

9th INTERNATIONAL CYSTINOSIS CONGRESS BEYOND BORDERS

Valencia, SPAIN
30 June - 3 July, 2016

www.cystinosisfoundation.org



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**Celebrating the Lives
of Our Cystinosis Families**

**Excellence
in Pediatrics[®]**
INSTITUTE

*Welcome to the 9th International Cystinosis Congress Beyond Borders.
Walk Onward*

It is our honor and great pleasure to welcome you to Valencia. There are several reasons that it is entirely appropriate for us to host the international congress in this beautiful and ancient city. The oldest center for higher learning in Spain, the University of Valencia, was founded in 1499 and is still in operation. Not only the academic tradition resides here, but there is also the name itself. The original Latin name of this city was Valentia, meaning "strength" or "valor" and nowhere in the world is there a greater gathering of individuals of strength and valor than the people you see at this conference.

Everyone here is on their own special journey, blazing their own pathway through a sometimes unpredictable and hazardous course. You will meet other patients and parents sharing their experiences. As isolated and alone as we may feel at times, in truth we are not alone. That is why we come to these conferences. By sharing our experiences we gain even more knowledge.

You will meet scientific experts in the field of Cystinosis research, to whom we are grateful for their industry and perseverance. When we began this journey together there was no treatment for Cystinosis. Many of the scientists in this room contributed to bringing forth treatments and increasing our knowledge of the mechanisms of Cystinosis. Many of the patients in this room participated in clinical trials and studies and to them we give our deepest thanks.

There are many more individuals who gave of themselves over many years in order for us to be here today. Some members of our global community are no longer with us and we honor their memory and their numerous contributions.

An international committee created this agenda. Our deep gratitude to Maya Baressi, Gail Daniels, Roy Forsyth, Lisa Frost, Don Hammond, Merle Mund, Anne-Claire Panisset-Hill and Freek Wonnink. It is a program that integrates all that a person needs to grow in this life; information on research developments, nutrition, medication regimens, emotional support, exercise and most important of all, social bonds.

Together families embark on a truly unique experience this year. We are grateful to Peter Bourquin for leading our Cystinosis Family Constellation workshops with volunteers from our audience.

A special thank you to our scientific chairs, Gema Ariceta, MD, Elena Levtchenko, MD and Jennifer Simpson, MD. The mini-symposium format of this conference is intended to aid in understanding the vast amount of information that is laid before you over the next few days. It is a great pleasure collaborating with the Excellence in Pediatrics Institute and Russell Hale to increase awareness about Cystinosis and launching our Parents' Advice videos. Deepest thanks to Lourdes Sanz and the Grupo Cistinosis Espagna.

A special thank you to the many individuals who contribute to the Cystinosis Foundation and help make this conference a reality. Thank you to our industry sponsors, Orphan Europe Recordati Group, Sigma-Tau Pharmaceuticals and Raptor Pharmaceuticals. Your enduring partnership on this path has vastly improved lives of many people you will never know. The Board of Directors of the Cystinosis Foundation thanks everyone.

Valerie Hotz
Executive Director
Cystinosis Foundation

*The Cystinosis Foundation
Making a difference since 1983 for the child born today.*

Welcome to the 9th International Cystinosis Congress Beyond Borders

On behalf of the Cystinosis Support Group of Spain, it is our pleasure to welcome you to Valencia. For us it is a great joy to co-host the 9th International Cystinosis Congress in Spain.

Our group was born from the pressing need to connect our community. The diagnosis of an ultra-rare condition like Cystinosis is very isolating and we do not want families to feel alone. Knowing that there are others in the same circumstances helps.

Having a rare disease does not mean one must be alone. Walking this path together helps make life easier and together we are stronger. We require help, support and understanding on this path, as we are visible in society.

This was and remains our main objective.

Our Spain Cystinosis Group has been growing in recent years, with new families joining. In Spain there are approximately 60 people affected with Cystinosis. We are in contact with about 30 of these individuals and we hope to reach everyone. Some of our adult patients are more than 40 years of age. Some adult women patients are mothers and others expect to be soon. Most of our adult patients are working and younger children are studying in school. All are looking to grow their ways and learn more.

