



CONFERENCE AGENDA

WEDNESDAY, JULY 12TH

12:00 pm - 8:00 pm

View on 2

CRN Board of Directors Meeting

Closed session – CRN board members

6:00 pm - 9:00 pm

Acorn A & B

Speak Up Speak Out Welcome Reception

Closed session – SUSO participants

THURSDAY, JULY 13TH

8:00 am - 5:00 pm

Parthenon

Ballroom Foyer

Registration/Information Desk

Please stop by the Registration Desk to check in and pick up your welcome information including conference booklet, t-shirts, and name badges.

QUALIFY-US Study

Mona Suck, MSc

8:00 am - 4:00 pm

Acorn A

Kirkland Hall

Speak Up/Speak Out:

Cystinosis & Me

Closed session – SUSO participants

Horizon Therapeutics is excited to announce the return of Speak Up, Speak Out:

Cystinosis & Me the groundbreaking spoken-word self-advocacy workshop for young people living with cystinosis, produced in partnership with Believe Limited and the Cystinosis Research Network. The two-day

workshop will focus on empowering cystinosis community members to use their own words, performed publicly, to tell the world about their journey, showcase the therapeutic and healing elements of the creative arts, build community, and teach the importance of self-advocacy, ultimately culminating in a final performance during CRN's Family Conference. This community experience will feature: Written and spoken word technique education, theatrical and performance training, relaxation, meditation and breathing exercises, a final performance during CRN's Family Conference, and so much more!

8:00 am - 9:00 am

Parthenon
Ballroom AB

**Symposium Welcome
and Breakfast**

Complimentary

9:00 am - 4:00 pm

Crescent Room

Scientific Symposium

*Host: Christy Greeley,
Executive Director, VP Research*

Chair: Elena Levtchenko, MD, PhD

9:00 am - 10:45 am

Crescent Room

**Session 1: Pathogenesis
& New Interventions**

9:00 am - 9:30 am

*New mechanisms of kidney damage in cystinosis – Minne Sarwal, MD, PhD
Understanding the unique susceptibility of the human kidney to pH dysfunction and injury in cystinosis is paramount to develop new therapies to preserve renal function. Renal proximal tubular epithelial cells (RPTECs) and fibroblasts, isolated from patients with cystinosis were transcriptionally profiled.*

Lysosomal fractionation, immunoblotting, confocal microscopy, intracellular pH, TEM, mitochondrial stress test, and membrane integrity assays were performed for validation. CRISPR, CTNS -/- RPTECs were generated. A new compound, Astaxanthin (ATX), was evaluated for rescue of the CTNS -/- phenotype. Alterations in cell stress, pH, autophagic turnover, and mitochondrial energetics, highlighted key changes in the vacuolar (V)-ATPases in patient derived and CTNS-/- RPTECs. ATP6V0A1 was significantly downregulated in cystinosis and highly co-regulated with loss of CTNS. Overexpression of ATP6V0A1 rescued cell stress and mitochondrial function. Treatment of CTNS -/- RPTECs with ATX, induced ATP6V0A1 expression and the resulting rescue of the RPTE cell and mitochondrial injury.

In conclusion, our exploratory transcriptional and in vitro cellular and functional studies confirm that loss of cystinosin in RPTEC, results in a reduction in ATP6V0A1 expression, with changes in intracellular pH, mitochondrial integrity, mitochondrial function, and autophagosome-lysosome clearance. ATX can rescue the cystinotic RPTEC injury, at least partially mediated by upregulating ATP6V0A1 expression. The availability of ATX as a well-tolerated oral supplement, offers its further clinical evaluation as a potential novel therapeutic to limit renal tubular injury in cystinosis, independent of cysteine depletion alone.

9:30 am - 10:00 am

*New pharmacological treatments for cystinosis beyond cystine accumulation –
Francesco Emma, MD
For more than 3 decades, patients with*

nephropathic cystinosis have been treated in western countries with cysteamine, which has dramatically changed the prognosis of the disease, but cannot prevent all complications. Some symptoms however, such as the renal Fanconi syndrome, are poorly or non-responsive to cysteamine, for reasons that are still poorly understood. In vitro and in vivo studies performed in the past decades have shown that in addition to lysosomal cystine accumulation, the disease is also characterized by altered mitochondrial bioenergetics and dynamics, impaired autophagy and mTOR signaling, dysfunction of endocytosis and intracellular trafficking processes, activation of the inflammasome, enhanced oxidative stress, and increased propensity to undergo apoptosis. Which of these anomalies represent primary events and which are secondary to the lysosomal dysfunction is still debated. These results however, have provided the rationale for developing new therapeutic approaches to be used in addition to cysteamine therapy. Several molecules have shown promising results using in vitro models of cystinosis and some have now been successfully tested in vivo using mouse and rat models of the disease. In addition, new dietary interventions have shown significant protection of the renal disease in animal models of cystinosis. In this presentation I will review these different approaches.

