SYSCILIA Newsletter 3 – November 2011

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**

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. http://cilia2012.org/

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.

Midterm review

The midterm 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. All SYSCILIA principal investigators will participate.

Annual report

The Annual Report was submitted to the European Commission by **July 28, 2011**. On September 30, we received the first comments of our Scientific Officer, concerning the explanation of resources per partner. The partners involved have given the requested specification of costs. On October 14, 2011 we have re-submitted a revised version of the report to the European Commission and we currently await further analysis of the scientific report and the Deliverable reports. The deadline for the EU is November 26, 2011.
The publishable summary can be found on our website. The entire report can be accessed by SYSCILIA partners only at the private section (privatewiki) of our website.

Semi-annual reporting

The Deliverables due at month 18 have to be submitted to the European Commission at latest on December 1st, 2011. Templates have been sent to the partners involved and are expected to be sent to the Project Manager by November 28, 2011.

Templates will be sent around on December 1st to start preparations of the semi-annual report, month 18. The deadline for finalizing this report is January 27, 2012.

Publications and Press releases

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

KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes
KIF7, the human ortholog of Drosophila Costal2, is a key component of the Hedgehog signaling pathway. Mutations in KIF7 are reported in individuals with hydrolethalus and acrocallosal syndromes, two multiple malformation disorders with overlapping features that include polydactyly, brain abnormalities and cleft palate. The publication shows the role of KIF7 in human primary cilia, especially in the Hedgehog pathway through the regulation of GLI targets, and expand the clinical spectrum of ciliopathies.
Nat Genet. 2011 Jun;43(6):601-6

Nephrocystin-4 is required for normal cloaca formation during zebrafish embryogenesis
NPHP4 mutations cause nephronophthisis, an autosomal recessive cystic kidney disease associated with renal fibrosis and kidney failure. The NPHP4 gene product nephrocystin-4 interacts with other nephrocystins, cytoskeletal and ciliary proteins; however, the molecular and cellular functions of nephrocystin-4 have remained elusive. Here we demonstrate that nephrocystin-4 is required for normal cloaca formation during zebrafish embryogenesis.
Hum Mol Genet. 2011 Aug 15;20(16):3119-28

Transport defects in the cilium cause renal dysfunction and abnormal skeleton development
Defects in the *WDR19* gene result in ciliary transport defects in rare genetic disorders (Sensenbrenner syndrome, Jeune syndrome, and isolated nephronophthisis), that are characterized by skeletal and/or chronic renal disease. *WDR19* encodes IFT144, a protein that is part of the IFT-A particle. Empowered by the cytoplasmic dynein motor, this intraflagellar transport module facilitates tip-to-basemolecular transport in the cilium. WDR19 is the third 'IFT' gene in line that could be associated with Sensenbrenner syndrome.
Am J Hum Genet. 2011 Oct 19.

Calendar

Webinars, online training sessions for the SYSCILIA database, will be planned regularly. These trainings are available for every participant, see the SYSCILIA private website.

Component Meetings and a Steering Committee Meeting have been held; minutes can be found on the website after login.

November 17: seminar by Erica Davis (Katsanis lab) "Genetic and functional dissection of total mutational load in ciliopathies". Utrecht, The Netherlands.

5-6 December: The first Innovation Convention organized by the European Commission. Square- Brussels Meeting Centre.
http://ec.europa.eu/research/innovation-union/ic2011/index_en.cfm

11-12 January 2012: **SYSCILIA training & meeting, Heidelberg.**

Venue: Greenier Room, 7th floor, BioQuant building, University of Heidelberg, Im Neuenheimer Feld 267, 69120 Heidelberg.

Website

Now available for SYSCILIA members on the private section of our website, the pdf-files of listed publications.

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