One of our goals is to share information from this conference with healthcare professionals at our hospitals where patients have Cystinosis.

We greatly appreciate that this year's congress will be held in our country. For us it is a great opportunity to learn about the latest research, treatments and developments in various fields and different aspects of Cystinosis. We also wish to strengthen ties between our many support groups and in so doing increase our global community.

We wish you a pleasant stay in Valencia. It is a city full of light, color and beside the Mediterranean Sea, so we are sure that you will enjoy its beauty and ancient history.

Lourdes Sanz
Secretaria AIRG-E
Coordinadora grupo cistinosis
www.grupocistinosis.org/www-airg-e.org

Bienvenidos al 9º Congreso Internacional de Cistinosis

Bienvenidos a Valencia, para nosotros es una alegría que el 9º Congreso Internacional de Cistinosis se celebre en España.

Nuestro grupo Cistinosis nació de la necesidad de: No sentirnos solos frente el diagnóstico de una enfermedad rara. Saber que hay otras personas en las mismas circunstancias. La necesidad de ayudarnos, apoyarnos y comprendernos en este camino y darnos a conocer y ser visibles en la sociedad. Tener una enfermedad rara no significa estar solos. Este grupo es necesario porque: ¡Juntos es más fácil! ¡Juntos somos más fuertes!

Este fue y sigue siendo nuestro principal objetivo.

El grupo de cistinosis España está creciendo en los últimos años. Cada vez somos más las familias que estamos en contacto. En España hay aproximadamente 60 personas afectadas con cistinosis, de las cuales estamos en contacto unas 30, pero esperamos conseguir llegar a todos. Nuestras pacientes adultas ya tienen más de 40 años, algunas de ellas son madres y otras esperan serlo pronto, la mayoría de los pacientes adultos trabajan, los más jóvenes están estudiando y los niños crecen buscando sus caminos.

Uno de nuestros objetivos es, que toda la información del congreso llegue a todos los hospitales donde tengan pacientes con cistinosis.

Agradecemos enormemente que este año el congreso se celebre en nuestro país. Para nosotros es una gran oportunidad para conocer los últimos avances en investigación, tratamientos y novedades en diferentes ámbitos y aspectos. Deseamos también que los lazos entre toda la comunidad se vayan estrechando y fortaleciendo cada día más.

Esperamos que tengáis una agradable estancia en Valencia. Es una ciudad llena de luz, color y al lado del Mar Mediterráneo, por lo que estamos seguros que la disfrutareis.

Lourdes Sanz
Secretaria AIRG-E
Coordinadora grupo cistinosis
www.grupocistinosis.org/www-airg-e.org

It is a great satisfaction and honour to welcome you to Valencia for the 9th International Congress of the Cystinosis Foundation. Thank you for choosing Spain and giving us the opportunity to host this conference in a new format, offering mini-symposiums to increase understanding of Cystinosis. We hope to contribute to its success and to help this sharing experience to be unforgettable and enjoyable.

This congress provides a unique scenario to educate and promote innovation, support patient-centred actions, and discover what therapeutic and research ideas are the important ones for patients with Cystinosis. This conference will also allow adopting a global approach to a disease that does not know borders. We will receive information about the way patients with Cystinosis live and develop in different countries, with the aim of helping each other, and reinforce friendships and partnerships between us. I am convinced that all together we will be able to share expertise, share our new knowledge about Cystinosis, and move forward to better management of this condition. The fact we are celebrating this International Cystinosis Congress Beyond Borders at the Mediterranean Sea, represents the symbol of different countries belonging to the same community of travellers, joining together towards a future of hope and healing on our journeys.

On behalf of the Spanish Cystinosis Community we wish you a nice, productive, and happy stay in Valencia. We are ready to make it real. Thank you again for your confidence and trust in our people. We are looking forward to welcoming everyone.

