the Presidio Boardroom.	Presidio Boardroom
5:00 pm – 8:00 pm	CRN Welcome Reception <i>Complimentary, Cash Bar</i> <i>Hosts: The Jordan and Jondle Families</i> Please join us for drinks and appetizers and an opportunity to meet the families and doctors attending this year's conference.	Nikko Foyer
Friday, July 15		Room
7:00 am - 8:30 am	Breakfast <i>Complimentary</i>	Nikko Foyer
8:00 am - 5:30 pm	Registration/Information Desk	Nikko Foyer
8:00 am - 5:45 pm	Child Care Open	Monterey I
8:00 am - 9:00 pm	Teen Lounge Open Teens are welcome to spend time in the teen lounge anytime. Snacks and drinks will be available, as well as video games and television.	Monterey II
8:30 am - 8:45 am	Welcome & Opening Remarks <i>Christy Greeley – President and Executive Director</i>	Nikko Ballroom
8:45 am - 9:15 am	Cystinosis Research Network – Your Advocacy Group <i>Christy Greeley – President and Executive Director</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.	Nikko Ballroom
9:15 am - 10:00 am	Reaching Beyond the Clouds: From Undiagnosed to Climbing Mt. Everest <i>Cindy Abbott, M.S., Health Science Instructor, California State University, Fullerton</i> On August 1 st 2007, Cindy was diagnosed with Wegener's Granulomatosis and on May 23 rd 2010, she summited Mt. Everest. With a rare and potentially life-threatening disease, at the age 51, and functionally blind in one eye she became one of the 400 Americans who have climbed the world's tallest mountain. Her journey to Mt. Everest began three years ago, but her story, like so many other victims of rare unknown diseases, started more than a decade earlier. www.reachingbeyondthecLOUDS.com	Nikko Ballroom

10:00 am – 10:15 am	Break	Nikko Foyer
10:15 am – 12:30 pm	Transitioning Project Interviews/Focus Group – Young Women’s Group <i>Maya Doyle, LCSW, ABD</i> The Cystinosis in Emerging Adulthood study is a focus group and interview based study being undertaken by Maya Doyle, LCSW, ABD. Focus groups for young adults (age 18-30) and their parents will be held during the CRN Family Conference on July 14 and 15. Follow-up interview will occur in the Fall 2011 and Winter 2012 at locations near participants’ homes. Patients and families interested in participating in the study can find out more and register at https://www.surveymonkey.com/s/yacys or in person at the CRN Family Conference Registration Desk, or just prior to the Focus Groups in the Presidio Boardroom.	Presidio Boardroom
10:15 am – 11:00 am	All About Cystinosis <i>William A. Gahl, M.D., Ph.D.</i> Nephropathic cystinosis is an autosomal recessive lysosomal storage disorder with an estimated incidence of one in 100-200,000 live births. Patients appear normal at birth but generally develop failure to thrive and manifest renal tubular Fanconi syndrome, with its concomitant metabolic (normal anion gap) acidosis and volume depletion, electrolyte imbalances, growth retardation, and hypophosphatemic rickets by 6-12 months of age. Later, photophobia reflects progressive corneal crystal accumulation. In the natural history of untreated cystinosis, kidney failure occurs at approximately ten years of age, requiring dialysis or kidney transplantation. Later in the untreated patient, extra-renal complications occur with varying frequencies. These include hypothyroidism, distal vacuolar myopathy, swallowing abnormalities, retinal blindness, diabetes mellitus, pancreatic exocrine insufficiency, decreased pulmonary function, and neurological deterioration. Early and diligent treatment with oral cysteamine (CystagonR) allows for a normal growth rate, slows progression of the kidney deterioration, and prevents the late, non-renal complications of the disease. Topical therapy with cysteamine eyedrops can dissolve the corneal crystals of cystinosis.	Nikko Ballroom
11:00 am – 12:00 pm	Family Introductions <i>Cheri Friend, Vice President Family Support</i> Overview of CRN Family Support activities will be given.	Nikko Ballroom
12:00 pm – 12:15 pm	CRN Research Update <i>Elva Smith, Vice President Research</i> Overview of CRN Research funding activities will be given.	Nikko Ballroom
12:15 pm – 1:15 pm	Luncheon <i>Complimentary</i>	Nikko Foyer
