

SYSCILIA Newsletter 5 – April 2012

2nd Annual Meeting

On May 14 – 16, SYSCILIA will be hosting 48 consortium members and 4 Scientific Advisory Board members for the second annual meeting of the project.

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



CILIA 2012 - Cilia in Development and Disease

1st International Conference in Europe for the Cilia Scientific and Clinical Community Initiated by the Ciliopathy Alliance, supported by SYSCILIA. [View conference website](#)



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

Publications and Press releases

Partner 11, University College Dublin, Oliver Blacque published in [Current Biology](#) “**Endocytosis Genes Facilitate Protein and Membrane Transport in *C. elegans* Sensory Cilia**”.

Multiple intracellular transport pathways drive the formation, maintenance, and function of cilia, a compartmentalized organelle associated with motility, chemo-/mechano-/photosensation, and developmental signaling. These pathways include cilium-based intraflagellar transport (IFT) and poorly understood membrane trafficking events. Defects in ciliary transport contribute to the etiology of human ciliary disease such as Bardet-Biedl syndrome (BBS). In this study, we employ the genetically tractable nematode *Caenorhabditis elegans* to investigate whether endocytosis genes function in cilium formation and/or the transport of ciliary membrane or ciliary proteins.



Partner 8, UMCU, Rachel Giles' paper about the etiology of pancreatic cysts has just been accepted by *Neurology*.

"**Inversin, Wnt signaling and primary cilia**" is the title of the paper published by partner 5, UKL FR, Gerd Walz in the February edition of [Differentiation](#).

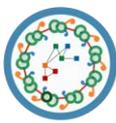
Erica Davis and Nico Katsanis from Duke, partner 4, and Colin Johnson, LIMM, partner 10, are co-authors on the paper published in [Science](#) "**Evolutionarily assembled cis-regulatory module at a human ciliopathy locus**"

Neighboring genes are often coordinately expressed within cis-regulatory modules, but evidence that nonparalogous genes share functions in mammals is lacking. Here, we report that mutation of either TMEM138 or TMEM216 causes a phenotypically indistinguishable human ciliopathy, Joubert syndrome. Despite a lack of sequence homology, the genes are aligned in a head-to-tail configuration and joined by chromosomal rearrangement at the amphibian-to-reptile evolutionary transition. Expression of the two genes is mediated by a conserved regulatory element in the noncoding intergenic region. Coordinated expression is important for their interdependent cellular role in vesicular transport to primary cilia. Hence, during vertebrate evolution of genes involved in ciliogenesis, nonparalogous genes were arranged to a functional gene cluster with shared regulatory elements.

For a complete overview of publications with SYSCILIA acknowledgement you are referred to our website: 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**, between partners about data exchange and analysis
- **International Conference on the [Systems Biology of Human Disease](#)**
2 – 4 May 2012; Heidelberg, Germany
- **[preARVO 2012 satellite meeting](#); Retina Ciliopathies: From Genes to Mechanisms and Treatment**
4 – 5 May 2012: Fort Lauderdale Florida, USA
- **[ARVO 2012](#); Translational research: Seeing the possibilities**
6 – 10 May 2012; Greater Fort Lauderdale/Broward County Convention Center, USA
- **31 May 2012 – end of 2nd reporting period 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
- [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.**
16 – 18 January 2013; Heidelberg, Germany

SYSCILIA participants

Gisela Slaats (UMCU)



Konstantinos Koutroumpas (CNRS)



Who are you?

My full name is Gisela Gijsbertje Gerretje Slaats and I was born in 1985 in Geldrop (NL). I moved to the Wageningen University and Research Center (NL) for my BSc and MSc Molecular Life Sciences with a biomedical specialization. I started with a short thesis at the Nutrition and Pharmacology department in Wageningen. Then I did an internship at the Erasmus Medical Center Rotterdam at the department of Genetics. Finally, I stayed in Stockholm for 8 months for my major thesis at the Karolinska Institutet at the Department of Medicine. In January 2011 I started as PhD student with Rachel Giles at the department of Nephrology, University Medical Center Utrecht (NL).

Konstantinos Koutroumpas; Post-Doc researcher at the *institute of Systems and Synthetic Biology (iSSB), Evry, France*

Education:

Apr. 2005 Diplomate Electrical and Computer Engineer (Five Year Diploma) University of Patras, Department of Electrical and Computer Engineering

May 2010 Doctor of Sciences “Stochastic Hybrid Systems in DNA replication modelling.”