Dr. Gema Ariceta
University Hospital Vall d'Hebron

Thursday, 30 June

17:00 - 19:30 **Welcome** Reception and Registration
Screening of Parents' Talk Videos

Friday, 1 July

07:00 - 08:30 **Buffet Breakfast**

PLENARY SESSION

08:30 - 08:35 **Welcome Remarks** : G. Ariceta, L. Sanz, V. Hotz
08:35 - 08:45 Presentation of Hobbs Humanitarian Award • V. Hotz, L. Sanz
Introduction of Support Group Leaders

PATIENTS AND FAMILIES ONLY PLENARY SESSIONS

08:45 - 08:55 Introduction of Speaker • L. Sanz, V. Hotz
08:55 - 09:30 Peter Bourquin Keynote Speaker "Cystinosis Family Constellation"
09:30 - 10:30 Focus: Patient Workshop • P. Bourquin
10:30 - 10:50 Coffee Break
11:50 - 11:50 Focus: Parent Workshop • P. Bourquin
11:50 - 12:50 Focus: Sibling Workshop • P. Bourquin
12:50 pm - 13:50 pm Lunch

PLENARY SESSION

13:50 - 14:20 Genetics of Cystinosis, a metabolic condition • W. Gahl
14:20 - 14:35 Managing Fanconi Syndrome • P. Goodyer
14:35 - 14:50 Renal Transplantation from A to Z • P. Grimm
14:50 - 15:10 Questions and Answers
15:10 - 15:25 Coffee Break

MINI-SYMPOSIUM "BE STRONG"

Development of Muscle Mass and Bone Density in Cystinosis • Moderated by P. Niaudet, MD

15:25 - 15:40 Muscle wasting in Cystinosis • D. Trauner
15:40 - 15:55 Feeding in Cystinosis • R. Dohil
15:55 - 16:10 Impact of Cystinosis on bone and muscle development • M. Leonard
16:10 - 16:30 Questions and Answers
16:30 - 17:15 Parents Panel Answering Questions from other Parents • M. Bos, G. Daniels, L. Sanz, S. Scott
17:15 - 18:15 Rare disease multi-specialty clinics; How it is done at my center Elena Levtchenko, MD will moderate this panel of medical experts discussing how their Cystinosis Clinic operates • A. Ariceta, W. Gahl, M. Greco, P. Grimm, K. Hohenfellner, M. Janssen, P. Niaudet, D. Trauner, A. Tsygin
19:30 - 21:30 Open Bus Ride and Tour of Valencia

Friday, 1 July
PARALLEL SESSIONS

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| 09:00 - 10:30 | Professionals only meeting |
| 10:30 - 10:50 | Coffee Break |
| 10:50 - 12:45 | Professionals only meeting |
| 14:05 - 15:05 | Parents of Adult Patients ONLY - This group discussion is restricted to individuals who are parents or caregivers of adult Cystinosis patients. Concerns and needs of adult Cystinosis patients will be addressed |
| 15:25 - 16:25 | Share the Bread - Panel discussion for Cystinosis patients only. Age 12 and above. • <i>P. Bourquin, D. Daniels, M. Drury, J. Hotz, F. Wonnink</i> |
| 14:00 - 18:00 | Team Building Art Project - Drop in with your family and create your own contribution to the Cystinosis Family Quilt, 2016 |