10:00 am - 10:40 am

*Potential of RNA therapy in genetic diseases -
Tjessa Bondue, MS*

Messenger RNA (mRNA) therapies are emerging in different disease areas, but have not yet reached the kidney field. Our aim is to study the feasibility to correct the genetic defect in nephropathic cystinosis using

*synthetic mRNA. We have used the synthetic mRNA to first introduce the CTNS protein into the lysosomes of cystinotic proximal tubular epithelial cells and podocytes, followed by functional assessment of the effect. The approach was also tested in an animal model of cystinosis, the *ctns*^{-/-} zebrafish. In the zebrafish, we can analyze the effect on the kidney phenotype, studying both the proximal tubular reabsorption function and the effect of the mRNA on the proteinuria. By establishing the proof-of-principle of mRNA-based protein replacement for cystinosis, we lay the basis for the future application of these types of therapies for a variety of genetic (kidney) diseases.*

The potential of Moderna mRNA technology to treat cystinosis - Paul Goodyer, MD
While cysteamine therapy has changed the natural history of cystinosis, it doesn't seem to tackle all aspects of the condition. We and others have tried to develop therapies that replace all functions of the CTNS. In 2019, we launched a clinical trial of ELX-02 to bypass CTNS nonsense mutations, but this was halted by the COVID pandemic. While disappointing, it is said that every cloud has a silver lining. During the pandemic, we were able to establish a collaboration with Moderna in which we will try to apply their mRNA/lipid nanoparticle technology to the therapy of cystinosis."

10:40 am - 11:00 am

Panel Discussion - Minnie Sarwal, MD, PhD, Francesco Emma, MD, Tjessa Bondue, MS, Paul Goodyer, MD

11:00 am - 11:15 am Coffee Break

Parthenon
Ballroom Foyer

11:15 am - 1:00 pm Session 2: Crescent Room Early Diagnosis & Biomarkers

11:15 am - 11:45 am

Genetic neonatal screening - Katharina Hohenfellner, MD

The presentation will give a background on the genetics of cystinosis, newborn screening in the USA, the pilot project for newborn screening in Germany and the future of newborn screening with new technologies.

11:45 am - 12:15 pm

Monitoring strategies and impact of pre-symptomatic treatment - Elena Levtchenko, MD, PhD

*The availability of cysteamine treatment and the success of kidney transplantation have dramatically improved long-term prognosis of patients with cystinosis, many of whom are currently reaching the age above 50. Studying large patients' cohorts in the US and in Europe has demonstrated that patients' kidney survival mostly depends on the age at start of cysteamine and on patients' compliance with cysteamine therapy (Nesterova, Gahl *Pediatr Nephrol* 2015, Emma et al. *Kidney Int* 2021). The majority of patients (60-70%) in whom cysteamine was initiated below the age of 1 year and who maintained an average white blood cell (WBC) cystine < 1 nmol ½ cystine/mg protein reached adult age without the need of kidney transplantation. A recent study in siblings with cystinosis demonstrated that starting cysteamine during the 1st month of life before*

the symptoms of renal Fanconi syndrome develop results in 30 ml/min/1.73 m² gain of kidney function (Veys et al. J Inher Metab Dis 2023). Measuring WBC cystine is technically challenging and is not available in many countries. Moreover, false-low WBC cystine levels can be observed in patients with milder forms of cystinosis (Bondue et al. Int J Mol Sci 2023). Therefore the cystinosis research field is exploring the potential of alternative biomarkers to monitor cystinosis that will be discussed in the current presentation (Emma et al. Cells 2022).