1:00 pm – 3:15 pm	Transitioning Project Interviews/Focus Group – Young Men’s Group <i>Maya Doyle, LCSW, ABD</i> The Cystinosis in Emerging Adulthood study is a focus group and interview based study being undertaken by Maya Doyle, LCSW, ABD. Focus groups for young adults (age 18-30) and their parents will be held during the CRN Family Conference on July 14 and 15. Follow-up interview will occur in the Fall 2011 and Winter 2012 at locations near participants’ homes. Patients and families interested in participating in the study can find out more and register at https://www.surveymonkey.com/s/yacys or in person at the CRN Family Conference Registration Desk, or just prior to the Focus Groups in the Presidio Boardroom.	Presidio Boardroom
1:15 pm – 2:45 pm	Poster Session This session will showcase a mix of science, medicine, industry, 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.	Carmel I & II
2:45 pm – 3:00 pm	Break	Nikko Foyer
3:00 pm – 3:15 pm	CRN National Institutes of Health Fellowship <i>Galina Nesterova, M.D., CRN NIH Fellow</i> A review of the CRN Fellowship goals and objective as a trainee to become an expert in nephropathic cystinosis at the NIH in order to serve the cystinosis community will be given. These include clinical evaluation and treatment of cystinosis patients with a focus on early detection of complications in order to prevent long term disabilities; monitoring of appropriate therapy to preserve patients’ kidney function; education of the medical community; publications and research projects; and review of a clinical case with tissue sample cystine level results.	Nikko Ballroom

3:20 pm – 3:35 pm	<p>Cystaran™ (Cysteamine Hydrochloride Ophthalmic Solution): Project Update <i>Karen Kuphal, Ph.D., Sr. Manager, Project Management, Sigma Tau Pharmaceuticals, Inc.</i> The development of ophthalmic cysteamine progressed at the National Eye Institute (NEI) over 19 years (1986 –2005). The presentation will highlight the conservative approach used to combine all available exposure data that was submitted to FDA and the status of the project.</p>	Nikko Ballroom
3:40 pm – 3:55 pm	<p>Raptor Pharmaceuticals Delayed Release Cysteamine Trial Update <i>Ted Daley, President</i> Raptor Pharmaceuticals is developing RP103 for patients with cystinosis. RP103 is a delay released form of cysteamine bitartrate. Phase 3 clinical results are expected later this summer. This presentation is an opportunity to learn more about the clinical trial design, study locations, and future development plans for RP103.</p>	Nikko Ballroom
4:00 pm – 4:15 pm	<p>CRF Cystinosis Gene Therapy Consortium <i>Stephanie Cherqui, Ph.D.</i> The CRF Cystinosis Gene Therapy Consortium is a unified research program to develop stem cell and gene therapy treatments for cystinosis. This program represents a collaboration of leaders in the fields of stem cell and gene therapy, molecular biology and clinical and molecular pathophysiology of cystinosis with the support of the Cystinosis Research Foundation. All the members of the consortium will work towards the same goal: finding successful therapies for cystinosis.</p> <p>Gene therapy adds a functional copy of the faulty gene and delivers it to the appropriate cells of the body. In the case of cystinosis, most of the tissues are damaged because of the lack of the <i>CTNS</i> gene. In other human disorders, a person’s own stem cells have already been used safely, and the stem cells could target several tissues. For cystinosis patients, this strategy might create a reservoir of healthy stem cells in the bone marrow for the lifetime of the patient that might respond to the progressive tissue damage of cystinosis and travel to repair the different organs of the patient.</p> <p>This consortium was created based on the promising pre-clinical studies performed in the mouse model for cystinosis. Transplantation of healthy bone marrow stem cells led to a significant decrease of cystine in all the tissues tested as well as prevention or treatment of tissue injury. The critical next step is to develop the strategies necessary to successfully deliver the <i>CTNS</i> gene to patients’ bone marrow stem cells using gene therapy in the mouse model. Finally, the translation of these studies to humans will require extensive safety studies as well as the expertise of well-established researchers and physicians to lead to the first clinical trial of stem cell and gene therapy for cystinosis.</p>	Nikko Ballroom
