



★ *The Cystinosis Foundation*

6th International Cystinosis Conference

September 23 – 26, 2010

Congress Village Ge.Tur.
Viale Centrale, 29
33054 Lignano Sabbiadoro (UD) ITALY
<http://www.getur.com>
midway (100 km) between Venice and Trieste

CONFERENCE PROGRAMME

THURSDAY, SEPTEMBER 23, 2010

17.00 - 20.00 Registration and Welcome
19.30 **DINNER**

FRIDAY, SEPTEMBER 24, 2010

07.00 - 07.45 CONFERENCE REGISTRATION
07.45 - 08.00 WELCOME & INTRODUCTION
F. EMMA, M. MAZZINA, J. HOBBS - HOTZ
08.00 - 08.30 50 years of cystinosis research and treatment
J. SCHNEIDER
08.30 - 10.10 GENETICS AND REGISTRIES
08.30 - 08.50 Genetics of cystinosis and prenatal counselling

UNDER THE PATRONAGE OF
REGIONE AUTONOMA FRIULI VENEZIA GIULIA
ISTITUTO SUPERIORE DI SANITÀ



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- 08.50 - 09.10 C. ANTIGNAC
The new European Registry
P. NIAUDET
- 09.10 - 09.30 Cure Cystinosis International Registry
V. HOTZ
- 09.30 - 09.50 Cystinosis in emerging countries
R. TOPALOGLU, A. TSYGIN
- 09.50 - 10.10 Time for questions
- 10.15 - 10.30 **COFFEE BREAK**
- 10.30 - 12.30 LONG-TERM OUTCOME AND NEW TREATMENTS
- 10.30 - 10.50 Long-term outcome in cystinosis
W. GAHL
- 10.50 - 11.10 Eye problems and new cysteamine collyriums
M. DEL MONTE
- 11.10 - 11.30 Muscle wasting in nephropathic cystinosis
D. TRAUNER
- 11.30 - 11.50 Update on cysteamine treatment and adverse events
E. LEVTCHENKO
- 11.50 - 12.10 Long-acting cysteamine
R. DOHIL
- 12.10 - 12.30 Time for questions
- 12.30 - 13.30 **LUNCH**
- 13.30 - 14.30 WORKSHOP FOR CHILDREN (6-12) WITH PARENTS PRESENT
F. EMMA
- 13.30 - 14.30 WORKSHOP FOR ADOLESCENTS AND YOUNG ADULTS
W. GAHL, M. OSTERMANN
- 14.30 - 14.50 Cognitive performance in cystinosis
D. TRAUNER
- 14.50 - 15.10 Psychological aspects and medication compliance
M. OSTERMANN
- 15.10 - 15.30 Time for questions
- 15.30 - 15.45 **COFFEE BREAK**
- 15.45 - 16.45 Adult patients and Parents Informal Discussion
P. COMBATTELLI, S. SCOTT, J. TERRY
- 16.45 - 17.05 Physical Activity Regime
M. MAXWELL
Parents retrieve children and everyone meet at pool



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17:45 - 18:30 Water Aerobics
M. MAXWELL

Session for Professionals

LESSONS FROM OTHER DISEASES

E. LEVTCHENKO

15.45 - 16.05 Molecular biology of Lowe disease

A. DE MATTEIS

16.05 - 16.25 Murine models of proximal tubular defects

E. CHRISTENSEN

16.25 - 16.45 Altered endocytosis in renal Fanconi syndromes

P. COURTOY

16.45 - 17.00 **BEVERAGE BREAK**

Session for Professionals

NEW FRONTIERS

C. ANTIGNAC

17.00 - 17.20 Bone marrow transplantation

S. CHERQUI

17.20 - 17.40 Gene therapy for metabolic diseases

F. FERRUA

17.40 - 18.00 Mesenchymal stem cells

P. GOODYER

18.00 - 18.30 Zinc finger nucleases for homology directed repair in cystinosis

P. HARRISON

17.30 Child care closes

19.30 Gala Dinner and Tribute to Dr. Schneider



SATURDAY, SEPTEMBER 25, 2010

Families & Professionals

- 08.00 - 08.10 Opening Remarks
08.10 - 08.30 Children's information on cystinosis
W. VAN'T HOFF
08.30 - 08.50 Lay presentation on bone marrow transplantation for families
S. CHERQUI
08.50 - 09.10 Lay presentation on gene therapy for families
S. CHERQUI
09.10 - 09.30 Time for questions
- 09.30 - 09.50 KIDNEY, GASTROINTESTINAL PROBLEMS AND GROWTH
Treatment of Fanconi Syndrome
P. GOODYER
- 09.50 - 10.10 **COFFEE BREAK**
- 10.10 - 10.30 Kidney transplantation – steroid avoidance protocols
P. NIAUDET
10.30 - 10.50 Growth, puberty and sexual development
M. GRECO
10.50 - 11.10 Gastro-intestinal problems related to cystinosis
R. DOHIL
11.10 - 11.45 Time for questions
- 11.45 - 13.15 **LUNCH**
- 13.15 - 14.30 WORKSHOP FOR ADULT PATIENTS
W. VAN' HOFF, M. OSTERMANN
- 13.15 - 15.00 POSTER SESSION / ROUND TABLE
Posters and displays from researchers, industry groups and others. Authors are present to discuss their work. In addition, professionals will sit at tables and Patients/families will be able to discuss issues with them on an individual basis.
- 15.00 - 15.15 **COFFEE BREAK**

Q&A SESSIONS W/ PANELS OF EXPERTS + 2 PARENTS/PATIENTS

- 15.15 - 16.15 Preserving renal function
R. TOPALOGLU, E. LEVTCHENKO, P. NIAUDET



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16.15 - 17.15 Gastrointestinal/pancreas/feeding/growth problems

R. DOHIL, M. GRECO, P. GOODYER

17.15 - 18.15 Neurological/psychological aspects

M. OSTERMANN, D. TRAUNER, W. VAN'T HOFF

18.15 Child care closes

19.30 *DINNER*

SUNDAY, SEPTEMBER 26, 2010

09.00 - 10.00 LIVING WITH CYSTINOSIS:

A discussion on living with cystinosis, focusing on education,
issues related to overprotection, transition to adult life and coping.

M. BOS, J. SHAHAR, Y. SHAHAR

10.00 - 10.30 Presentation of W.L. Hobbs Award.

10.30 - 11.00 Closing remarks and conclusion of conference.

F. EMMA, M. MAZZINA, J. HOBBS-HOTZ