12:15 pm - 12:45 pm

Development of neuromarkers in cystinosis - John Foxe, PhD

12:45 pm - 1:05 pm

Panel Discussion - Katharina Hohenfellner, MD, Elena Levtchenko, MD, PhD, John Foxe, PhD

1:05 pm - 2:00 pm

Parthenon
Ballroom AB

Symposium Lunch

Complimentary

1:05 pm

Parthenon
Ballroom Foyer

Poster Session Kickoff

This session will showcase a mix of science, medicine, industry, and advocacy group topics to provide an interactive experience for both family and professional attendees. Researchers, clinicians, industry, and advocacy representatives will be invited to exhibit their latest research findings, treatment breakthroughs, and advocacy group updates. Posters will remain on display for the entirety of the conference.

2:00 pm - 4:00 pm
Crescent Room

Session 3: Treatment, Advocacy & Quality of Life

2:00 pm - 2:20 pm

*New strategies in kidney transplantation -
Roslyn Mannon, MD*

The number of kidneys transplanted has reached an all time high, with over 25,000 kidney transplants in 2022, and on track to do even more in 2023. In spite of this, the number of people on the waitlist continues to rise, with 89,000 registered. This mismatch in available organs has propelled the field to consider alternatives but also to take deeper study into the access issues for patients to kidney transplants and to examine those barriers. Moreover, there has been little developed in terms of new drugs after transplant, and late kidney graft failure continues to be a challenging problem.

In this talk, we will discuss a number of issues in the transplant field. To solve the organ shortage, we will talk a little about preservation strategies, how organs are dispersed (“allocated”), and the promise and challenge of xenotransplantation. We will talk about potential new drugs in the pipeline and some of the cell therapy trials. Finally, we will discuss causes of late graft failure, and how this is being addressed in terms of noninvasive biomarkers, focus on cardiovascular and diabetes management, and immunologic management.

This is an exciting time to be in the transplant field, and there is optimism that we will / can solve many of these issues in this decade.

2:20 pm - 2:40 pm

*Management of fertility issues in cystinosis –
Aude Servais, MD, PhD*

There is an increasing number of adult

patients with cystinosis which raises the question of fertility. In women, there are no specific concerns: fertility and pregnancy course depend mainly on renal function. Results from an European study of pregnancies in cystinosis patients will be shown. We will discuss the main situations and expected outcomes of pregnancies. In males, there is a risk of delayed puberty and infertility. We will present recent data regarding the mechanisms of this complication, management and new available tools for fertility preservation.

2:40 pm - 3:00 pm

*Harmonization of cystinosis treatment around the globe and monitoring quality of life - Katharina Hohenfellner, MD, Mona Suck, MSc
QUALIFY-US – Development of a patient-reported outcome to measure the health-related quality of life of children and adolescents with cystinosis. Studies examining health-related quality of life (HrQoL) in patients with cystinosis suggest a limited HrQoL for patients and their caregivers. Primarily generic instruments are used; no disease-specific patient-reported outcome measures (PROMs) exist so far. The QUALIFY-US study aims to develop and validate a disease-specific PROM to measure HrQoL in children, adolescents, and young adults with cystinosis in Germany, France, Spain, and the United States of America. Questionnaire development includes the following steps: (1) developing a semi-structured interview guide based on a systematic literature review and feedback from physicians, patients, and representatives from patient organizations, (2) conducting focus (group) interviews in all participating countries to identify relevant aspects of the disease-specific HrQoL,*

(3) data analysis and item formulation for the preliminary questionnaire, including a cognitive debriefing, (4) a field and re-test for psychometric testing. The final result of the study will be a cross-cultural, psychometrically validated, practically feasible, and conceptually suitable instrument to assess HrQoL in children, adolescents, and young adults with cystinosis available in German, English, Spanish, and French for further (inter-)national studies.

SELECT - Cystinosis Guideline - The goal is to develop an evidence-based guideline for cystinosis. Since it is funded by the German Government, we have to follow certain requirements. The Cochraine Institute in Krems is involved regarding the evidence. A total of 85 medical colleagues and patient support groups worldwide are involved. The project started on 01.10.2022 and has a time frame of 24 months. Starting in fall of 2023, proposals will begin to be formulated and will be finalized at the guideline meeting in April 2024. Subsequently, a corresponding patient guide and a guideline for medical staff (in German and English) will be prepared and published.

