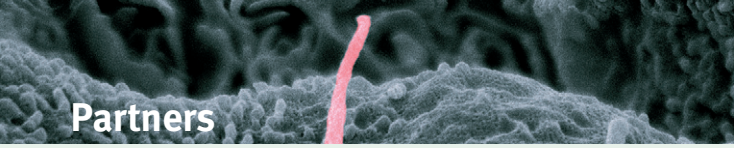

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Syscilia

Syscilia

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http://syscilia.org/

SYSCILIA

A systems biology approach to dissect cilia function and its disruption in human genetic disease.

The aim of SYSCILIA is to identify the molecular mechanisms characterizing cilium function, and the discrete perturbations associated with dysfunction caused by mutations in inherited ciliopathies, applying a systems biology approach.

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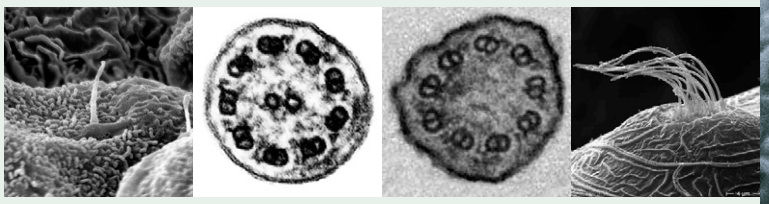

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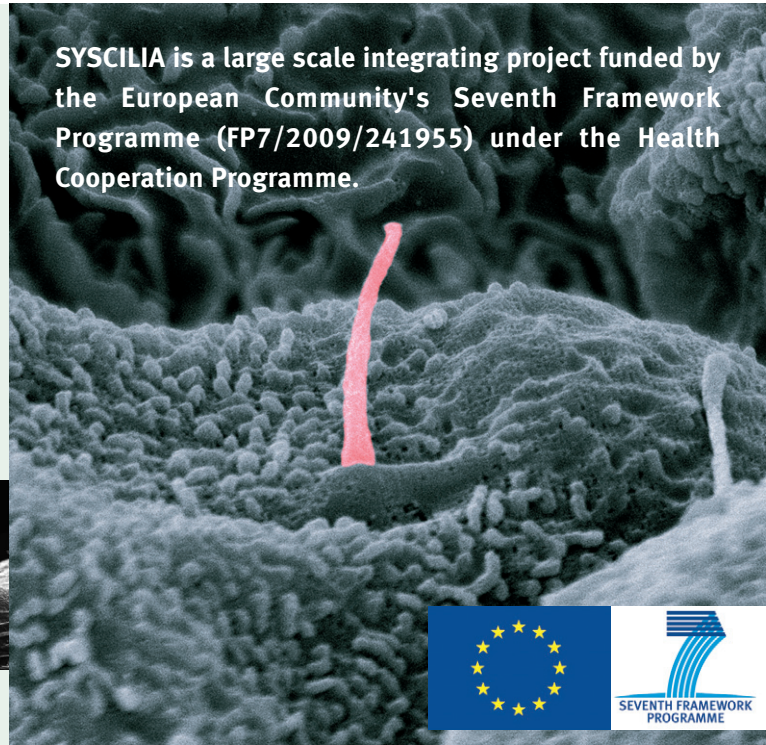
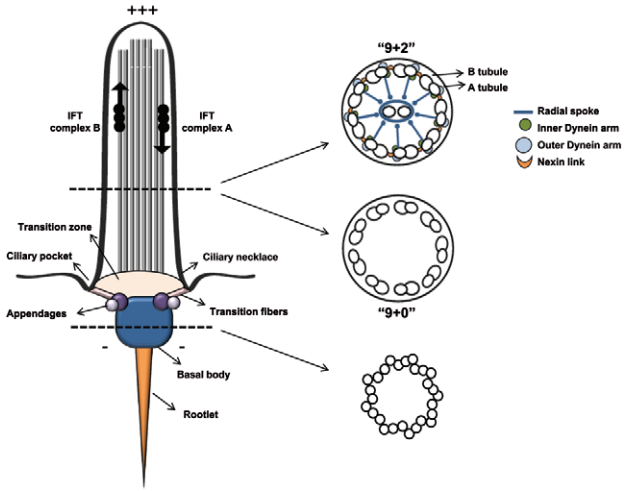


SEVENTH FRAMEWORK PROGRAMME

Ciliopathies - *hereditary diseases caused by defects in the function of cilia*

The cilium

A cilium is a longitudinal projection from the surface of almost every vertebrate cell. Cilia are derived from the mature centriole of the centrosome, and perform essential cellular functions. Some cilia are motile, which are used by the cell to propel fluid. Most cilia however are non-motile, also named primary cilia. These were long thought to be vestigial organelles that had lost their crucial motile function throughout evolution. In the last decade however, they were found to harbour key receptors and other components of cellular signalling pathways that are vitally important in the regulation of many biological processes. They turned out to be essential signalling hubs, acting like the cell's antenna.



Ciliopathies

Consistent with their broad tissue and cellular distribution, it is now recognized that defects in cilia give rise to an equally broad but consistent range of phenotypes in mammals. They underlie a number of often chronically disabling and sometimes life-threatening genetic conditions, such as **blindness, deafness, chronic respiratory infections, kidney disease, heart disease, infertility, obesity, diabetes, and even some types of cancer**. What links these diverse individual disorders is that the proteins, known to cause the disease upon mutation of their cognate genes, are localized to the cilia or their anchoring structure, the basal body and the adjacent centriole and play some role in their function. Therefore, these hereditary diseases are called ciliopathies

Scientific & Technical Objectives

Our overall objectives are to **establish a paradigm for studying and modelling complex eukaryotic systems**, to **understand how system perturbation contributes to the modulation of clinical phenotypes**, and to provide a **better understanding of ciliary processes in biology and their associated diseases**.
Our objectives focus on all critical components of the systems biology process, namely:

- assay development and application,
- data generation, handling and integration,
- model building and testing followed by refinement.

We also exploit the models to find new insights into biological mechanism and human disease.