

EUNEFRON Symposium on Rare Inherited Kidney Disorders

Date: March 9–10, 2012



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Venue: Charité Hospital, Campus Mitte, Berlin
Lecture hall Internal Medicine, south wing
Sauerbruchweg 2, 10117 Berlin

For further information about this program, please contact
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This project is supported through Coordination Theme 1 (Health) of the European Community's FP7,
Grant agreement number HEALTH-F2-2008-201590

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Friday, March 9th, 2012

8:30 – 8:55

Registration

8:55 – 9:00

Welcome **O. Devuyst, D. Müller**

9:00 – 10:30

Session 1: Orphan nephropathies: new perspectives

9:00 – 9:30

L. Guay-Woodford: *Perspective from pediatric nephrology*

9:30 – 10:00

J-P. Grunfeld: *Perspective from adult nephrology*

10:00 – 10:30

H. Murer: *Perspective from physiology*

10:30 – 11:00

Coffee break

11:00 – 12:30

Session 2: Cystinosis

11:00 – 11:30

F. Emma: *Update on cystinosis*

11:30 – 11:50

C. Antignac: *Proteins interacting with cystinosis*

11:50 – 12:10

P. Courtoy/H. Gaide Chevronnay: *Physiopathology of cystinosis progression in CTNS^{-/-}*

12:10 – 12:30

E. Levtchenko: *Renal epithelial cell dysfunction in cystinosis*

12:30 – 14:00

Lunch break + **Poster session 1**

14:00 – 16:50

Session 3: From Mendelian disorders to GWAS

14:00 – 14:30

A. Köttgen: *GWAS in nephrology*

14:30 – 14:50

P. Ronco/H. Debiec: *Immunopathology: An example of membranous nephropathy*

14:50 – 15:10

E. Christensen: *Proximal tubular endocytosis: From mouse models to human disease*

15:10 – 15:30

Y. Pirson/K. Dahan: *Familial chronic interstitial nephritis: UMOD gene and what else?*

15:30 – 15:50

L. Rampoldi: *Uromodulin-associated kidney disease*

15:50 – 16:20

Coffee break

16:20 – 16:50

R. Kleta: *Next generation sequencing for rare diseases*

17:45 – 22:15

EUNEFRON social event (PIs, Invited Speakers, Associate members and invited guests)

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Saturday, March 10th, 2012

From 8:15 Arrival and coffee

8:45 – 10:15 Session 4: Differentiation and developmental disorders

8:45 – 9:15 **M. Pontoglio:** *HNF1 transcription factors*

9:15 – 9:35 **N. Knoers:** *The importance of exome sequencing in nephrogenetics*

9:35 – 9:55 **O. Devuyst:** *Transcriptional factors in the proximal tubule: From development to disease*

9:55 – 10:15 **P. Ronco/E. Plaisier:** *Basement membrane collagen diseases: Novel roles for COL4A1*

10:15 – 10:45 Coffee break

10:45 – 14:15 Session 5: Distal tubule and magnesium disorders

10:45 – 11:15 **J. Hoenderop/R. Bindels:** *Disorders of magnesium handling*

11:15 – 11:35 **D. Müller:** *From men to mice and back: CNNM2 and magnesium homeostasis*

11:35 – 11:55 **X. Jeunemaitre:** *A new pathway for Na/K reabsorption and NCC regulation*

11:55 – 12:15 **C. Wagner:** *Renal acid excretion - new insights into mechanisms and diseases*

12:15 – 12:35 **P. Deen:** *Towards a better (understanding of) treatment for nephrogenic diabetes insipidus*

12:35 – 13:45 Lunch break + **Poster session 2**

13:45 – 14:15 **R. Unwin:** *Effect of calcineurin inhibitors on the distal convoluted tubule*

14:15 – 16:25 Session 6: Policies and registries

14:15 – 14:45 **S. Parker:** *European perspective on registries*

14:45 – 15:05 **R. Vargas/D. Kahila:** *The example of tubulopathies databases*

15:05 – 15:25 **W. van 't Hoff:** *The EUNEFRON cystinosis study*

15:25 – 15:45 Coffee break

15:45 – 16:00 **X. Wang:** *Rare inherited kidney diseases in China*

16:00 – 16:15 **M. Taylor:** *RaDaR – Rare disease plan in UK*

16:15 – 16:25 **O. Devuyst:** *Lessons and insights from EUNEFRON*

16:25 Departure

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