

## SYSCILIA Newsletter 6 – June 2012

### 2<sup>nd</sup> Annual Meeting very successful

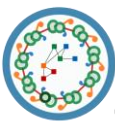
On May 14 – 16, SYSCILIA welcomed 47 consortium members and 4 Scientific Advisory Board members to the **second annual meeting** of the project at the Institute of Child Health in **London**.



Photo: [Toby Gibson](#)

The progress of the project and the latest results (some even finalized at the meeting) were presented by representatives of all workpackages, and these as well as future directions were critically and vividly discussed. The interactive way in which work was discussed and planned, resulted in the initiation of a “Project Shop” for which subjects were already discussed during the brain storm session on Wednesday. The participants were subdivided into 5 groups/topics and results, including timeline and lead partners, were presented at the end of the morning.

A very warm ‘thank you’ to all speakers for their exciting presentations, as well as for sharing their latest unpublished results, and to all participants for their fruitful discussions and ample willingness to foster new collaborations: you all made the meeting very successful!



Photos: **Toby Gibson**

A special 'thank you' is warranted for all 4 members of our Scientific Advisory Board who very actively participated in the discussions. They drafted a report with their remarks at the end of the meeting, which we will use, together with their other suggestions, to further strengthen our research.

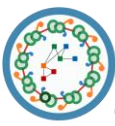
Their overall impression was very positive, and reflected the general open and positive character of our second annual meeting:

"We are impressed with the interdisciplinarity of the whole program and with the spectrum of expertise and the interactive way in which the work has been planned and executed in many work-packages...

.... In conclusion, we enjoyed hearing so much strong new data presented and congratulate the consortium on their achievements to date. We look forward to seeing more progress over the coming year."

## **CILIA 2012 - Cilia in Development and Disease**

The first international conference in Europe for the cilia scientific and clinical community initiated by the Ciliopathy Alliance and supported by SYSCILIA, took place in London on May 16-18, 2012, right after the SYSCILIA annual meeting. It was the first international scientific conference devoted to Cilia in Development and Disease, and had a massive response of over 280 registrants. An additional 50 applicants unfortunately had to be disappointed, as the maximum capacity of the venue at the UCL Institute of Child Health was reached.



On May 16 the Conference started with an evening reception with the Ciliopathy Alliance patient groups and VIP guests. Joe Gleeson gave his keynote talk at this evening.

The Ciliopathy Alliance brings together patient support groups, researchers, doctors and allied health professionals representing patients and families living with and affected by **ciliopathies** - diseases caused by defects in the function of cilia.

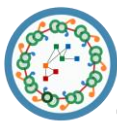


On May 17 and 18, the cilia community was presented with an impressive line-up of both keynote speakers that represented the leaders in the cilia- and ciliopathy field and exciting papers selected for oral presentations from submitted abstracts. SYSCILIA was well-represented in both categories. In between the talks, about 150 posters of nearly equal high quality were presented, many of which contained unpublished work, which also made the scientific content of the conference of outstanding quality.



Photo: Toby Gibson





On Thursday evening there was the Gala Dinner in the Titanic Ballroom at the Hotel Russell. It attributed to the nice, friendly and interacting atmosphere during the conference. The dinner was enlivened by a musical performance on harp and flutes by an ensemble, of which two members were ciliopathy patients themselves (suffering from congenital blindness, LCA).



Kate Osborne

Photos: Toby Gibson



James Risdon.



Frederica Mossone

I think we speak not only on behalf of the SYSCILIA partners present, but on behalf of the entire cilia community when we express our tremendous 'thank you' to **Phil Beales** and his organizing committee for this vibrant and inspiring conference. The conjunction with our Annual meeting was a perfect opportunity to indulge in a full week of ciliary research, meet with patients, clinicians and industry, and explore new collaborative avenues that will further boost our success.

### Pre-ARVO conference on Retinal Ciliopathies

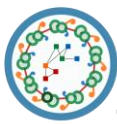
Just prior to the SYSCILIA annual meeting, on May 4-5, a more focused event was held as a pre-conference to the annual Association for Research in Vision and Ophthalmology (ARVO) meeting in Ft. Lauderdale, USA. This concerned the 14th Vision research Conference, with this year's theme "**Retina Ciliopathies, from Genes to Mechanisms and Treatment**", co-organized by **Marius Ueffing** who also chaired the session on non-syndromic retinal ciliopathies. SYSCILIA research was importantly presented in invited talks in the sessions focusing on "Cilia, Intraflagellar Transport and Disease" (**Karsten Boldt**), "Syndromic Ciliopathies" (**Nico Katsanis** and **Uwe Wolfrum**), and "Ciliary Systems Biology" (**Ronald Roepman**, also chair of this session). Ronald Roepman used this opportunity to inform the ciliary community about the concept of and progress made in SYSCILIA, which generated a lot of interest and inquiries to connect to the project.