3:00 pm - 3:20 pm

*Cystinosis in Africa: challenges and opportunities - Neveen Soliman, MD, PhD
Africa, the second most populous continent with a population of 1.4 billion, faces serious challenges when it comes to rare diseases. In the African context, cystinosis is probably underdiagnosed where physicians and patients are still facing numerous diagnostic, therapeutic, economic and ethical challenges. Here we discuss the current situation of cystinosis in Africa and why it should be given commensurate attention to overcome the existing hurdles. This could be achieved*

through an integrated approach that includes: 1) Promoting cystinosis awareness and genomic literacy among healthcare providers and the public to address the unmet needs of affected patients; 2) Ending the diagnostic odyssey is the very first step towards identifying cystinosis patients in different African nations through networking and collaboration to ensure access to diagnosis, genetic testing and counseling; 3) Working on providing equitable access to treatment mainly cysteamine therapy and kidney transplantation; and 5) Developing multidisciplinary and multinational research programs to improve our understanding of cystinosis not only within the continent but globally. This integrated approach is expected not only to bridge the healthcare gap between Africa and Europe/North America; but also to reduce the existing disparities between various African nations with the ultimate goal of improving global cystinosis outcomes.

3:20 pm - 3:40 pm

*A non-orphan use for an orphan drug:
Cysteamine for SARS-CoV-2 - Jess Thoene,
MD*

Global COVID 19 pandemic is caused by infection with severe acute respiratory syndrome coronavirus 2(SARS- CoV-2). Continuous emergence of new variants and their rapid spread are jeopardizing vaccine countermeasures to a significant extent. While currently available vaccines are effective at preventing illness associated with SARS- CoV-2 infection, these have been shown to be less effective at preventing breakthrough infection and transmission from a vaccinated individual to others. Here we demonstrate broad antiviral activity of cysteamineHCl in vitro against major emergent infectious variants of SARS-

CoV-2 in a highly permissible Vero cell line. CysteamineHCl inhibited infection of wild type, alpha, beta, gamma, delta, lambda, and omicron variants effectively. Cysteamine is a very well-tolerated USFDA-approved drug used chronically as a topical ophthalmic solution to treat ocular cystinosis in patients who receive it hourly or QID lifelong at concentrations 6 times higher than that required to inhibit SARSCoV-2 in tissue culture. Application of cysteamine as a topical nasal treatment can potentially 1) mitigate existing infection 2)prevent infection in exposed individuals,and 3) limit the contagion in vulnerable populations. Thoene J, Gavin RF, Towne A, Wattay L, Ferrari MG, Navarrete J, Pal R. In vitro activity of cysteamine against SARS-CoV-2 variants. Mol Genet Metab. 2022 Sep-Oct;137(1-2):192-200. doi: 10.1016/j.ymgme.2022.08.009. Epub 2022 Sep 5. PMID: 36115282; PMCID: PMC9444157

3:40 pm - 4:00 pm

Panel discussion - Roslyn Mannon, MD, Aude Servais, MD, PhD, Katharina Hohenfellner, MD, Mona Suck, MSc, Neveen Soliman, MD, PhD, Jess Thoene, MD

4:00 pm - 5:00 pm

Franklin Hall

First Time Attendee Orientation and Networking Session

Hosts: Jonathan Dicks, President, VP Development and Jen Wyman, VP Family Support

We look forward to welcoming and connecting our new families as well as providing details on what to expect during the conference. Please bring your entire family.

5:00 pm - 7:00 pm

Parthenon Ballroom

CRN Welcome Reception

Hosts: CRN Board of Directors

Complimentary dinner and open bar

7:00 pm - 10:00 pm

Parthenon Ballroom

Family Introductions/Networking

Hosts: Jonathan Dicks, President, VP Development, Tim Wyman, Treasurer, Jen Wyman, VP Family Support, Kristina Sevel, Board of Directors

A great opportunity to put faces to names of each family attending the conference as all families will have the opportunity to introduce themselves. Bring your entire family!