Saturday, 2 July

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| 07:00 - 08:30 | Buffet Breakfast |
| PLENARY SESSION | |
| 08:30 - 08:40 | Welcome Remarks : <i>G. Ariceta, L. Sanz, V. Hotz</i> |
| 08:40 - 09:40 | Laughter Team Event |
| 09:40 - 10:25 | Cystinosis Family Affair - Panel of Adult Patients Sharing Their Pathways • <i>D. Daniels, M. Drury, J. Hotz, S. Scott, M. Tromp, F. Wonnink</i> |
| 10:25 - 10:45 | Coffee Break |
| MINI-SYMPOSIUM "SEE THE WORLD" | |
| Cystinosis and the Visual System • Moderated by <i>J. Simpson, MD</i> | |
| 10:45 - 10:55 | Overview of ocular complications • <i>J. Simpson</i> |
| 10:55 - 11:10 | Novel diagnostic modalities for corneal cystinosis and Update on Cysteamine gel formulation • <i>L. Hong</i> |
| 11:10 - 11:25 | Ocular cystinosis without treatment • <i>R. Lopez-Ladron</i> |
| 11:25 - 11:40 | Intracranial hypertension and Nephropathic Cystinosis • <i>N. Martin Begue</i> |
| 11:40 - 11:55 | From theory to practice • <i>M. Hua</i> |
| 11:55 - 12:10 | Visual spatial deficit in Cystinosis and school related challenges • <i>D. Trauner</i> |
| 12:10 - 12:35 | Questions and Answers |
| 12:35 - 12:45 | Our Heroes • <i>D. Hammond, V. Hotz</i> |
| 12:45 - 13:50 | Lunch |
| 13:50 - 14:50 | Poster Session - Displays from researchers and industry. Authors are present to discuss their work. Patients and families are able to discuss issues with medical experts on an individual basis. |

MINI-SYMPOSIUM "LEARN AND LOVE"
Transitioning to Adulthood in the Face of Cystinosis • Moderated by *E. Levtchenko, MD*

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| 14:50 - 15:05 | Endocrine system in Cystinosis • <i>A. Bertholet-Thomas</i> |
| 15:05 - 15:20 | Medication compliance issues • <i>G. Ariceta</i> |
| 15:20 - 15:35 | Male Fertility • <i>E. Levtchenko</i> |
| 15:35 - 15:50 | Pregnancy • <i>A. Servais</i> |
| 15:50 - 16:05 | Care of adult Cystinosis patients • <i>A. Servais</i> |
| 16:05 - 16:25 | Questions and Answers |
| 16:25 - 16:40 | Coffee Break |

MINI SYMPOSIUM "THE FUTURE IS BRIGHT"

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| Novel Drugs, Diagnostics and Therapeutics in Cystinosis • Moderated by <i>G. Ariceta, MD</i> | |
| 16:40 - 16:55 | Update on delayed-release Cysteamine • <i>R. Dohil</i> |
| 16:55 - 17:10 | Drug Library Screening in Cystinosis • <i>F. Emma</i> |
| 17:10 - 17:25 | Prodrug Update • <i>R. Anderson</i> |
| 17:25 - 17:40 | High definition optical coherence tomography in monitoring Nephropathic Cystinosis • <i>K. Veys</i> |
| 17:40 - 18:00 | Stem cell transplantation and gene therapy • <i>C. Rocca</i> |
| 18:00 - 18:20 | Questions and Answers |
| 20:00 - 23:00 | Gala Dinner Celebration. Presentation of Cystinosis Family Quilt 2016 |

Saturday, 2 July
PARALLEL SESSIONS

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| 08:00 - 14:00 | Drop in and create your own contribution to the Cystinosis Family Quilt 2016 |
| 11:40 - 11:55 | Cure Cystinosis International Registry • <i>V. Hotz</i> |
| 15:30 - 16:00 | Administering Cysteamine Eye Treatment • <i>J. Simpson, N. Martin-Begue</i> Learn the technique for efficient administration of eye drops from pediatric ophthalmologists |

Sunday, 3 July

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| 09:00 - 09:05 | Welcome Remarks : <i>G. Ariceta, L. Sanz, V. Hotz</i> |
| 09:05 - 10:05 | Cystinosis patient care around the globe • <i>A. Bertholet-Thomas, M. Janssen, N. Soliman, R. Topaloglu, M. Kagan, A. Tsygin, R. Hoseini</i> |
| 10:05 - 11:05 | Preserving renal function - Questions and Answers Forum for All • <i>F. Emma, P. Niaudet, R. Topaloglu, E. Levtchenko</i> |
| 11:05 - 11:45 | Gastrointestinal/Feeding and Questions about G-tube Questions and Answers Forum for All • <i>R. Dohil, P. Goodyer</i> |
| 11:45 - 12:00 | Farewell until 2018! • <i>G. Ariceta, L. Sanz, V. Hotz</i> |