4:15 pm – 5:45 pm	<p>Medical Panel Moderator: <i>William A. Gahl, M.D., Ph.D.</i> Panelists: <i>Rick Kaskel, M.D., Ph.D., Craig Langman, M.D., Jerry Schneider, M.D., Jess Thoene, M.D., Corinne Antignac, M.D., Ph.D., Ranjan Dohil, M.D., Ewa Elenberg, M.D., Paul Goodyer, M.D., Galina Nesterova, M.D., Stephanie Cherqui, Ph.D., Bruce Barshop, M.D., Ph.D., Larry Greenbaum, M.D., Susan Thomas M.D., Elena Levtchenko, M.D., Ph.D., Francesco Emma, M.D., Richard Simon, M.D., Doris Trauner, M.D., Don Cairns, Ph.D., Roz Mannon, M.D., Roz Anderson, Ph.D., Minnie Sarwal, M.D., Leticia Belmont, M.D., Catherine Tuleu, Ph.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</p>	Nikko Ballroom
5:45 pm – 6:15 pm	<p>Session Wrap-Up and Group Photo <i>Christy Greeley, President and Executive Director</i> Group photos will be taken of all those with cystinosis, as well as all families and professionals in attendance.</p>	Nikko Ballroom
Saturday, July 16		Room
7:00 am - 8:30 am	<p>Breakfast <i>Complimentary</i></p>	Nikko Foyer
8:00 am - 5:30 pm	Registration/Information Desk	Nikko Foyer
8:00 am - 5:30 pm	Child Care Open	Monterey I
8:00 am - 9:00 pm	<p>Teen Lounge Open Teens are welcome to spend time in the teen lounge anytime. Snacks and drinks will be available, as well as video games and television.</p>	Monterey II
8:30 am - 8:45 am	<p>Opening Comments <i>Paula Shal – VP Education and Awareness</i> Overview of the day’s agenda and CRN Education and Awareness overview will be given.</p>	Nikko Ballroom

8:45 am - 9:30 am	CRN Adult Care Excellence Initiative <i>Maya Doyle, LCSW, ARB and Colleen Hammond, CRN Board Member</i>	Nikko Ballroom
9:30 am – 10:15 am	Patient Panel Presentation <i>Moderators: Pam Woodward and Marybeth Krummenacker, CRN Board Members</i> <i>Panelists: Jessica Jondle, Laura Krummenacker, Laura McGinnis, Tahnine Woodward, Andrew Hoffman, Steve Schleuder, Christian Morales</i> Patient panel presentation during which panel members will answer prepared questions and address topics related to the use of coping mechanisms through the ups and downs that cystinosis brings related to not only developmental and transitional issues of daily life but also medical issues. Teens and adults living with cystinosis will be featured and will share how they have recognized and lived to their full potential given the challenges they have faced. Audience participation will be encouraged.	Nikko Ballroom
10:15 am – 10:30 am	Break	Nikko Foyer
10:30 am – 12:45 pm	Transitioning Project Interviews/Focus Group – Mixed Young Adult Group <i>Maya Doyle, LCSW-R</i> The Cystinosis in Emerging Adulthood study is a focus group and interview based study being undertaken by Maya Doyle, LCSW, ABD. Focus groups for young adults (age 18-30) and their parents will be held during the CRN Family Conference on July 14 and 15. Follow-up interview will occur in the Fall 2011 and Winter 2012 at locations near participants' homes. Patients and families interested in participating in the study can find out more and register at https://www.surveymonkey.com/s/yacys or in person at the CRN Family Conference Registration Desk, or just prior to the Focus Groups in the Presidio Boardroom.	Presidio Boardroom
10:30 am – 10:40 am	Advocacy Groups: Collaboration is the Key <i>Host: Marybeth Krummenacker, CRN Board Member and NORD Board Member</i> During this morning's session, organizations that advocate for the entire rare disease community, pharmaceutical companies with a broad interest in rare diseases, as well as cystinosis advocacy groups from around the world will give updates on their activities. The importance of collaboration among these groups is key to our community's success.	Nikko Ballroom
10:45 am – 11:00 am	Advocacy Through Collaboration <i>Mary E. Cobb, Senior Vice President, National Organization for Rare Disorders</i> In this presentation, Mary will share examples of how The National Organization for Rare Disorders (NORD) collaborates with different stakeholders on initiatives that support their mission of being the voice of the rare disease community. As an umbrella organization that is dedicated to helping people with rare "orphan" diseases and assisting the patient organizations that serve them, NORD believes in the <i>power of collaboration and the value of diverse perspectives – all in the interest of supporting the needs of patients.</i> This is reflected through their advocacy, education and research activities, as well as through a partnership with EURORDIS which helps to provide global connections on critical rare disease issues.	Nikko Ballroom
11:05 am – 11:15 am	AIRG-France: Association for Information and Research on Genetic Renal Diseases <i>Beatrice Coupey</i>	Nikko Ballroom
11:20 am – 11:35 am	Cystinosis Foundation <i>Valerie Hotz, Executive Director</i>	Nikko Ballroom