FRIDAY, JULY 14TH

7:00 am - 8:30 am

Parthenon Ballroom

Breakfast

Complimentary

8:00 am - 5:00 pm

Parthenon
Ballroom Foyer

Registration/Information Desk

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Parthenon
Ballroom Foyer

Poster Session

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QUALIFY-US Study

Mona Suck, MSc

8:00 am - 5:30 pm

Acorn A & B

Childcare and Teen Activities

Sponsored by Leadiant Biosciences, Inc. The CRN is proud to offer CHILDCARE for all the children and siblings from ages 0-12 attending the conference! We will have a full staff of nannies to watch the children. Childcare rooms will be open both Friday and Saturday excluding lunch time. There will be age-appropriate activities, games, and tons of crafts to make and take! All ages are encouraged to participate in the entertainment and crafts in the rooms! Parents MUST sign their child into daycare each morning and SIGN OUT every time they come to get their child(ren) (ages 0-12). Parents need to take their children out of childcare for lunch. Dosing of medications can only be administered by parents, not nannies or volunteers. Snacks and plenty of water will be provided. Guaranteed to allow the kids to not only bond, but to have the time of their lives!

Acorn C

Ages 13 and up will be provided a safe and fun TEEN ROOM, where they can come and go as they please. There will be room to hang out with other teens, along with games to play including an Xbox Series S, Nintendo Switch, and plenty of board and card games! Come check it out! Teens are also welcome to participate in any entertainment or craft activity that is taking place in the childcare room.

8:15 am - 8:45 am

Parthenon Ballroom

Welcome and Opening Remarks

Jonathan Dicks, President, VP Development

8:45 am - 9:15 am

Parthenon Ballroom

**Cystinosis Research Network:
Your Advocacy Group**

*Christy Greeley, Executive Director,
VP Research*

9:00 am - 11:30 am

View on 2
Kirkland Hall

**Speak Up Speak Out:
Cystinosis & Me**

Closed session – SUSO participants

9:15 am - 9:45 am

Parthenon Ballroom

Cystinosis 101

Bill Gahl, MD, PhD

Cystinosis is a genetic disorder due to failure to remove the amino acid cystine from a compartment within cells called the lysosome. The consequent accumulation of cystine causes crystals to form and damages different organs of the body at different rates. For example, the kidney fails to reabsorb water and small molecules in the first year of life; crystals form in the cornea by about a year of age; muscle and other organ involvement occurs in the second to fourth decades. Without treatment, kidney failure occurs around age 10 years and requires dialysis or transplantation. Symptomatic treatment includes replacement of kidney losses of small molecules, thyroid replacement, and sometimes growth hormone and other therapies. Directed treatment to remove cystine from lysosomes and cells involves oral cysteamine; for the corneal crystals, topical cysteamine eye drops are effective. Early diagnosis is critical.

9:45 am - 10:15 am

Parthenon Ballroom

**Preparing for Your 1st, or Next,
Kidney Transplant**

Paul Grimm, MD

Unfortunately, even when children are

diagnosed with cystinosis at a young age and they can tolerate the cystine depleting therapy, there may be substantial damage already done to the kidneys. This damage may occur without the usual tests of kidney function being abnormal. Creatinine might be “normal” even after significant amounts of kidney damage has occurred. We are usually born with a lot of extra kidney function, so we don’t start feeling ill from kidney failure until we are down at 10 or 15% function. Therefore, we have to rely on the kidney numbers that come with routine blood testing, but they are not so easily interpreted. So, as a parent or a patient, how worried should you be about your kidney numbers? What are the kidney numbers that are good, or bad, or worse? Are there any special considerations for a cystinosis patient compared to any other patient with chronic kidney disease? We will discuss the kidney numbers that include CKD Stages (1, 2, 3, 4 and 5/ESRD) serum creatinine, BUN and a relative newcomer called Cystatin C. These are used singly and in combination to get a better understanding of the kidney function. We may also discuss measuring protein loss in the urine to help determine how far advanced the kidney function is. We will talk about plotting these kidney numbers on a graph and using it to help predict when the kidney function might be falling to the level where some kind of kidney replacement therapy/renal replacement therapy will be necessary. At the end of the session, we will also discuss ways to slow the progress of kidney failure; including nutritional choices, lifestyle choices and medication.

10:15 am - 10:30 am Break

Parthenon
Ballroom Foyer

10:30 am - 11:00 am Making Sense of Clinical Trials

Parthenon Ballroom

J.J. Zaritsky, MD

We will review how clinical trials are designed and performed from a patient and family perspective. Specifically, we will go through the steps that are needed to bring a new therapeutic to market. We will also spend some time on the current state of therapies that target genes. Finally, we will review the process of therapeutic development for rare diseases.