ETH Zurich, Department of Information Technology and Electrical Engineering

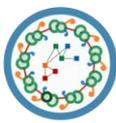
Work Experience:

2005-2006 Research Assistant; University of Patras, Department of Electrical and Computer Engineering.

Participated in the implementation of an innovative concept for lunar exploration on behalf of ESA’s project MANET Intercommunication for Robotic Applications for Lunar Exploration (MIRACLE) funded by Alcatel Alenia Space Italia SpA.

2005-2010 Research Assistant ETH Zurich, Department of Information Technology and Electrical Engineering.

Researcher in the project Hybrid systems for biochemical network Modeling and analysis (HYGEIA)



funded by European Commission (FP6–NEST–4995).
2011 - Post Doc Researcher *institute of Systems and Synthetic Biology (iSSB), Evry, France*
Researcher in the project SYSCILIA, "A systems biology approach to dissect cilia function and its disruption in human genetic disease", funded by the European Community's [Seventh Framework Programme \(FP7/2007-2013\)](#)

What have you learned about yourself over the past one and a half year?

I learned that I should consider which tasks are important and urgent in planning my work. Time management is an important factor during your PhD, since there are so many things you can do!

Participating in a highly interdisciplinary project like SYSCILIA, the willing to learn about different topics is a prerequisite. This necessity helped me to overcome the fear of plunging into new unfamiliar areas and revealed the excitement of discovering new field.

What dream do you want to come true in the coming year?

I hope my first article in the cilia field will be published!

I would like my country, Greece, to overcome the economical crisis and to give the opportunity to scientists working in and out of the country to contribute to its development.

What do you want to achieve next year with the project?

Together with Ludo Westerveld (student) I hope we will find some nice targets in the kinesome screen which can be validated. Overall, I hope all SYSCILIA partners will have interesting results during their screens and modeling, because this is important input for follow-up experiments.

The main aim is to understand the molecular mechanisms of diseases by uncovering how gene mutations disrupt molecular networks giving rise to disease phenotypes. My hope is that this analysis will provide us with a genotype to phenotype map that we will be able to exploit by predict new disease candidates.

What is your life motto?

If you are going to do something, do it right! (In Dutch: als je iets doet, doe het dan goed)

"Imagination is more important than knowledge." - Albert Einstein

What trait would you like to change about yourself and what do you find a nice personal trait?

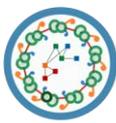
I wish I could be a bit more patient. I don't like waiting, especially when I have something planned for example. What I do like is my organized way of working. I rarely forget or loose things.

What I consider my strong character skill is my ability to become familiar with new scientific areas, an ability developed through my participation in diverse and multidisciplinary projects. On the other hand, I always get passionate with what I do and this results in dedicating all my time on the work putting aside my personal life.

What helps you to relax in times of rush and stress?

Since I was little I took dance classes, and nowadays I still do. Zumba and comparable dance lessons make me feel relaxed! Besides that, I also like to bake a nice cake or pie to relax.

A long bicycle ride always helps me to **relax and** to run away from the rush hours of the day. Moreover, it is a great way to refresh my mind and come back with new ideas.



If you could choose another profession what would you choose and why?

I think this is a very difficult question! You can think of many positive and negative things about each profession. I think I would choose to be a pilot....fly across the world, and reserve some extra time to visit places and see a lot of the world! There are still so many places on my to-visit list!

I would rather choose an education related job. I would like to be a teacher, mainly because of the satisfaction you feel when you see that you can transfer your knowledge to other people.

Who is your idol and why?

I don't have one at the moment. But during my puberty my idols were Phillip Cocu (soccer player PSV) and Pieter van den Hoogenband (swimmer, also born in Geldrop). And yes, posters were above my bed (I guess this is the most important indicator?)

Hiroaki Kitano, for his significant contributions to engineering and Systems Biology.

Where do you become inspired by?

Classical answer...Mother Nature! And good music!

My PhD advisor, Prof. J. Lygeros, who showed me that research is not only a nice occupation, even when some of our goals are not achieved, but also a way of life.

Anything to add?

I'm looking forward to see you all in London during the 2nd annual meeting!

News: University of Heidelberg hired a postdoc, Qianhao Lu, who will start May 1st. We will be able to welcome him to the project during our annual meeting in London.

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

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