11:40 am – 11:50 am	Cystinosis Foundation Ireland <i>Mick Swift, Chairman</i>	Nikko Ballroom
11:55 am – 12:05 pm	Cystinosis Mexico <i>Victor Gomez, President</i>	Nikko Ballroom
12:10 pm – 12:25 pm	Cystinosis Research Foundation <i>Tricia Sturgis, Board Member</i> The Cystinosis Research Foundation is the largest fund provider of cystinosis research in the world. Our mission is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. We have issued over \$12.9 million in research grants since 2003. The CRF funded every study that led to the discovery of delayed-release cysteamine, we are funding cutting-edge stem and gene therapy research, we established the CRF Cystinosis Gene Therapy Consortium led by Dr. Stephanie Cherqui and CRF established the first post-doctoral cystinosis fellowship program. In addition, the CRF is dedicated to educating the medical community about cystinosis and connecting with cystinosis families in order to unify the cystinosis community. The CRF hosts the annual <i>Day of Hope</i> , a family conference, in an effort to build community fellowship and share research progress. In 2010, the CRF, with support from thirteen other family foundations, established the first international patient registry, the Cure Cystinosis International Registry (CCIR) to assist the scientific community, accelerate cystinosis research and to understand more about cystinosis and its complications.	Nikko Ballroom
12:30 pm – 1:30 pm	Luncheon <i>Complimentary</i>	Nikko Foyer

1:30 pm – 3:00 pm	<p>Workshop Session A These 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>A1 – Childhood Issues <i>Hosts: Jeff Larimore, CRN Treasurer, Frankie McGinnis, CRN Board Member</i> <i>Panelists: Jess Thoene, M.D., Ranjan Dohil, M.D., Larry Greenbaum, M.D., Francesco Emma, M.D., Don Cairns, Ph. D.</i></p> <p>A2 – Pediatric to Adult Care Transition Issues <i>Hosts: Colleen Hammond and Jessica Jondle, CRN Board Members</i> <i>Panelists: Rick Kaskel, M.D., Ph.D., Craig Langman, M.D.</i></p> <p>A3 – Neurology and Educational Issues <i>Hosts: Jen Wyman, CRN VP Development, Lorna Smith, CRN Board Member</i> <i>Panelists: Doris Trauner, M.D. , Lynn Thomas, Parent Advocate</i></p> <p>A4 – Adult Issues <i>Hosts: Tahnje Woodward, CRN Board Member, Karen Gledhill, CRN Secretary</i> <i>Panelists: William A. Gahl, M.D., Ph.D., Ewa Elenberg, M.D., Susan Thomas, M.D., Richard Simon, M.D.</i> <i>First half hour of the session will be a private session for adults with cystinosis only.</i></p> <p>A5 – Transplantation <i>Hosts: Pam Woodward and Jose Morales, CRN Board Members</i> <i>Panelists: Paul Goodyer, M.D., Roz Mannon, M.D., Minnie Sarwal, M.D.</i></p>	<p>Nikko Ballroom III</p> <p>Carmel I</p> <p>Carmel II</p> <p>Mendocino I</p> <p>Mendocino II</p>
3:00 pm – 3:15 pm	Break	Nikko Foyer
3:15 pm – 4:45 pm	<p>Workshop Session B These 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>B1 – Childhood Issues <i>Hosts: Jen Sexstone and Lorna Smith, CRN Board Members</i> <i>Panelists: Jess Thoene, M.D., Ranjan Dohil, M.D., Larry Greenbaum, M.D., Francesco Emma, M.D., Don Cairns, Ph. D.</i></p> <p>B2 – Pediatric to Adult Care Transition Issues <i>Hosts: Colleen Hammond and Jessica Jondle, CRN Board Members</i> <i>Panelists: Rick Kaskel, M.D., Ph.D., Craig Langman, M.D.</i></p> <p>B3 – Neurology and Educational Issues <i>Hosts: Elva Smith, CRN VP Research</i> <i>Panelists: Doris Trauner, M.D. , Lynn Thomas, Parent Advocate</i></p> <p>B4 – Adult Issues <i>Hosts: Paula Shal and Mack Maxwell, CRN Board Members</i> <i>Panelists: William A. Gahl, M.D., Ph.D., Ewa Elenberg, M.D., Susan Thomas, M.D., Richard Simon, M.D.</i> <i>First half hour of the session will be a private session for adults with cystinosis only.</i></p> <p>B5 – Transplantation <i>Hosts: Pam Woodward and Jose Morales, CRN Board Members</i> <i>Panelists: Paul Goodyer, M.D., Roz Mannon, M.D., Minnie Sarwal, M.D.</i></p>	<p>Nikko Ballroom III</p> <p>Carmel I</p> <p>Carmel II</p> <p>Mendocino I</p> <p>Mendocino II</p>
4:45 pm – 5:30 pm	<p>Conference Wrap Up <i>Christy Greeley, President and Executive Director</i> Summary of conference proceedings, announcement of 2013 conference site and announcement of "Above and Beyond" Achievement Award</p>	Nikko Ballroom
6:30 pm – 10:00 pm	<p>Farewell Dinner Dance <i>Complimentary, Cash Bar</i> Please join all conference attendees for dinner and dancing to celebrate the end of our time together in San Francisco.</p>	Nikko Ballroom