11:00 am - 11:45 am Stem cell gene therapy for cystinosis: updated results of the clinical trial

Parthenon Ballroom

Stephanie Cherqui, PhD and Jordan Janz

We will report results from the phase 1/2 clinical trial (NCT03897361) evaluating safety and efficacy of CTNS-RD-04 in adult patients with cystinosis. CTNS-RD-04 consists of autologous CD34+ hematopoietic stem and progenitor cells (HSPCs) transduced with a lentiviral vector (LV) carrying the CTNS cDNA encoding for cystinosin. The trial is fully enrolled, and six participants (ages 20 to 46 years) have been treated with CTNS-RD-04 with follow-up ranging from 1 to 36 months. Jordan, patient 1 of the clinical trial, will share his experience and answer questions.

11:45 am - 12:15 pm Speak Up Speak Out: Cystinosis & Me

Parthenon Ballroom

Group Performance

12:15 pm - 12:30 pm Group Photograph

Parthenon Ballroom

All conference attendees should gather for a group photograph and a photograph of all individuals in attendance living with cystinosis. Please pick up your children from childcare and bring them to the ballroom for this wonderful remembrance of the conference.

12:30 pm - 1:30 pm Lunch

Parthenon Ballroom

Complimentary

1:45 pm - 4:00 pm

Meet in Acorn C

Scavenger Hunt Through Centennial Park

Teens/Young Adults only, join us for a scavenger hunt through Centennial Park! This 2 mile scavenger hunt gives you the opportunity to see sites of Nashville, along with meeting other teens/young adults at the conference. Please meet in the Teen Room (Acorn C) at 1:45 pm to start the hunt and come prepared to walk! Sign up available pre-conference and at the registrations desk on Thursday. Nashville Scavenger Hunt: Centennial Park Pursuit Palooza | Let's Roam (letsroam.com)

1:30 pm - 4:00 pm

Panel Sessions 1 & 2

Panel sessions are targeted towards different phases in the cystinosis journey, divided into tracks by age or topic. They will be smaller group sessions with a panel of experts presenting information, guiding discussion and fielding questions. Attendees should feel free to attend sessions of most interest to them. Some sessions are closed as noted below.

1:30 pm - 2:30 pm Panel Session 1

Ballroom A-C

Newly Diagnosed Families

Host: Kristina Sevel, Board of Directors

Panelists: Rachel Bishop, MD, J.J. Zaritsky, MD, Neveen Soliman, MD, PhD, Albert Freedman, PhD, Christina Nguyen, MD, Ranjan Dohil, MD, Galina Nesterova, MD, Francesco Emma, MD

Ballroom DE

Middle Childhood and Teen Years

Host: Chelsea Meschke and Herberth Sigler, Board of Directors

Panelists: Rezan Topaloglu, MD, FESPN, Doris Trauner, MD, Paul Goodyer, MD, Minnie Sarwal, MD, PhD, Katharina Hohenfellner, MD, Maya Doyle, MSW, PhD, Larry Greenbaum, MD, PhD, Donald Cairns, PhD

Crescent Room

Adults

Host: Karen Gledhill, Secretary

Panelists: Bill Gahl, MD, PhD, Elena Levtschenko, MD, PhD, Roz Mannon, MD, Paul Grimm, MD, Jess Thoene, MD, Ewa Elenberg, MD, Med, Aude Servais, MD, PhD

2:30 pm - 2:45 pm Break

Parthenon Ballroom
Foyer

2:45 pm - 3:45 pm Panel Session 2

Ballroom A-C

Newly Diagnosed and Middle Childhood: Physician Q & A

Host: Tim Wyman, Treasurer

Panelists: Rezan Topaloglu, MD, FESPN, J.J. Zaritsky, MD, Neveen Soliman, MD, PhD, Doris Trauner, MD, Katharina Hohenfellner,

MD, Larry Greenbaum, MD, PhD, Ranjan Dohil, MD, Galina Nesterova, MD

Ballroom DE

Transplant and Dialysis

Host: *Marybeth Krummenacker, VP Education & Awareness*

Panelists: *Minnie Sarwal, MD, PhD, Paul Goodyer, MD, Roz Mannon, MD, Christina Nguyen, MD, Francesco Emma, MD*

Crescent Room

Adults: Living with Cystinosis as an Adult

(closed session for adults 18 + and their partners)

