EUNEFRON
European Network for the Study of Orphan Nephropathies
www.eunefron.org

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
Iwan Meij, *EUNEFRON* Project Manager, i.meij@mdc-berlin.de or call +49 30 9406 4225

This project is supported through Coordination Theme 1 (Health) of the European Community's FP7,
Grant agreement number HEALTH-F2-2008-201590

SEVENTH FRAMEWORK PROGRAMME

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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 cystinosin*
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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Please visit www.eunefron.org for free registration and more information

Speakers:

C. Antignac, *FR: Cystinosin interacting proteins*
E. Christensen, *DK: Proximal tubular endocytosis*
P. Courtoy/H. Gaide Chevronnay, *BE: Cystinosis in CTNS$^{-/-}$*
P. Deen, *NL: Nephrogenic diabetes insipidus treatment*
O. Devuyst, *CH: Transcriptional factors in proximal tubule*
F. Emma, *IT: Update on cystinosis*
J-P. Grunfeld, *FR: Adult orphan nephropathies*
L. Guay-Woodford, *UK: Pediatric orphan nephropathies*
J. Hoenderop/R. Bindels, *NL: Magnesium handling disorders*
X. Jeunemaitre, *FR: Na/K reabsorption and NCC regulation*
R. Kleta, *UK: Next generation sequencing*
N. Knoers, *NL: Exome sequencing in nephrogenetics*
A. Köttgen, *DE: GWAS in nephrology*
E. Levtchenko, *BE: Epithelial dysfunction in cystinosis*
D. Müller, *DE: CNNM2 and magnesium homeostasis*
S. Parker, *FR: European perspective on registries*
Y. Pirson/K. Dahan, *BE: Chronic interstitial nephritis*
M. Pontoglio, *FR: HNF1 transcription factors*
L. Rampoldi, *IT: Uromodulin-associated kidney disease*
P. Ronco/H. Debiec/E. Plaisier, *FR: Immunopathology/COL4A1*
M. Taylor, *UK: Rare disease plan in the UK*
R. Unwin, *UK: Calcineurin inhibitors*
W. van 't Hoff, *UK: The Eunefron cystinosis study*
R. Vargas/D. Kahila, *FR: Tubulopathy databases*
C. Wagner, *CH: Renal acid excretion*
X. Wang, *CH: Kidney disease registries in China*

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