

SYSCILIA Newsletter 4 – January 2012

Annual report.

The Annual Report for period 1 has received the final comments by the European Committee on January 19, 2012. From some partners more elaborate or detailed information is requested concerning the scientific reporting. The modifications are requested in order to set a good example for the first progress report which will be followed in the next reporting periods. Submission of the revised version of the report will be done at the latest on January 30, after which the payment requests can be processed.

Semi-annual report month 18

A draft of this internal report has been distributed amongst the PIs just before the cross-component meeting in Heidelberg (January 11-12). A final version will be generated soon and will then be published on the wiki, available for all SYSCILIA participants.

Cross-Component meeting Heidelberg, 11-12 January 2012.

Venue: Greenier Room, 7th floor, BioQuant building, University of Heidelberg.

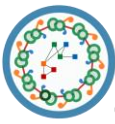
On January 11, 2012 the cross-component meeting was preceded by a very interesting and educational training on project management, given by Dr. Robert Lang, M.D., M.Sc.



Dr. Lang is trained as a General Surgeon, specializing in endoscopic surgery. He quit the clinical career with the upcoming of the DRG reimbursement system in Germany and embarked into hospital management. Worked as Head of the Department “Medical Controlling and Strategy Development” until 2008. Last position was as a Medical Director in Abu Dhabi, UAE.

Now working internationally as a trainer, coach and lecturer with a focus on ethical leadership, identification of bad leadership, soft skills and intercultural training. Teaches corporate ethics leadership ethics, international project management and intercultural management at several universities and business schools. Appointed professor Health Care Management from April 2012.

The cross-component meeting started after an introduction by Ronald Roepman with presentations and discussions from the bio-informatics, Rob Russell (UHEI), Toby Gibson



(EMBL), Konstantinos Koutroumpas (CNRS), John van Dam (RUNMC) and Gordana Apic (CCNet).

The meeting focused on training and workshops. For SYSCILIA participants the details and presentations can be found on the wiki page 'meetings' after login.

Besides presentations, members decided to split into four groups each with a different topic. Discussion in groups, then reassemble all participants and a summary was presented to the whole meeting.



Theme 1) **Cilia Genotype/phenotype**

Theme 2) **Structure BBSome Group**

Theme 3) **Edgetics Group – mapping mutations for understanding ciliopathies**

Theme 4) **Therapies/drugs/pathways Group - Computational models to animal models**

This successful and fruitful meeting will be held annually so we meet again in Heidelberg next year.

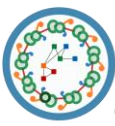
Publications and Press releases

We are very proud to inform you that for the third month in a row an article with SYSCILIA acknowledgement has been published in the high impact journal 'American Journal of Human Genetics'.

It is still a special thrill to identify a new genetic cause for an inherited condition. It is particularly satisfying when the results can be translated into clinical benefit through improvements in diagnosis or genetic counseling. Once the genetic cause for a condition is known, a succession of insights into the function of the encoded protein in the healthy and disease state is then possible.

Chronic renal disease and skeletal defects due to mutations in WDR19/IFT144. [Am J Hum Genet.](#) (R. Roepman)

Expanding the role of the TMEM family of proteins at the ciliary transition zone. [Am J Hum Genet.](#) (N. Katsanis, O. Blacque, C.A. Johnson)



Mutations in a ciliary-expressed gene, *C8orf37*, lead to disrupted ciliary processes and are associated with the pathogenesis of retinal dystrophies. [Am J Hum Genet](#) (R. Roepman).

Beside the publication in AJHG, Colin Johnson and co-workers also published an article in Human Molecular Genetics: "The orphan receptor meckelin needs the cytoskeletal protein filamin A to mediate ciliogenesis". [Hum Mol Genet](#).

The group of Marius Ueffing published a curated and structure-based PPI network central to rhodopsin in [Mol Syst Biol](#).

For a complete overview of publications with SYSCILIA acknowledgement you are referred to our website: syscilia.org/results.

2nd Annual Meeting

The next annual meeting of the project will be on **May 14, 15 & 16, 2012**.

Venue: the Institute of Child Health, **London**

Please send your registration to s.dusseljee@antrg.umcn.nl before February 17, 2012.



CILIA 2012 - Cilia in Development and Disease

Subsequent to our SYSCILIA annual meeting, the first international scientific conference organised by the Ciliopathy Alliance, Cilia 2012, will be held at the Institute of Child Health, London on May 16-18.

A two day conference on cilia biology and the ciliopathies - the wide range of life-threatening and sometimes fatal conditions caused by ciliary defects and dysfunction.

On May 16, all participants are invited to an evening reception with the Ciliopathy Alliance patient groups and VIP guests.

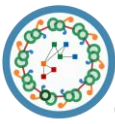
The CILIA 2012 Conference is supported by SYSCILIA.

Deadline for submitting abstracts: 31 January 2012.

Mid term review

The mid term review will be held on September 21, 2012 in Heidelberg.

The aim of this technical review by independent experts is to assess the work carried out under the project and provide recommendations to the European Commission.



Upon request of our Scientific Officer the mid-term review will start on September 20, with discussions on overall coordination, management, training and dissemination activities with the members of the Steering Committee. This will render ample time for the technical review per Workpackage on September 21.

Calendar 2012

Webinars, online training sessions for the SYSCILIA database, will be planned every last Monday of the month. These trainings are available for every participant, see the SYSCILIA private website. Invitations will be send around one week in advance.

Website

Now available on the website for SYSCILIA members: mentoring folders!

Mentoring (formal and informal) has been shown to be particularly effective during the critical final years of post-doctoral training. SYSCILIA will offer the CV's and contact information of all PIs in the form of a "mentoring folder" to SYSCILIA-related PhD-students and post-docs of both genders. The young scientist can contact any PI to enquire about mentoring opportunities. If mentor and protégé agree to proceed, telephone conversations or face-to-face (see preferred method of mentoring in each folder) will be dedicated to improving the post-doc's CV to best suit her/his career aspirations. 2-3 post-doc's can be mentored per PI per year, and the Project Manager will monitor who is mentoring which post-doc.

Young women pursuing a successful science career have few women role models to follow and are more likely to suffer from perceptions of inadequacy. Every year an extra call to enroll will be sent to female post-docs to encourage 100% participation.

Do you have any news item that you would like to see announced in this Newsletter? Input can be send to: simone.dusseljee@gmail.com

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