Host: *Megan Morrill, Board of Directors*

Panelists: *Bill Gahl, MD, PhD, Rachel Bishop, MD, Elena Levtchenko, MD, PhD, Maya Doyle, MSW, PhD, Paul Grimm, MD, Jess Thoene, MD, Ewa Elenberg, MD, Med, Aude Servais, MD, PhD*

Kirkland Hall

Parents of Adults with Cystinosis Breakout

(closed session for parents of adults living with cystinosis)

Panelists: *Carol Hughes, Gail Potts, Terri Schleuder, Maya Doyle, MSW, PhD*

The purpose of the session is intended to be a private setting for parents of adults with cystinosis to share information, insight, advice, and encouragement. This session will provide an opportunity to learn from others who face similar challenges and allow you to talk about your experiences.

3:45 pm - 4:00 pm

Break

Parthenon
Ballroom Foyer

4:00 pm - 5:30 pm
Parthenon Ballroom

Medical Panel

Moderator: Bill Gahl, MD, PhD

Host: Christy Greeley, Executive Director, VP Research

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 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.

Dinner on your own for general attendees

Take this opportunity to explore Nashville!

6:30 pm - 7:30 pm
View on 2

ALAB Adult Meetup

Join the Adult Leadership Advisory Board and fellow adults with cystinosis for a mix and mingle. This is an opportunity to get to know others in the adult community to plan for a night out in Nashville or just a relaxing get together. Snacks and beverages will be provided.

6:45 pm

Speaker Dinner

*Closed Event - invited speakers/CRN Board
The 404 Kitchen | American Restaurant in
Nashville, TN (the404nashville.com)*

Bus will depart hotel at 6:30 pm

SATURDAY, JULY 15TH

7:00 am - 8:30 am

Parthenon Ballroom

Breakfast

Complimentary

8:00 am - 4:00 pm

Parthenon

Ballroom Foyer

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Franklin Hall

QUALIFY-US Study

Mona Suck, MSc

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sign their child into daycare each morning and SIGN OUT every time they come to get their child(ren) (ages 0-12). Parents need to take their children out of childcare for lunch. Dosing of medications can only be administered by parents, not nannies or volunteers. Snacks and plenty of water will be provided. Guaranteed to allow the kids to not only bond, but to have the time of their lives!

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8:30 am - 8:45 am
Parthenon Ballroom

Opening Comments
*Marybeth Kruppenacker,
VP Education & Awareness*

8:30 am - 9:30 am
Kirkland Hall

ALAB Board Meeting
Closed session - ALAB Board

8:45 am - 9:15 am
Parthenon Ballroom

Horizon Therapeutics Update
*Andrea Atherton and Tovah Toomasson,
Horizon Therapeutics*

9:15 am - 9:45 am
Parthenon Ballroom

**CYSTARAN: From Research
to an Approved Therapy**
Lesli King, Leadiant Biosciences

9:30 am - 11:30 am
Crescent Room

**ALAB Session for Adults and Teens
Living with Cystinosis**

Hosts: ALAB Board Members

The Adult Leadership Advisory Board of CRN are people living with cystinosis themselves, who seek to provide a safe, inclusive and supportive environment for all adults and teens living with cystinosis. We develop programs to inform and include and support adults with cystinosis. This session will give us a chance to meet each other, share our stories and develop ideas about future resources and services for the community.

9:45 am - 10:00 am
Parthenon Ballroom

Recordati Rare Diseases Update

Anna Vorobeva, Recordati Rare Diseases

10:00 am - 10:30 am
Parthenon Ballroom

**Cystinosis Network Europe/
Community Advisory Update**

Denise Dunne

Cystinosis Network Europe (CNE) is an umbrella grouping of patient support, advocacy and research organizations in Europe and beyond. The group originally came together informally to provide peer support and to support the hosting of a family and research conference every two years in Europe. CNE coordinates the Cystinosis Community Advisory Board (CAB) which is a group of patient representatives who offer their expertise to public or private sponsors of clinical and other research. This presentation will give an update on the work of CNE and the CAB in the last four years and show the value of including patient experts in the development of research.

