

ENCONTRO SCIENTIA

November 28

12h00

Room 2.3.13, Ciências ULisboa

Restoring Motile cilia to make a good beat

Primary Ciliary Dyskinesia (PCD) is an untreatable rare disease affecting 1:7500 people, which can be caused by at least 50 different defective genes. PCD is an autosomal recessive disease that affects organs with motile cilia. The respiratory system, for example, is progressively compromised due to an impaired mucociliary transport. However, PCD is more than a respiratory disease, as it can lead to infertility and half of the PCD patients present *situs inversus* or heterotaxy, a condition highly correlated with congenital heart disease.

We generated a zebrafish model for a severe PCD variant and we tested mRNA therapy for cilia motility restoration provided by Ethris GmbH. We have shown cilia motility can be rescued using lipid nanoparticles formulated with a mRNA-based technology.

Zebrafish is a small vertebrate comparable to a living test tube, it shares 70% of genes with humans and our genomes are so conserved that human mRNAs rescue zebrafish mutations.



**Susana
S. Lopes**

**Cilia
in Development
and Disease,
CE3C,
Ciências ULisboa**