### Publications and Press releases

**Rachel Giles**, Partner 8, UMCU is co-author on a publication in **Nature Genetics**, accepted 13/5/2012.

***FAN1 mutations cause karyomegalic interstitial nephritis, linking chronic kidney disease to DNA damage response signaling.***

Zhou W, Otto E, Cluckey A, Airik R, Hurd T, Chaki M, Diaz K, Lach F, Bennet G, Ghosh A, Natarajan S, Thongthip S, Veturi U, Allen S, Janssen S, Ramaswami G., Dixon J, Burkhalter F, Spoendin M, Moch H, Mihatch M, Verine J, Reade R, Sollman H, Godin M, Kiss D, Monga G, Mazzucco G, Amann K, Artunc F, Newland R, Wiech T, Zschiedrich S, Huber T, Slaats G, Joles J, Goldschmeding R, **Giles RH**, Washburn J, Levy S, Smogorzewska A, Hildebrandt F.



Several SYSCILIA partners are co-author on a publication in *Cell*, accepted 13/2/2012.

***Exome capture reveals mutations of MRE11, ZNF423 and CEP164, linking renal ciliopathies to DNA damage response signaling.***

Moumita Chaki, Rannar Airik, Amiya K. Ghosh, **Rachel H. Giles**, Rui Chen, Gisela G. Slaats, Hui Wang, Toby W. Hurd, Weibin Zhou, Andrew Cluckey, Heon-Yung Gee, Gokul Ramaswami, Chervenka, Ranjani Sri Ganji, Vitezslav Bryja, Heleen H. Arts, Jeroen van Reeuwijk, Machteld M. Oud, Stef J.F. Letteboer, **Ronald Roepman**, Hervé Husson, Oxana Ibraghimov-Beskrovnaya, Takayuki Ysunaga, **Gerd Walz**, Lorraine Eley, John A. Sayer, Bernhard Schermer, Max C. Liebau, Thomas Benzing, Stephanie Le Corre, Iain Drummond, Jaap A. Joles, Sabine Janssen, Susan J. Allen, Sivakumar Natarajan, John F. O. Toole, Massimo Attanasio, Sophie Saunier, Corinne Antignac, Robert K. Koenekoop, Huanan Ren, Irma Lopez, Ahmet Nayir, Corinne Stoetzel, Helene Dollfus, Rustin Massoudi, Joseph G. Gleeson, Sharon P. Andreoli, Dan G. Doherty, Anna Lindstrad, Christelle Golzio, **Nicholas Katsanis**, Lars Pape, Emad B. Abboud, Ali A. Al-Rajhi, Richard A. Lewis, James R. Lupski, Roel Goldschmeding, **Heymut Omran**, Eva Lee, Shaohui Wang, JoAnn M. Sekiguchi, Rudel Saunders, **Colin A. Johnson**, Elizabeth Garner, Jens Andersen, Joseph Shlomei, Gudrun Nurnberg, Peter Nurnberg, Shawn Levy, Agata Smogorzewska, Edgar A. Otto & Friedhelm Hildebrandt.

**Nico Katsanis** and Erica Davis are the authors of the article *“The ciliopathies: a transitional model into systems biology of human genetic disease”* in *Current Opinion in Genetics & Development* 2012, 22:1–14.

For a complete overview of publications with SYSCILIA acknowledgement, please refer to our website: [syscilia.org/results](http://syscilia.org/results).

## Calendar 2012

**Webinars**, online training sessions with different topics using the SysWiz database, are planned once every month. These trainings are available for every participant, see the SYSCILIA private website.

**IT-meetings, monthly via Skype**, between SYSCILIA partners about data exchange and analysis

**28 June 2012 – deadline for 2<sup>nd</sup> annual report SYSCILIA**

**European Human Genetics Conference 2012 (ESHG 2012)**

23 – 26 June 2012; Nürnberg Convention Center Ost, Nürnberg, Germany

**4th Conference on Systems Biology of Mammalian Cells (SBMC)**

9 – 11 Jul 2012; Gewandhaus zu Leipzig, Germany

**[ICSB2012](#)**

19 – 23 Aug 2012; Toronto, Canada

**Midterm Review by the European Committee**

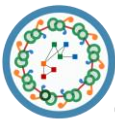
20 & 21 September 2012; Heidelberg, Germany.

**[7th French Meeting on Cilia, Flagella and Centrioles](#)**

22-24 Oct, 2012; Calanque des Eaux Salées, 13620 Carry-le-Rouet, France

**[American Society of Nephrology 2012](#)**

30 Oct – 4 Nov 2012; San Diego, California, USA



[The American Society of Human Genetics](#), 62nd Annual meeting

6 – 10 Nov 2012; San Francisco, USA

**2013 – Cross Component meeting SYSCILIA.**

16 – 18 January 2013; Heidelberg, Germany

Do you have any news item that you would like to see announced in this Newsletter? Input can be send to:  
[s.dusseljee@gen.umcn.nl](mailto:s.dusseljee@gen.umcn.nl)

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