10:30 am - 10:45 am Break

Parthenon Ballroom

10:45 am - 11:45 am Finding Our Roots

Parthenon Ballroom

Moderator: Jean Campbell

Panelists: Steve Groft, PharmD, Marybeth Krummenacker, Jess Thoene, MD, Bill Gahl, MD, PhD

A discussion and a history of how far we have come in living with a rare disease like cystinosis. From the initial diagnosis Marybeth received for her daughter Laura, we will discuss her initial visit to the NIH and meeting Dr. Gahl, what it means to be accepted into a research protocol and being a part of something she didn't really understand. NIH, FDA, NIDD - all acronyms to people and places that would play an integral part in not only her life but to a community that she didn't know existed. Dr. Gahl, Dr. Groft and Dr. Thoene will each address the importance of the relationships between government institutions and industry and ways to collaborate to help create hope and promise which was previously non-existent. We will also discuss the importance of the support of basic biomedical research funding, how it is funded and the critical need for diseases like cystinosis. All of this would have NEVER have happened without ADVOCATES. The beginnings of ADVOCATING for patients who didn't have a voice and how those collaborations and those voices - certainly RARE DISEASE voices - are now not only heard, but are highly respected and are often called upon as "the experts" in the community will be emphasized. CRN is a clear example of one of those organizations. The partnerships and the collaborations are immeasurable. The journey has been

extraordinary and we understand that it should not be taken for granted.

11:45 am - 12:30 pm Meeting the Challenge of Rare Disease in the Family: 25 Years of Lessons Learned

Parthenon Ballroom

Al Freedman, PhD

This session, led by a psychologist and the father of a young man who lived 26 years with a rare disease, will highlight the common challenges faced by rare families, with a focus on creating a path from fear to hope.

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12:30 pm - 1:30 pm Lunch

Parthenon Ballroom

Complimentary

1:30 pm - 2:00 pm QUALIFY-US – Development of a patient-reported outcome to measure the health-related quality of life of children and adolescents with cystinosis

Parthenon Ballroom

Mona Suck, MSc

The introduction of oral cysteamine therapy in the early 1980s has changed the course of cystinosis from a primarily lethal to a chronic multiorgan disease. Both the disease-specific symptoms as well as the therapy have a significant impact on health-related quality of life (HrQoL). The few studies examining HrQoL in patients with cystinosis suggest a limited HrQoL for patients and their caregivers. In recent years, validated disease-specific questionnaires have played an increasing role in contributing to improved care for patients. For example, by demonstrating the superiority of a drug or therapy on the basis of HrQoL. Studies

assessing HrQoL in patients with cystinosis primarily use generic questionnaires since no disease-specific patient-reported outcome measures (PROMs) exist so far. Such a questionnaire will be developed within the scope of the QUALIFY-US project, so that the relevant aspects of HrQoL from the patient's point of view can be considered in the treatment and also be included in the benefit assessment of new therapies. Within this presentation we would like to give you an insight into the topics of HrQoL and PROM development as well as to discuss with you which aspects you consider relevant with regard to cystinosis and your HrQoL.

2:00 pm - 3:00 pm
Parthenon Ballroom

Parents of Children and Adults with Cystinosis Panel

Moderator: Kristina Sevel, Board of Directors
Panelists: Heather Rothrock, Ina Gardner, Bryan and Chelsea Meschke, Kim Forrester, Tim Wyman

Panel presentation during which parents of children and adults with cystinosis 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. Parents of individuals at every stage of the disease will be featured and will share how they have managed the variety of challenges they have faced. Audience participation will be encouraged.

3:00 pm - 4:00 pm
Parthenon Ballroom

Adults Living with Cystinosis Panel

Moderator: Ella Balasa, ALAB Coordinator
Panelists: Jordan Janz, Megan Morill, Emily Mello, Katie Morrison, Jana Healy, Victor Gardner, Heidi Hughes

Panel presentation during which individuals living with cystinosis, including members of the Adult Leadership Advisory Board, will answer prepared questions and address topics related to the use of coping mechanisms and strategies for success through the ups and downs that Cystinosis brings. Audience participation will be encouraged.

4:00 pm - 4:30 pm

Parthenon Ballroom

Closing Remarks

*Jonathan Dicks, President, VP Development
Christy Greeley, Executive Director, VP
Research*

6:00 pm - 11:00 pm

Parthenon Ballroom

Farewell Dinner Dance

*Complimentary dinner/open bar
Sponsored by Horizon Therapeutics
All conference attendees – pack your dancing shoes for our final event of the week – the dinner dance! Before saying goodbye, recharge with a delicious dinner then show your moves on the dance floor! After all, this evening is to celebrate YOU and our cystinosis community.*