Comunicados de Imprensa
Faculdade de Ciências
UNIVERSIDADE DE LISBOA

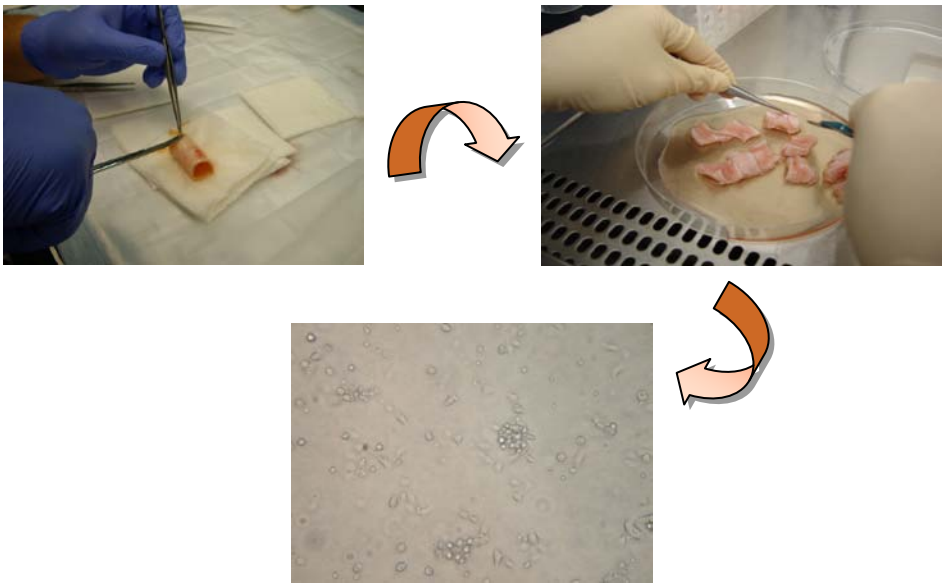
“Personalized Medicine”

Researchers test new investigational drug for patients with Cystic Fibrosis

There is already a **therapeutic approach** for Cystic Fibrosis patients with the **second most frequent mutation in Portugal** (A561E). Seven researchers from the **Faculty of Sciences of the University of Lisboa**, in collaboration with two clinicians from **Hospital La Fe in Valencia**, Spain, pre-clinically tested this new investigational drug, which is expected to be approved soon by the US (FDA) and EU (EMA) regulatory agencies.

"Our results show that, similarly to what happens to patients with the most frequent CF-causing mutation (F508del), patients with the A561E mutation may also benefit from this new drug", says **Margarida Amaral, principal investigator of this study**, professor at the Department of Chemistry and Biochemistry, Faculty of Sciences , University of Lisboa and **coordinator of the new research centre from FCT (Portuguese Foundation for Science and Technology) - BioISI-Biosystems & Integrative Sciences Institute**.

Within the scope of a protocol established in 2011, clinicians from **Hospital La Fe in Valencia** send to Portugal **lungs explanted from patients with Cystic Fibrosis**, who undergo lung transplantation in that hospital. **BioISI researchers grow the cells from these lungs**, and use them in various types of studies. The goal is to understand the molecular mechanisms underlying this genetic disease.



The **A561E mutation** is also relatively frequent in **Cystic Fibrosis patients** from **Spain** and **Brazil**.

"In the particular case of the present study, we have assessed the response of lung cells from patients with different (rare) mutations in the CFTR gene (which is gene that is mutated in this disease) to a new investigational drug for cystic fibrosis. If we find a positive response (as it was indeed the case here) we have **a scientific basis justifying the widening the scope of this new drug** to more patients. This is **a 'personalized medicine' approach**", explains Margarida Amaral.

The results of this study are published in **EBioMedicine**. The article "**Measurements of Functional Responses in Human Primary Lung Cells as a Basis for Personalised Therapy for Cystic Fibrosis**" is authored by **Nikhil Awatade, Inna Uliyakina, Carlos Farinha, Luka Clarke, Karina Mendes, Maria Margarida Ramos and Margarida Amaral**, researchers of BioISI (Lisboa, Portugal) and also **Amparo Solé and Juan Pastor**, from Hospital La Fe (Valencia, Spain).

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